Contents

1 Mapping of disease names onto ICD-9 codes

2 Disease list and descriptions: A
   2.1 Amino acid (AA) metabolism (aromatic) (Albinism, Alkaptonuria, Tyrosinemia, Waardenburg syndrome) ......................................................... 15
   2.1.1 Albinism ................................................................. 15
   2.1.2 Alkaptonuria ............................................................ 15
   2.1.3 Tyrosinemia ............................................................. 15
   2.1.4 Waardenburg syndrome ............................................ 15
   2.2 Amino acid (AA) metabolism (branched) (maple syrup urine disease) ................................................................. 16
   2.2.1 Maple syrup urine disease ........................................ 16
   2.3 Amino acid (AA) metabolism (Lowe) (Beta-alaninemia, Hydroxyprolinemia, Hyperprolinemia, Sarcosinemia) .................................................. 16
   2.3.1 Beta-alaninemia ...................................................... 16
   2.3.2 Hydroxyprolinemia ................................................... 16
   2.3.3 Hyperprolinemia ...................................................... 16
   2.3.4 Sarcosinemia ......................................................... 17
   2.4 Amino acid (AA) metabolism (sulfur-bearing) (Homocystinuria) ................................................................. 17
   2.4.1 Homocystinuria ....................................................... 17
   2.5 Amino acid (AA) metabolism (straight-chain) (Hyperglycinemia, Hyperlysinemia, Pipecolic acidemia, Saccharopinuria) ........................................ 17
   2.5.1 Hyperglycinemia ...................................................... 17
   2.5.2 Hyperlysinemia ...................................................... 17
   2.5.3 Pipecolic acidemia .................................................. 17
   2.5.4 Saccharopinuria ....................................................... 18
   2.6 Amino acid (AA) transport (Cystinosis, Cystinuria, Hartnup disease) ................................................................. 18
   2.6.1 Cystinosis ............................................................... 18
   2.6.2 Cystinuria ............................................................... 18
   2.6.3 Hartnup disease ....................................................... 18
   2.7 Acanthosis nigricans .................................................... 18
   2.8 Acidosis ................................................................. 19
   2.9 (Organic) Aciduria ........................................................ 19
   2.10 Actinomycosis ........................................................... 19
   2.11 Acute leukemia .......................................................... 20
   2.12 Acute promyelocytic leukemia ........................................ 20
   2.13 Ainhum ................................................................. 20
   2.14 Albright (-McCune)-Sternberg syndrome ................................. 21
   2.15 Alcoholism ............................................................... 21
   2.16 Allergic rhinitis ........................................................ 21
   2.17 Alkalosis ................................................................. 21
   2.18 Alopecia (baldness) .................................................... 22
2.19 Alopecia areata .................................................. 22
2.20 Alzheimer’s disease ............................................ 22
2.21 Amebiasis ........................................................ 22
2.22 Amyotrophic lateral sclerosis ............................... 23
2.23 Anaerobic infection ............................................. 23
2.24 Aniridia (eye abnormalities) ................................. 23
2.25 Ankylosing spondylitis ....................................... 23
2.26 Anthrax .......................................................... 24
2.27 Aortic aneurysm ............................................... 24
2.28 Aplastic anemia ................................................. 24
2.29 Attention Deficit Hyperactivity Disorder .................. 24
2.30 Autism .......................................................... 25
3 Disease list and descriptions: B ................................. 25
  3.1 Behcet’s syndrome ........................................... 25
  3.2 Benign neoplasms ............................................ 26
  3.3 Bipolar disorder .............................................. 26
  3.4 Breast cancer (male) .......................................... 26
  3.5 Breast cancer .................................................. 26
  3.6 Brucellosis .................................................... 27
  3.7 Budd-Chiari syndrome ...................................... 27
  3.8 Bundle branch block ........................................ 27
  3.9 Buphthalmos .................................................. 27
  3.10 Burkitt lymphoma ........................................... 28
4 Disease list and descriptions: C .................................. 28
  4.1 Carbohydrate transport and metabolism (Glycogen storage disease, Lactose intolerance, Galactosemia) .................................................. 28
  4.1.1 Glycogen storage disease ................................. 28
  4.1.2 Lactose intolerance ...................................... 28
  4.1.3 Galactosemia ............................................... 29
  4.2 Carcinoma in situ ............................................. 29
  4.3 Cardiomyopathy .............................................. 29
  4.4 Celiac sprue (celiac disease) ............................... 29
  4.5 Cerebral palsy ............................................... 30
  4.6 Cervical rib .................................................. 30
  4.7 Charcot-Marie-Tooth disease .............................. 30
  4.8 Cholelithiasis (gallstones) ................................. 30
  4.9 Cholera ...................................................... 31
  4.10 Chondrodystrophy (achondroplasia) ...................... 31
  4.11 Congenital absence of vertebrae ......................... 31
  4.12 Congenital spinal fusion (congenital spinal stenosis) ... 32
  4.13 Cystic fibrosis .............................................. 32
5 Disease list and descriptions: D ................................. 33
  5.1 Dejerine-Sottas syndrome ................................ 33
  5.2 Depression .................................................. 33
  5.3 Dermatomyositis-polymyositis ............................ 34
  5.4 Diabetes mellitus .......................................... 34
  5.5 Diphtheria .................................................. 34
6 Disease list and descriptions: E

6.1 *E. coli* intestinal disease ................................................................. 35
6.2 Edward’s syndrome ................................................................. 35
6.3 Enzyme-deficiency (hemolytic anemia) .................................................. 35
6.4 Epilepsy (seizure disorder) .......................................................... 36
6.5 Erythematous squamous dermatosis (seborrheic dermatitis) ................. 36

7 Disease list and descriptions: F

7.1 Food poisoning ................................................................. 36
7.2 Fragile X syndrome .......................................................... 36
7.3 Friedreich’s ataxia .............................................................. 37

8 Disease list and descriptions: G

8.1 Giant cell arteritis .............................................................. 37
8.2 Gram-negative folliculitis ...................................................... 37
8.3 Goiter ................................................................. 38
8.4 Good pastures syndrome ..................................................... 38
8.5 Gout ................................................................. 38

9 Disease list and descriptions: H

9.1 Hemivertebra ................................................................. 38
9.2 *Helicobacter pylori* infection .................................................. 39
9.3 Hepatitis A ................................................................. 39
9.4 Hepatitis B ................................................................. 39
9.5 Hepatitis C ................................................................. 39
9.6 Hepatitis D ................................................................. 40
9.7 Hepatitis E ................................................................. 40
9.8 Hereditary spastic paraplegia .................................................. 40
9.9 HIV disease ................................................................. 40
9.10 Hodgkin’s disease ........................................................ 41
9.11 Hyperosmolality (hypernatremia) ............................................... 41
9.12 Hypersensitivity angiitis .......................................................... 41
9.13 Hypoglycemia ............................................................. 41
9.14 Hypoosmolality (hyponatremia) ............................................... 42
9.15 Hypertrophic obstructive cardiomyopathy ...................................... 42

10 Disease list and descriptions: I

10.1 Ichthyosis congenita ........................................................ 42

11 Disease list and descriptions: K

11.1 Kawasaki disease .......................................................... 43
11.2 Klippel-Feil syndrome .......................................................... 43

12 Disease list and descriptions: L

12.1 Leprosy (Hansen disease) ...................................................... 43
12.2 Lethal midline granuloma ....................................................... 44
12.3 Leukodystrophy ................................................................. 44
12.4 Lipid metabolism (Hypercholesterolemia, Hypertriglyceridemia, Hyperlipoproteinemia, Combined hyperlipidemia, Abetalipoproteinemia, Lipodystrophy, Gaucher’s disease, Niemann-Pick disease, Carnitine-acylcarnitine translocase deficiency, Mitochondrial trifunctional protein deficiency) ..................................................... 44
12.4.1 Hypercholesterolemia ..................................................... 44
12.4.2 Hypertriglyceridemia ..................................................... 45
12.4.3 Hyperlipoproteinemia ..................................................... 45
12.4.4 Combined hyperlipidemia ..................................................... 45
12.4.5 Abetalipoproteinemia .................................................. 45
12.4.6 Lipodystrophy ........................................................... 45
12.4.7 Gaucher’s disease ....................................................... 46
12.4.8 Niemann-Pick disease ................................................ 46
12.4.9 Carnitine-acylcarnitine translocase deficiency ................. 46
12.4.10 Mitochondrial trifunctional protein deficiency ............... 47
12.5 Lown-Ganong-Levine syndrome ...................................... 47
12.6 Lumbosacral spondylysis ................................................. 47
12.7 Lymphosarcoma (lymphoma) ............................................ 47

13 Disease list and descriptions: M ....................................... 48
13.1 Migraine ........................................................................ 48
13.2 Meningococcal infection .................................................. 48
13.3 Mineral metabolism (Fe) (Haemochromatosis) ..................... 48
13.4 Mineral metabolism (Cu) (Wilson’s disease, Menkes’ disease) ... 48
13.4.1 Wilson’s disease ......................................................... 48
13.4.2 Menkes’ disease ......................................................... 49
13.5 Mineral metabolism (Mg) (Hypermagnesemia, Hypomagnesemia) . 49
13.5.1 Hypermagnesemia ...................................................... 49
13.5.2 Hypomagnesemia ....................................................... 49
13.6 Mineral metabolism (Ca) (Hypocalcaemia, Hypercalcaemia, Pseudohypoparathyroidism) . 50
13.6.1 Hypocalcaemia ......................................................... 50
13.6.2 Hypercalcaemia ......................................................... 50
13.6.3 Pseudohypoparathyroidism ....................................... 50
13.7 Mitochondrial disease .................................................... 50
13.8 Moyamoya ................................................................. 51
13.9 Multiple epiphyseal dysplasia ......................................... 51
13.10 Multiple sclerosis ......................................................... 51
13.11 Mumps ................................................................. 51
13.12 Muscular dystrophy ..................................................... 52
13.13 Mycoplasma ............................................................... 52
13.14 Myotonic disorders (myotonia) ..................................... 52

14 Disease list and descriptions: N ....................................... 52
14.1 Neurofibromatosis .......................................................... 52
14.2 Neuromyelitis optica (NMO, Devic’s disease) ..................... 53

15 Disease list and descriptions: O ....................................... 53
15.1 Ornithosis (psittacosis) ................................................... 53
15.2 Osteogenesis imperfecta ................................................ 53
15.3 Osteopetrosis ............................................................. 53

16 Disease list and descriptions: P ....................................... 54
16.1 Parkinson’s disease ....................................................... 54
16.2 Patau’s syndrome ......................................................... 54
16.3 Pertussis (whooping cough) ........................................... 54
16.4 Pervasive development disorders .................................... 55
16.5 Phenylketonuria .......................................................... 55
16.6 Pick’s disease ............................................................. 55
16.7 Plasma cell leukemia .................................................... 55
16.8 Poliomyelitis ............................................................... 56
16.9 Plague ................................................................. 56
16.10 Pneumonia ............................................................... 56
16.11 Polyarteritis nodosa ..................................................... 56
16.12 Polyostotic fibrous dysplasia ....................................... 57
<table>
<thead>
<tr>
<th>Section</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>16.13</td>
<td>57</td>
</tr>
<tr>
<td>16.14</td>
<td>57</td>
</tr>
<tr>
<td>16.15</td>
<td>57</td>
</tr>
<tr>
<td>16.16</td>
<td>58</td>
</tr>
<tr>
<td>16.17</td>
<td>58</td>
</tr>
<tr>
<td>17.1</td>
<td>58</td>
</tr>
<tr>
<td>17.2</td>
<td>59</td>
</tr>
<tr>
<td>17.3</td>
<td>59</td>
</tr>
<tr>
<td>17.4</td>
<td>59</td>
</tr>
<tr>
<td>18.1</td>
<td>60</td>
</tr>
<tr>
<td>18.2</td>
<td>60</td>
</tr>
<tr>
<td>18.3</td>
<td>60</td>
</tr>
<tr>
<td>18.4</td>
<td>60</td>
</tr>
<tr>
<td>18.5</td>
<td>60</td>
</tr>
<tr>
<td>18.6</td>
<td>60</td>
</tr>
<tr>
<td>18.7</td>
<td>60</td>
</tr>
<tr>
<td>18.8</td>
<td>60</td>
</tr>
<tr>
<td>18.9</td>
<td>60</td>
</tr>
<tr>
<td>18.10</td>
<td>61</td>
</tr>
<tr>
<td>18.11</td>
<td>61</td>
</tr>
<tr>
<td>19.1</td>
<td>62</td>
</tr>
<tr>
<td>19.2</td>
<td>62</td>
</tr>
<tr>
<td>19.3</td>
<td>63</td>
</tr>
<tr>
<td>19.4</td>
<td>63</td>
</tr>
<tr>
<td>19.5</td>
<td>63</td>
</tr>
<tr>
<td>19.6</td>
<td>63</td>
</tr>
<tr>
<td>20.1</td>
<td>64</td>
</tr>
<tr>
<td>21.1</td>
<td>64</td>
</tr>
<tr>
<td>21.2</td>
<td>64</td>
</tr>
<tr>
<td>21.2.1</td>
<td>64</td>
</tr>
<tr>
<td>21.2.2</td>
<td>64</td>
</tr>
<tr>
<td>21.2.3</td>
<td>65</td>
</tr>
<tr>
<td>21.2.4</td>
<td>65</td>
</tr>
<tr>
<td>21.3</td>
<td>65</td>
</tr>
<tr>
<td>22.1</td>
<td>65</td>
</tr>
<tr>
<td>22.2</td>
<td>66</td>
</tr>
</tbody>
</table>
List of Tables

1  Disorders, record counts, and the raw and adjusted prevalence. . . . . . . . . . . . . . . . 67
2  Disorders, record counts, and the raw and adjusted prevalence. . . . . . . . . . . . . . . . 68
3  Disorders, record counts, and the raw and adjusted prevalence. . . . . . . . . . . . . . . . 69
4  Disorders, record counts, and the raw and adjusted prevalence. . . . . . . . . . . . . . . . 70
1 Mapping of disease names onto ICD-9 codes

International classification of diseases, ninth revision (ICD-9) codes [?] are used to record medical findings (primarily for billing purposes) in hospital data bases, including the one that we used for our analysis. Because this system was designed for financial purposes (patient billing) rather than for research, and multiple ICD-9 codes typically map into a single disease (and often multiple rare disorders map to a single ICD-9 code), additional work is required to match data base ICD-9 codes into disease names. The correspondence between the disease names and ICD-9 codes that we assumed in this study is as follows.

Amino acid (AA) metabolism (aromatic) disorders
—270.2
Amino acid (AA) metabolism (branched) disorders
—270.3
Amino acid (AA) metabolism (Lowe) disorders
—270.8
Amino acid (AA) metabolism (sulfur-bearing) disorders
—270.4
Amino acid (AA) metabolism (straight-chain) disorders
—270.7
Amino acid (AA) transport disorders
—270, 277.0
Acanthosis nigricans
—701.2
Acidosis
—276.2
Aciduria
—270.9
Actinomycosis
—039.0, 039.1, 039.2, 039.3, 039.4, 039.8, 039.9
Acute leukemia
—208.0
Acute promyelocytic leukemia
—205.0
Ainhum
—136.0
Albright-Sternberg syndrome
—756.59
Alcoholism
—291.0, 291.1, 291.2, 291.3, 291.4, 291.5, 291.81, 291.89, 291.9, 303.00, 303.01, 303.02, 303.03, 303.90, 303.91, 303.92, 303.93, 305.00, 305.01, 305.02, 305.03, 357.5, 425.5, 571.0, 571.1, 571.2, 571.3, 790.3
Allergic rhinitis
—477.1, 477.2, 477.8, 477.9
Alkalosis
—276.3
Alopecia
—704.00, 704.02, 704.09
Alopecia areata
—704.01
Alzheimer’s disease
—290.0, 290.10, 331.0
Amebiasis
—006.0, 006.1, 006.2, 006.3, 006.4, 006.5, 006.6, 006.8, 006.9
Amyotrophic lateral sclerosis
—335.20
Anaerobes — 041.82, 041.83, 041.84, 038.3
Aniridia — 743.45
Ankylosing spondylitis — 720.0
Anthrax — 022.0, 022.1, 022.2, 022.3, 022.8, 022.9
Aortic aneurysm — 441.1, 441.2, 441.3, 441.4, 441.5, 441.6, 441.7, 441.9
Aplastic anemia — 284.0, 284.8, 284.9
Attention deficit — 314.0, 314.00, 314.01
Autism — 299.00, 299.01
Behcet’s syndrome — 136.1, 711.20, 711.21, 711.22, 711.23, 711.24, 711.25, 711.26, 711.27, 711.28, 711.29
Benign neoplasms — 210.0, 210.1, 210.2, 210.3, 210.4, 210.5, 210.6, 210.7, 210.8, 210.9, 211.0, 211.1, 211.2, 211.3, 211.4, 211.5, 211.6, 211.7, 211.8, 211.9, 212.0, 212.1, 212.2, 212.3, 212.4, 212.5, 212.6, 212.7, 212.8, 212.9, 213.0, 213.1, 213.2, 213.3, 213.4, 213.5, 213.6, 213.7, 213.8, 213.9, 214.0, 214.1, 214.2, 214.3, 214.4, 214.8, 214.9, 215.0, 215.2, 215.3, 215.4, 215.5, 215.6, 215.7, 215.8, 215.9, 216.0, 216.1, 216.2, 216.3, 216.4, 216.5, 216.6, 216.7, 216.8, 216.9, 217, 218.0, 218.1, 218.2, 218.9, 219.0, 219.1, 219.8, 219.9, 220, 221.0, 221.1, 221.2, 221.3, 221.8, 221.9, 222.0, 222.1, 222.2, 222.3, 222.4, 222.8, 222.9, 223.0, 223.1, 223.2, 223.3, 223.81, 223.89, 223.9, 224.0, 224.1, 224.2, 224.3, 224.4, 224.5, 224.6, 224.7, 224.8, 224.9, 225.0, 225.1, 225.2, 225.3, 225.4, 225.8, 225.9, 226, 227.0, 227.1, 227.3, 227.4, 227.5, 227.6, 227.8, 227.9, 228.00, 228.01, 228.02, 228.03, 228.04, 228.09, 228.1, 229.0, 229.8, 229.9
Bipolar disorder — 296.00, 296.01, 296.02, 296.03, 296.04, 296.05, 296.06, 296.10, 296.11, 296.12, 296.13, 296.14, 296.15, 296.16, 296.40, 296.41, 296.42, 296.43, 296.44, 296.45, 296.46, 296.50, 296.51, 296.52, 296.53, 296.54, 296.55, 296.56, 296.60, 296.61, 296.62, 296.63, 296.64, 296.65, 296.66, 296.67, 296.80, 296.81, 296.82, 296.89, 296.90, 296.99
Breast cancer, male — 175.0, 175.9
Brucellosis — 023.0, 023.1, 023.2, 023.3, 023.8, 023.9
Budd-Chiari — 453.0
Bundle branch block — 426.0, 426.10, 426.11, 426.12, 426.13, 426.2, 426.3, 426.4, 426.50, 426.51, 426.52, 426.53, 426.54, 426.6
Buphthalmos — 743.20, 743.21, 743.22
Carcinoma in situ — 230.0, 230.1, 230.2, 230.3, 230.4, 230.5, 230.6, 230.7, 230.8, 230.9, 231.0, 231.1, 231.2, 231.8, 231.9, 232.0, 232.1, 232.2, 232.3, 232.4, 232.5, 232.6, 232.7, 232.8, 232.9, 233.0, 233.1, 233.2, 233.3, 233.4, 233.5, 233.6, 233.7, 233.9, 234.0, 234.8, 234.9, 235.0, 235.1, 235.2, 235.3, 235.4, 235.5, 235.6, 235.7, 235.8, 235.9, 236.0, 236.1, 236.2, 236.3, 236.4, 236.5, 236.6, 236.7, 236.90, 236.91, 236.99, 237.0, 237.1, 237.2, 237.3, 237.4, 237.5, 237.6, 237.70, 237.71, 237.72, 237.9, 238.0, 238.1, 238.2, 238.3, 238.4, 238.5, 238.6, 238.7, 238.8, 238.9, 239.0, 239.1, 239.2, 239.3, 239.4, 239.5, 239.6, 239.7, 239.8, 239.9
Carbohydrate transport and metabolism
—271, 271.8, 271.0, 271.3, 271.1, 271.9
Cardiomyopathy, primary
—425.4
Celiac sprue
—579.0
Cerebral palsy
—343.0, 343.1, 343.2, 343.3, 343.4, 343.8, 343.9
Cervical rib
—756.2
Charcot-Marie-Tooth
—356.1
Cholelithiasis
—574.0, 574.01, 574.02, 574.11, 574.12, 574.21, 574.22, 574.30, 574.31, 574.40, 574.41, 574.50, 574.51, 574.60, 574.61, 574.70, 574.71, 574.80, 574.81, 574.90, 574.91
Cholera
—001.0, 001.1, 001.9
Chondrodystrophy
—756.4
Congenital absence of vertebra
—756.13
Congenital spinal fusion
—756.15
Cystic fibrosis
—277.0, 277.00, 277.01, 277.02, 277.03, 277.09
Dejerine-Sottas disease
—356.0
Depression
Dermatomyositis-polymyositis
—710.3, 710.4
Diabetes mellitus type 1
—250.01, 250.03, 250.11, 250.13, 250.21, 250.23, 250.31, 250.33, 250.41, 250.43, 250.51, 250.53, 250.61, 250.63, 250.71, 250.73, 250.81, 250.83, 250.91, 250.93
Diabetes mellitus type 2
—250.00, 250.02, 250.10, 250.12, 250.20, 250.22, 250.30, 250.32, 250.40, 250.42, 250.50, 250.52, 250.60, 250.62, 250.70, 250.72, 250.80, 250.82, 250.90, 250.92
Diphtheria
—032.0, 032.1, 032.2, 032.3, 032.81, 032.82, 032.83, 032.84, 032.85, 032.89, 032.9
E. coli intestinal
—008.00, 008.01, 008.02, 008.03, 008.04, 008.09
Edward’s
—758.2
Enzyme-deficiency (hemolytic anemia)
—282.3
Epilepsy
—345.00, 345.01, 345.10, 345.11, 345.2, 345.3, 345.40, 345.41, 345.50, 345.51, 345.60, 345.61, 345.70, 345.71, 345.80, 345.81, 345.90, 345.91
Erythematous-squamous dermatosis
—690
Food poisoning
—005.0, 005.1, 005.2, 005.3, 005.4, 005.81, 005.89, 005.9
Friedreich’s ataxia
—334.0
Fragile X syndrome
— 759.83
Giant cell arteritis
— 446.5
Gram-negative bacteria (infection)
— 038.40, 038.41, 038.42, 038.43, 038.44, 038.49, 041.3, 041.4, 041.5, 041.6, 041.7
Goiter
Goodpasture’s syndrome
— 446.21
Gout
— 274, 274.1, 274.11, 274.19, 274.8, 274.81, 274.82, 274.89
HIV
— 042
Helicobacter pilori infection
— 041.86, 041.89, 041.9
Hemivertebra
— 756.14
Hepatitis A
— 070.0, 070.1
Hepatitis B
— 070.20, 070.21, 070.22, 070.23, 070.30, 070.31, 070.32, 070.33
Hepatitis C
— 070.41, 070.44, 070.51, 070.54, 070.70, 070.71
Hepatitis D
— 070.42, 070.52
Hepatitis E
— 070.43, 070.53
Hereditary spastic paraplegia
— 334.1
Hodgkin’s disease
— 201.9
Hyperosmolality
— 276.0
Hypoosmolality
— 276.1
Hypersensitivity angiitis
— 446.20, 446.29
Hypertrophic obstructive cardiomyopathy
— 425.1
Hypoglycemia
— 251.1, 251.0
Ichthyosis congenita
— 751.1
Kawasaki’s disease
— 446.1
Keratoderma
— 701.1
Klippel-Feil syndrome
— 756.16
Leprosy
— 030.0, 030.1, 030.2, 030.3, 030.8, 030.9
Lethal midline granuloma
— 446.3
Leukodystrophy
Lipid metabolism disorders
—272, 272.0, 272.1, 272.2, 272.3, 272.4, 272.5, 272.6, 272.7, 272.8, 272.9
Lown-Ganong-Levine syndrome
—426.81
Lumbosacral spondylolysis
—756.11
Lymphosarcoma
Meningococcus
—036.0, 036.1, 036.2, 036.3, 036.40, 036.41, 036.42, 036.43, 036.81, 036.82, 036.89, 036.9
Migraine
—346.00, 346.01, 346.10, 346.11, 346.20, 346.21, 346.80, 346.81, 346.90, 346.91
Mineral metabolism disorders (Fe)
—275.0
Mineral metabolism disorders (Cu)
—275.1
Mineral metabolism disorders (Mg)
—275.2
Mineral metabolism disorders (Ca)
—275.3, 275.4, 275.41, 275.42, 275.49
Mitochondrial disorders
—277.87
Moyamoya
—437.5
Multiple epiphyseal dysplasia
—756.56
Multiple sclerosis
—340
Mumps
—072.0, 072.1, 072.2, 072.3, 072.71, 072.72, 072.79, 072.8, 072.9
Muscular distrophy
—359.1
Mycoplasma
—041.81
Myotonic disorders
—359.2
Neurmyelitis optica
—341.0
Neurofibromatosis
—237.70, 237.71, 237.72, 237.7
Ornithosis
—073.0, 073.7, 073.8, 073.9
Osteogenesis imperfecta
—756.51
Osteopetrosis
—756.52
Parkinson’s disease
—332, 332.0, 332.1
Patau’s syndrome
—758.1
Pertussis
—033.0, 033.1, 033.8, 033.9
Pervasive developmental disorders
—299.00, 299.01, 299.10, 299.11, 299.80, 299.81, 299.90, 299.91
Phenylketonuria
—270.1
Pick’s disease
—331.11, 331.19
Plague
—020.0, 020.1, 020.2, 020.3, 020.4, 020.5, 020.8, 020.9
Plasma cell leukemia
—203.1
Pneumonia
—038.2, 041.2
Polionyelitis
—045.00, 045.01, 045.02, 045.03, 045.10, 045.11, 045.12, 045.13, 045.20, 045.21, 045.22, 045.23, 045.90, 045.91, 045.92, 045.93
Polyarteritis nodosa
—4460
Polyostotic fibrous dysplasia of bone
—756.54
Prader-Willi’s syndrome
—759.81
Primary cerebellar degeneration
—334.2
Prion-related disorders
—046.0, 046.1
Progeria
—259.8
Psoriasis
—696.1, 696
Reiter’s syndrome
—099.3, 711.10, 711.11, 711.12, 711.13, 711.14, 711.15, 711.16, 711.17, 711.18, 711.19
Renal glycosuria
—271.4
Reticulosarcoma
—200.00, 200.01, 200.02, 200.03, 200.04, 200.05, 200.06, 200.07, 200.08
Rheumatoid arthritis
—714.0, 714, 714.2, 714.30, 714.32, 714.33
Systemic lupus erythematosus
—710.0
Salmonella
—003.0, 003.1, 003.20, 003.21, 003.22, 003.23, 003.24, 003.29, 003.8, 003.9
Schilder’s syndrome
—341.1
Schizophrenia
Scleroderma
—710.1
Shigella
—004.0, 004.1, 004.2, 004.3, 004.8, 004.9
Sjogren’s syndrome
—047.0, 047.1, 047.8, 048, 049.0, 049.1, 049.8, 049.9, 050.0, 050.1, 050.2, 050.9, 051.0, 051.1, 051.2, 051.9, 052.0, 052.1, 052.7, 052.8, 052.9, 053.0, 053.10, 053.11, 053.12, 053.13, 053.19, 053.20, 053.21, 053.22, 053.29, 053.71, 053.79, 053.8, 053.9, 054.0, 054.10, 054.11, 054.12, 054.13, 054.19, 054.2, 054.3, 054.40, 054.41, 054.42, 054.43, 054.44, 054.49, 054.5, 054.6, 054.71, 054.72, 054.73, 054.79, 054.8, 054.9, 055.0, 055.1, 055.2, 055.71, 055.79, 055.8, 055.9, 056.00, 056.01, 056.10, 056.09, 056.71, 056.79, 056.8, 056.9, 057.0, 057.8, 057.9, 060.0, 060.1, 060.9, 061, 062.0, 062.1, 062.2, 062.3, 062.4, 062.5, 062.8, 062.9, 063.0, 063.1, 063.2, 063.8, 063.9, 064, 065.0, 065.1, 065.2, 065.3, 065.4, 065.8, 065.9, 066.0, 066.1, 066.2, 066.3, 066.40, 066.41, 066.42, 066.49, 066.8, 066.9

**Viral infections of the central nervous system (CNS)**
— 046.2, 046.3, 046.8, 046.9

**Vitamin deficiency**
—266.2, 266.9, 269.1

**WPW**
—426.7

**Wegener’s granulomatosis**
—446.4
2 Disease list and descriptions: A

2.1 Amino acid (AA) metabolism (aromatic) (Albinism, Alkaptonuria, Tyrosinemia, Waardenburg syndrome)

2.1.1 Albinism

Albinism is a lack of pigmentation in the eyes, skin and hair. Albinism is an inherited condition resulting from the combination of recessive alleles passed from both parents of an individual. The gene which results in albinism prevents the body from making the usual amounts of the pigment melanin. About 1 in 17,000 people have some type of albinism, although up to 1 in 75 are carriers.

Source:
http://en.wikipedia.org/wiki/Albinism

2.1.2 Alkaptonuria

Alkaptonuria also known as alcaptonuria or ochronosis is a rare inherited genetic disorder of tyrosine metabolism. This is an autosomal recessive trait that is caused by a defect in the enzyme homogentisic acid oxidase. The enzyme normally breaks down a toxic tyrosine byproduct, homogentisic acid (also called alkapton), which is harmful to bones and cartilage and is excreted in urine. A distinctive characteristic of alkaptonuria is that urine exposed to air turns dark (or black) after several hours because of the homogentisic acid. In adulthood, persons suffering from alkaptonuria develop progressive arthritis (especially of the spine), due to its degenerative effects on bones and cartilage.

Source:
http://en.wikipedia.org/wiki/Alkaptonuria

2.1.3 Tyrosinemia

Tyrosinemia (or "Tyrosinaemia") is an error of metabolism, usually inborn, in which the body can not effectively break down the amino acid tyrosine, found in most animal and plant proteins. Tyrosinemia is inherited in an autosomal recessive pattern.

Source:
http://en.wikipedia.org/wiki/Tyrosinemia

2.1.4 Waardenburg syndrome

Waardenburg syndrome is a rare genetic disorder most often characterized by varying degrees of deafness, minor defects in structures arising from the neural crest, and pigmentation anomalies. It is named after Dutch ophthalmologist Petrus Johannes Waardenburg, who first described it. Other names for the syndrome include Van der Hoeve / Halbertsma / Gualdi syndrome, Ptosis-Epicanthus syndrom, and Mende syndrome.

Source:
http://en.wikipedia.org/wiki/Waardenburg_symdrome
2.2 Amino acid (AA) metabolism (branched) (maple syrup urine disease)

2.2.1 Maple syrup urine disease

Maple syrup urine disease (MSUD) is an inherited metabolic disorder due to a deficiency of decarboxylase enzyme that leads to elevated concentrations of leucine, isoleucine, and valine (branched amino acids) in the blood and urine. Characterized by the urine having an odor similar to that of maple syrup. This results in severe mental retardation, and seizures. Also called branched chain ketoaciduria. Maple syrup urine disease affects an estimated 1 in 185,000 infants worldwide.

Source:

2.3 Amino acid (AA) metabolism (Lowe) (Beta-alaninemia, Hydroxyprolinemia, Hyperprolinemia, Sarcosinemia)

2.3.1 Beta-alaninemia

Beta-alaninemia is an inherited metabolic disorder characterized by deficiency of a certain enzyme. Symptoms include seizures and drowsiness but untreated it can result in death.

Source:
http://www.wrongdiagnosis.com/medical/beta_alaninemia.htm

2.3.2 Hydroxyprolinemia

Hydroxyproline is an imino acid normally present in human plasma. It is derived primarily from endogenous collagen turnover and the breakdown of dietary collagen. The finding of elevated (5- to 10-fold increase from the normal of less than 50 micromoles) serum hydroxyproline is thought to be an inherited defect in the catabolism of hydroxyproline. Even when only 1 case, in a female, was reported, this condition was thought to be an autosomal recessive because of its nature as an inborn error of metabolism and because the parents were thought to have been sibs (Scriver and Efron, 1972).

Source:

2.3.3 Hyperprolinemia

Hyperprolinemia Type I (HP-I) is characterized by abnormally high levels of proline in the blood. The high level of blood proline is the result of a deficiency of the enzyme proline oxidase, which is essential to the normal breakdown (metabolism) of proline. There are often no clinical manifestations of HP-I. Hyperprolinemia II (HP-II) results from the deficiency of another enzyme and also results in high blood proline levels, as well as other more severe clinical manifestations than are seen in HP-I. Mild mental retardation and convulsions are commonly associated with HP-II.

Source:
http://www.webmd.com/hw/raising_a_family/nord581.asp
2.3.4 **Sarcosinemia**

Sarcosinemia is an autosomal recessive condition characterized by an increased concentration of sarcosine in plasma and urine, due to sarcosine dehydrogenase deficiency. Sarcosinemia is most probably a benign condition without significant clinical problems. Mutations in the gene for sarcosine dehydrogenase, located on chromosome 9q34. Prevalence has been estimated to be 1:28,000 to 1:350,000 in newborn screening programs.

*Source:*
http://www.orpha.net/data/patho/GB/uk-sarco.pdf

2.4 **Amino acid (AA) metabolism (sulfur-bearing) (Homocystinuria)**

2.4.1 **Homocystinuria**

Homocystinuria, also known as Cystathionine beta synthase deficiency, is an inherited disorder of the metabolism of the amino acid methionine. It is an inherited autosomal recessive trait, which means the child is to inherit the defective gene from both parents. This defect leads to a multisystemic disorder of the connective tissue, muscles, CNS, and cardiovascular system. Homocystinuria represents a group of hereditary metabolic disorders characterized by an accumulation of homocysteine in the serum and an increased excretion of homocysteine in the urine. Infants appear to be normal and early symptoms, if any are present, are vague.

*Source:*
http://en.wikipedia.org/wiki/Homocystinuria

2.5 **Amino acid (AA) metabolism (straight-chain) (Hyperglycinemia, Hyperlysinemia, Pipecolic acidemia, Saccharopinuria)**

2.5.1 **Hyperglycinemia**

Hyperglycinemia is an autosomal recessive metabolic disorder caused by deficiencies in the mitochondrial glycine cleavage system. Clinical features include seizures, hypotonia, apnea, and coma. When the illness presents in childhood there tends to be an associated progressive dementia accompanied by extrapyramidal signs.

*Source:*
http://www.wrongdiagnosis.com/medical/hyperglycinemia.htm

2.5.2 **Hyperlysinemia**

Hyperlysinemia is a group of hereditary disorders characterized by an abnormal increase of lysine in the blood and associated with mental retardation, convulsions, and anemia.

*Source:*
http://en.wikipedia.org/wiki/Hyperlysinemia

2.5.3 **Pipecolic acidemia**

Pipecolic acidemia formerly described patients with accumulated pipecolic acid in the blood and/or urine. Pipecolic acid is an intermediate compound in the lysine metabolism that is synthetised by the pipecolic oxidase in peroxysomes. Recent findings show that the vast majority of pipecolicacidemias are actually due to general peroxysome storage disorders. However a few exceptional patients have isolated cases of pipecolicacidemia combined with mental retardation, hypotonia, or Joubert’s syndrome. There is no specific treatment.
2.5.4 Saccharopinuria

Saccharopinuria a variant form of hyperlysine mia caused by a partial enzyme deficiency.

**Source:**
http://en.wikipedia.org/wiki/Saccharopinuria

2.6 Amino acid (AA) transport (Cystinosis, Cystinuria, Hartnup disease)

2.6.1 Cystinosis

Cystinosis is a hereditary disorder of the renal tubules characterized by the presence of carbohydrates and amino acids in the urine, excessive urination, and low blood levels of potassium ions and phosphates. The body accumulates the amino acid cystine within cells. Excess cystine forms crystals that can build up and damage cells. These crystals negatively affect many systems in the body, especially the kidneys and eyes.

**Source:**
http://en.wikipedia.org/wiki/Cystinosis

2.6.2 Cystinuria

Cystinuria is characterized by the inadequate reabsorption of cystine during the filtering process in the kidneys, thus resulting in an excessive concentration of this amino acid. Cystine will precipitate out of the urine, if the urine is neutral or acidic, and form crystals or stones in the kidneys, ureters, or bladder. The disorder affects 1 in 10,000 people and is inherited in an autosomal recessive pattern.

**Source:**
http://en.wikipedia.org/wiki/Cystinuria

2.6.3 Hartnup disease

Hartnup disease is a genetic metabolic disorder in the absorption of the amino acid tryptophan that leads to the insufficient production of nicotinamide. Nicotinamide is necessary for neutral amino acid transporter production in the proximal renal tubules found in the kidney, and intestinal mucosal cells found in the small intestine. Therefore, a symptom stemming from this disorder results in increased amounts of amino acids in the urine. Pellagra is also caused by low nicotinamide; this disorder results in dermatitis, diarrhea and dementia.

**Source:**

2.7 Acanthosis nigricans

A skin condition characterized by dark thickened velvety patches, especially in the folds of skin in the axilla (armpit), groin and back of the neck. The condition is complex. It can occur with endocrine diseases such as Cushing disease, tumors of the pituitary, and diabetes mellitus. It is common in people who have insulin resistance — whose body is not responding correctly to the insulin that they make in their pancreas. Acanthosis nigricans also occurs with underlying malignancies (especially carcinomas of the viscera), administration of certain drugs, and as a genetic disorder inherited in an autosomal dominant manner.
2.8 Acidosis

Too much acid in the body, a distinctly abnormal condition resulting from the accumulation of acid or from the depletion of alkaline reserves. In acidosis, the pH of the blood is abnormally low. Acidosis is associated with diabetic ketoacidosis, lung disease, and severe kidney disease. The opposite of acidosis is alkalosis in which there is too high a pH due to excess base or insufficient acid in the body.

2.9 (Organic) Aciduria

Organic acidurias are a class of inherited metabolic diseases characterized by urinary excretion of abnormal amounts or types of organic acids. Most of them affect more than one organ system. Some affect central nervous system development, causing developmental delay. Some affect physical growth. Some cause episodic illnesses with vomiting and metabolic acidosis; some of these may be precipitated by prolonged fasting, minor viral infections, or any other catabolic state. Some are associated with hypoglycemia and ketosis or ketoacidosis. Most of the organic acidoses result from defective autosomal genes for various enzymes important to intermediary metabolism. Typically, most of the harm is caused by an impaired ability to synthesize an essential substance or by toxicity to specific organs from excessive amounts of a specific metabolite. Most are inherited as autosomal recessive diseases. The diagnosis is usually made by detecting an abnormal pattern of organic acids in a urine sample by tandem mass spectrometry. In some conditions, the urine is always abnormal, in others the characteristic substances are only present intermittently. There are no effective treatments for some of the conditions. Treatments for some of the others may include avoidance of fasting, extra carbohydrates during illness, or intravenous fluid and dextrose to reverse catabolism. Some can be ameliorated with extra vitamins or other metabolic substrates.

2.10 Actinomycosis

Actinomycosis is a rare infectious disease of humans caused by Actinomyces bacteria. Characterized by the formation of painful abscesses in the mouth, lungs, or digestive organs, actinomycosis abscesses grow larger as the disease progresses, often over a period of months. In severe cases, the abscesses may penetrate the surrounding bone and muscle to the skin, where they break open and leak large amounts of pus. Actinomycosis occurs in cattle and other animals as a disease called lumpy jaw. This name refers to the large abscesses that grow on the head and neck of the infected animal.
2.11 Acute leukemia

Acute leukemia is a disease of the leukocytes and their precursors. It is characterized by the appearance of immature, abnormal cells in the bone marrow and peripheral blood and frequently in the liver, spleen, lymph nodes, and other parenchymatous organs. The clinical picture is marked by the effects of anemia, which is usually severe (fatigue, malaise), an absence of functioning granulocytes (proneness to infection and inflammation), and thrombocytopenia (hemorrhagic diathesis). The spleen and liver usually are moderately enlarged, while enlarged lymph nodes are seen mainly in the pediatric lymphoblastic leukemias. Fever and a very high ESR complete the picture. Leukocyte counts vary greatly in the acute leukemias. About one-fourth to one-third of cases begin with a low white blood count (sub- or aleukemic leukemia), while about half show some degree of leukocytosis. Mature granulocytes may still be found in the peripheral blood in addition to abnormal forms. The coexistence of immature and mature cell forms is termed “hiaetus leucaemicus.” The leukocytopenic forms are the most difficult to differentiate from aplastic anemias, pancytopenias, and the myelodysplastic syndromes. Bone marrow aspiration is usually necessary to establish a diagnosis. Aspirated marrow is found to be permeated by abnormal cells (paramyeloblasts, paraleukoblasts, nonclassifiable cells (N. C.), leukemic cells, blasts, etc.) with little or no evidence of normal hematopoiesis.

Source:

2.12 Acute promyelocytic leukemia

Acute promyelocytic leukemia: Commonly called APL, a malignancy of the bone marrow in which there is a deficiency of mature blood cells in the myeloid line of cells and an excess of immature cells called promyelocytes. APL is due to a translocation (an exchange of chromosome material) between chromosomes 15 and 17 which is symbolized t(15;17). This translocation is not a mere marker of APL. It is the cause of APL. The signs and symptoms of APL are nonspecific and include fatigue (feeling tired), minor infections, or a tendency to bleed (hemorrhagic diathesis). There is usually pancytopenia with low levels of red blood cells (anemia), low levels of the granulocytes and monocytes (types of white blood cells that fight infections), and low levels of platelets (that are needed for blood to clot normally). Patients with APL may therefore receive transfusions. APL is consistently associated with a disorder that resembles (but is not identical to) disseminated intravascular coagulation (DIC). There is in APL a pronounced tendency to hemorrhage (bleeding). The bleeding can manifest itself as petechiae (little bleeding spots in the skin or elsewhere), small ecchymosis (bruises), epistaxis (nose bleeds), bleeding in the mouth, hematuria (blood in the urine), bleeding from venipuncture and bone marrow sites and in girls and women who are menstruating may have menometrorrhagia (excessive irregular menstrual bleeding). The hemorrhagic diathesis (bleeding condition) may precede the diagnosis of leukemia by 2-8 weeks.

Source:

2.13 Ainhum

Ainhum disease is the autoamputation of the fifth toe. This could be due to formation of a constricting scar around the base of the fifth toe. It is seen in Africa, in people walking barefoot.

Source:
http://en.wikipedia.org/wiki/Ainhum
2.14 Albright (-McCune)-Sternberg syndrome

A rare congenital developmental disorder beginning in childhood or early adolescence, combining polystotic fibrous dysplasia of the bone, café-au-lait pigmentation of the skin, and endocrine disorders, especially precocious puberty in girls. The long bones are most frequently affected. Short stature, round face, shortened metacarpals and metatarsals, difficulty in walking. Orthopaedic care may be necessary for disability resulting from pathologic fractures and bony overgrowth. Varying degree of mental retardation. Puberty is usually reached at 5 to 10 years of age. Predominantly in females (3:2). This condition is similar to the Jaffe-Lichtenstein syndrome, but distinguished from that by the patchy skin pigmentation and sexual precocity.

Source:
http://www.whonamedit.com/synd.cfm/1844.html

2.15 Alcoholism

Alcoholism is also known as “alcohol dependence.” It is a disease that includes alcohol craving and continued drinking despite repeated alcohol-related problems. Alcoholism includes four symptoms: craving, impaired control, physical dependence, and tolerance. It is a chronic and often progressive disease. Like many diseases, it has a generally predictable course and is influenced by both genetic (inherited) and environmental factors. It is estimated that 14 million people in the United States — 1 in every 13 adults — abuse alcohol or are alcoholic. More men than women are alcohol dependent or experience alcohol-related problems. Rates of alcohol problems are also highest among young adults ages 18–29 and lowest among adults 65 years and older.

Source:
http://www.medicinenet.com/alcohol_abuse_and_alcoholism/article.htm

2.16 Allergic rhinitis

Symptoms of allergic rhinitis frequently include nasal congestion, a clear runny nose, sneezing, nose and eye itching, and tearing eyes. Allergic rhinitis can lead to other diseases such as sinusitis and asthma. Commonly, allergic rhinitis is a result of an allergic person coming in contact several times with protein from plants. Approximately 5–10 % of Americans at times suffer from allergic rhinitis. A person is programmed to be allergic by his/her genetic makeup and is destined to be allergic from birth. Approximately one in four persons with allergic rhinitis also has asthma.

Source:
http://www.medicinenet.com/hay_fever/article.htm

2.17 Alkalosis

A dangerous decrease in the normal acidity of the blood. There is too much base in the blood and body. This is a distinctly abnormal condition. It results from the accumulation of base or from the depletion of acid. The pH of the alkalotic body is above normal. Alkalosis can be caused by high altitudes, hyperventilation, and excessive vomiting. The opposite of alkalosis is acidosis in which there is too low a pH due to excess acid or insufficient base in the body.

Source:
http://www.medterms.com/script/main/art.asp?articlekey=6852
2.18 Alopecia (baldness)

Alopecia, commonly known as baldness, is a set of disorders which involves the state of lacking hair where it would normally grow, especially on the head. The most common form of baldness is a progressive hair thinning condition called androgenic alopecia or 'male pattern baldness' that occurs in adult human males and some primate species. Nonetheless, the severity and nature of baldness can vary greatly; it ranges from male and female pattern alopecia (androgenetic alopecia, also called androgenic alopecia or alopecia androgenetica), alopecia areata, which involves the loss of some of the hair from the head, and alopecia totalis, which involves the loss of all head hair, to the most extreme form, alopecia universalis, which involves the loss of all hair from the head and the body.

Source: http://en.wikipedia.org/wiki/Alopecia

2.19 Alopecia areata

Alopecia areata is a hair loss condition which usually affects the scalp. It can, however, sometimes affect other areas of the body. Hair loss tends to be rather rapid and often involves one side of the head more than the other. Alopecia areata affects both males and females. This type of hair loss is different than male pattern baldness, an inherited condition. Current evidence suggests that alopecia areata is caused by an abnormality in the immune system. This particular abnormality leads to autoimmunity. Alopecia areata is sometimes associated with other autoimmune conditions such as allergic disorders, thyroid disease, vitiligo, lupus, rheumatoid arthritis, and ulcerative colitis. Sometimes, alopecia areata occurs within family members, suggesting a role of genes and heredity.

Source: http://www.medicinenet.com/alopecia_areata/article.htm

2.20 Alzheimer’s disease

The clinical hallmarks of Alzheimer’s disease are progressive impairment in memory, judgment, decision making, orientation to physical surroundings, and language. Alzheimer disease is the most common of all neurodegenerative diseases. It accounts for about two-thirds of cases of dementia with vascular causes and other neurodegenerative diseases making up most of the rest. According to the Alzheimer’s Association, about 4 million Americans suffer from the disease. There is no cure for Alzheimer’s disease. The average time of survival from the initial diagnosis of Alzheimer’s disease was found (in a study reported in 2004) to be 4.2 years for men and 5.7 years for women. Men had poorer survival across all age groups compared with women and survival was decreased in all age groups compared with the life expectancy of the US population.

The German psychiatrist and pathologist Alois Alzheimer (1864–1915) first described this form of presenile dementia in 1907.


2.21 Amebiasis

Infection of the intestines with amebae, especially with the ameba Entamoeba histolytica, characterized by frequent, loose stools flecked with blood and mucus.

2.22 Amyotrophic lateral sclerosis

Amyotrophic lateral sclerosis (also called ALS or “Lou Gehrig’s disease”) is a classic motor neuron disease. ALS is progressive and fatal. These illnesses are often infections. ALS occurs most often in adults in the fifth through seventh decades of life. It progressively leads to death in 2 to 7 years. The cause is unknown. All forms of ALS cause progressive muscle weakness and wasting. Spontaneous tiny local areas of muscle twitching, called fasciculations, are characteristic in most patients.

Source:
http://www.medicinenet.com/amyotrophic_lateral_sclerosis/article.htm

2.23 Anaerobic infection

An anaerobic infection is an infection caused by bacteria (called anaerobes) which cannot grow in the presence of oxygen. Anaerobic bacteria can infect deep wounds, deep tissues, and internal organs where there is little oxygen. Anaerobes tend to invade skin and muscle tissue that has been damaged by injury or surgery—particularly if the tissue has a poor blood supply. Spontaneous infections sometimes develop in people who have certain cancers or a weakened immune system. Also common are infections in the mouth.

Source:
http://www.healthatoz.com/healthatoz/Atoz/ency/anaerobic_infections.jsp

2.24 Aniridia (eye abnormalities)

Rare, congenital absence or partial absence of the iris; genetically caused by an autosomal dominant or recessive hereditary pattern. Often, the iris is vestigial (little more than a margin is present) and the eye appears to have no color (only a larger than normal pupil). Other deformities of the anterior chamber are also often present (e.g., cataract), and glaucoma frequently develops before adolescence. There is usually decreased acuity (circa 20/200), photophobia, possible nystagmus, cataracts, displaced lens, and underdeveloped retina; visual fields are usually normal, unless glaucoma develops.

Source:
http://www.tsbvi.edu/Education/anomalies/aniridia.htm

2.25 Ankylosing spondylitis

Ankylosing spondylitis is a form of chronic inflammation of the spine and the sacroiliac joints. Chronic inflammation in these areas causes pain and stiffness in and around the spine. Over time, chronic spinal inflammation (spondylitis) can lead to a complete cementing together (fusion) of the vertebrae, a process referred to as ankylosis. Ankylosis leads to loss of mobility of the spine. Ankylosing spondylitis is also a systemic rheumatic disease, meaning it can affect other tissues throughout the body. Accordingly, it can cause inflammation in or injury to other joints away from the spine, as well as other organs, such as the eyes, heart, lungs, and kidneys. Ankylosing spondylitis shares many features with several other arthritis conditions, such as psoriatic arthritis, reactive arthritis, and arthritis associated with Crohn's disease and ulcerative colitis. Ankylosing spondylitis is 2–3 times more common in males than in females. Ankylosing spondylitis affects all age groups, including children. The most common age of onset of symptoms is in the second and third decades of life. The tendency for developing ankylosing spondylitis is believed to be genetically inherited, and the majority (nearly 90%) of patients with ankylosing spondylitis are born with the HLA-B27 gene. The onset of pain and stiffness is usually gradual and progressively worsens over months.
2.26 Anthrax

Anthrax is a highly infectious disease that normally affects animals, especially ruminants (such as goats, cattle, sheep, and horses), but which can be transmitted to humans by contact with infected animals or their products or by biological warfare. The agent of anthrax is a bacteria called Bacillus anthracis that looks like a large rod under the microscope. Anthrax is now rare in humans in the United States and comparable countries. The incubation period is relatively short. It generally ranges from 3 to 5 days but can be as brief as 12 hours. There are three forms of disease caused by anthrax: cutaneous (skin) anthrax, pulmonary (lung) anthrax, and gastrointestinal (intestinal) anthrax.

Source:
http://www.medicinenet.com/anthrax/article.htm

2.27 Aortic aneurysm

The aorta bulges at the site of the aneurysm like a weak spot on an old worn tire. Aortic aneurysms can develop anywhere along the length of the aorta. The majority, however, are located along the abdominal aorta. Abdominal aortic aneurysms are most common after age 60. Males are 5 times more likely than females to be affected. Approximately 5% of men over age 60 develop an abdominal aortic aneurysm. The most common cause of aortic aneurysms is “hardening of the arteries” called arteriosclerosis. At least 80% of aortic aneurysms are from arteriosclerosis. Most abdominal aortic aneurysms produce no symptoms (they are asymptomatic). When they produce symptoms, the most common symptom is pain.

Source:

2.28 Aplastic anemia

Anemia due to failure of the bone marrow to produce blood cells, including red and white blood cells as well as platelets. Aplastic anemia frequently occurs without a known cause. Known causes include exposure to chemicals (benzene, toluene in glues, insecticides, solvents), drugs (chemotherapy, gold, seizure medications, antibiotics, and others), viruses (HIV, Epstein-Barr), radiation, immune conditions (systemic lupus erythematosus, rheumatoid arthritis), pregnancy, paroxysmal nocturnal hemoglobinuria, and inherited disorders (Fanconi’s anemia). Symptoms of aplastic anemia include fatigue, bruising, bleeding, shortness of breath, fever, chills, and less frequently, bone pain.

Source:

2.29 Attention Deficit Hyperactivity Disorder

Attention Deficit Hyperactivity Disorder (ADHD) is a condition that becomes apparent in some children in the preschool and early school years. It is hard for these children to control their behavior and/or pay attention. It is estimated that between 3 and 5 percent of children have ADHD, or approximately 2 million children in the United States. This means that in a classroom of 25 to 30 children, it is likely that at least one will have ADHD.
ADHD was first described by Dr. Heinrich Hoffman in 1845. A physician who wrote books on medicine and psychiatry, Dr. Hoffman was also a poet who became interested in writing for children when he couldn’t find suitable materials to read to his 3-year-old son. The result was a book of poems, complete with illustrations, about children and their characteristics. “The Story of Fidgety Philip” was an accurate description of a little boy who had attention deficit hyperactivity disorder. Yet it was not until 1902 that Sir George F. Still published a series of lectures to the Royal College of Physicians in England in which he described a group of impulsive children with significant behavioral problems, caused by a genetic dysfunction and not by poor child rearing—children who today would be easily recognized as having ADHD. Since then, several thousand scientific papers on the disorder have been published, providing information on its nature, course, causes, impairments, and treatments.

A child with ADHD faces a difficult but not insurmountable task ahead. In order to achieve his or her full potential, he or she should receive help, guidance, and understanding from parents, guidance counselors, and the public education system. This document offers information on ADHD and its management, including research on medications and behavioral interventions, as well as helpful resources on educational options.

Because ADHD often continues into adulthood, this document contains a section on the diagnosis and treatment of ADHD in adults.

Source:
http://www.nimh.nih.gov/publicat/adhd.cfm

2.30 Autism

Autism is a developmental disorder that is characterized by impaired development in communication, social interaction, and behavior. The range of these disorders varies from severely impaired individuals with autism to other individuals who have abnormalities of social interaction but normal intelligence—Asperger’s syndrome. Autism is, undoubtedly, a biologically-based disorder. In support of a biological theory of autism, several known neurological disorders are associated with autistic features. Autism is one of the symptoms of these disorders. These conditions include tuberous sclerosis (an inherited disorder), the fragile X syndrome, cerebral dysgenesis (abnormal development of the brain), Rett syndrome, and some of the inborn errors of metabolism (biochemical defects). There is a strong association between autism and seizures.

Source:
http://www.medicinenet.com/autism/article.htm

3 Disease list and descriptions: B

3.1 Behcet’s syndrome

Behcet’s syndrome is classically characterized as a triad of symptoms that include recurring crops of mouth ulcers (called aphthous ulcers), genital ulcers, and inflammation of a specialized area around the pupil of the eye, the uvea. Behcet’s syndrome is also sometimes referred to as Behcet’s disease. The cause of Behcet’s syndrome is not known. The disease is more frequent and severe in patients from the Eastern Mediterranean and Asia than those of European descent. Both inherited (genetic) and environmental factors, such as microbe infections, are suspected to be factors that contribute to the development of Behcet’s syndrome. Behcet’s syndrome has not been felt to be contagious.

Source:
http://www.medicinenet.com/behcets_syndrome/article.htm
3.2 Benign neoplasms

A benign tumor is one that does not spread or “metastasize” to other parts of the body. A benign tumor is caused by cell overgrowth, and thus is different from a cyst or an abscess. Although benign is better news than malignant for a cancer biopsy, it does not always mean “harmless,” though many are almost harmless. Benign tumors cause more than 13,000 annual deaths in the USA, compared to more than 500,000 annual deaths from cancer (i.e. from malignant tumors).

Source:
http://www.wrongdiagnosis.com/b/benign/intro.htm

3.3 Bipolar disorder

Bipolar disorder, also known as manic-depressive illness, is a brain disorder that causes unusual shifts in a person’s mood, energy, and ability to function. Different from the normal ups and downs that everyone goes through, the symptoms of bipolar disorder are severe. Bipolar disorder can be treated, and people with this illness can lead full and productive lives. More than 2 million American adults, or about 1 percent of the population age 18 and older in any given year, have bipolar disorder. Bipolar disorder typically develops in late adolescence or early adulthood.

Source:
http://www.medicinenet.com/bipolar_disorder/article.htm

3.4 Breast cancer (male)

Like breast cancer in women, cancer of the male breast results from the uncontrolled growth of cells within this breast tissue. Male breast cancer is a rare condition, accounting for only about 1% of all breast cancers. The American Cancer Society estimates that approximately 1,690 new cases of male breast cancer will be diagnosed in 2005, resulting in about 460 deaths (in comparison, over 40,000 women die of breast cancer each year). Most cases of breast cancer are detected in men between the ages of 60 and 70, although the condition can develop in men of any age. Infiltrating ductal carcinoma is the most common type of male breast cancer. A lump beneath the nipple is the most common symptom of male breast cancer. Male breast cancer is staged (reflecting the extent of tumor spread) identically to breast cancer in women. Surgery is the most common initial treatment for male breast cancer; chemotherapy, radiation therapy, and hormonal therapy are also administered. The prognosis of male breast cancer, like breast cancer in women, is predominantly influenced by tumor stage.

Source:
http://www.medicinenet.com/male_breast_cancer/article.htm

3.5 Breast cancer

Other than skin cancer, breast cancer is the most common type of cancer among women in the United States. More than 180,000 women are diagnosed with breast cancer each year. Breast cancer is not just one disease, but rather is a general term used to describe a number of different types of cancers which occur in the breast. Each different type of breast cancer behaves differently and has a different prognosis. The exact causes of breast cancer are not known. However, studies show that the risk of breast cancer increases as a woman gets older. This disease is very uncommon in women under the age of 35. Most breast cancers occur in women over the age of 50, and the risk is especially high for women over age 60. Also, breast cancer occurs more often in white women than African American or Asian women.

Source:
http://www.medicinenet.com/breast_cancer/article.htm
3.6 Brucellosis

An infectious disease due to the bacteria *Brucella* that causes rising and falling (undulant) fevers, sweats, malaise, weakness, anorexia, headache, myalgia (muscle pain) and back pain. It also called undulant fever because the fever is typically undulant, rising and falling like a wave. Brucellosis is transmitted through contaminated and untreated milk and milk products and by direct contact with infected animals (cattle, sheep, goats, pigs, camels, buffaloes, wild ruminants and, very recently, seals), and animal carcasses. The incubation period of brucellosis is usually one to three weeks, but sometimes may be several months after exposure. Brucellosis is an extremely variable disease. Millions of people worldwide are at risk for the disease, especially in developing countries where the infection in animals has not been brought under control, heat treatment procedures of milk (e.g. pasteurization) are not routinely applied, and food habits such as consumption of raw milk and poor hygienic conditions favor human infection. In the US, there are fewer than 0.5 cases per 100,000 population. Most cases are reported from California, Florida, Texas, and Virginia.

*Source:*


3.7 Budd-Chiari syndrome

Budd-Chiari syndrome is clotting of the hepatic vein, the major vein that leaves the liver. Most patients with Budd-Chiari syndrome have an underlying condition that predisposes to blood clotting. The most common symptom in Budd-Chiari syndrome is ascites, or fluid accumulation in the abdomen. Some individuals with Budd-Chiari syndrome may be jaundiced (yellow skin). In the US, Budd-Chiari syndrome is rare. Budd-Chiari syndrome is a potentially fatal disorder if untreated. The syndrome occurs in persons of all races. The syndrome is equally present in both sexes. Emergent presentation is more common in women than in men. Age at presentation is usually the third or fourth decade of life, although the condition may also occur in children or elderly persons.

*Source:*

http://www.cumc.columbia.edu/dept/gi/BC.html
http://www.emedicine.com/MED/topic2694.htm

3.8 Bundle branch block

Bundle branch block (BBB) is a disruption in the normal flow of electrical pulses that drive the heart beat. Bundle branch block belongs to a group of heart problems called intraventricular conduction defects (IVCD). Left bundle branch block usually happens as a consequence of other diseases such as arteriosclerosis, rheumatic fever, congenital heart disease, myocarditis, myocardial infarction, metastatic heart tumors, or other invasions of the heart tissue. Right bundle branch block happens less often from underlying heart disease.

*Source:*

http://www.chfpatients.com/text/bbb.txt

3.9 Buphthalmos

The ocular globe is usually large as a result of the increased intraocular pressure dating from intrauterine life, hence the term buphthalmos, meaning “ox eye.” In only about half of cases are both eyes involved, and males are affected somewhat more often than females. The defect is thought to involve the permeability of the trabeculum to aqueous humor. The primary mode of therapy is surgical. Autosomal recessive inheritance is quite certain in a significant proportion of glaucoma cases.
3.10 Burkitt lymphoma

Burkitt lymphoma is a lymph gland tumor classified as a non-Hodgkin’s type of lymphoma. This type of tumor was first discovered in Africa, but it has now been found in the U.S. as well. African Burkitt lymphoma is closely associated with the Epstein-Barr virus (EBV), the primary cause of infectious mononucleosis. The American form of Burkitt lymphoma is less closely associated with EBV. Both types of tumor are caused by defective immune cells called B lymphocytes. Burkitt lymphoma may first be noticed as a swelling of the lymph nodes (glands) in the neck or below the jaw. These swollen lymph nodes are often painless and can grow very rapidly. The cause of Burkitt lymphoma is not known, but in the African type of Burkitt lymphoma there is a strong association with early childhood infection by the Epstein-Barr virus. Burkitt lymphoma is usually curable if treated aggressively with chemotherapy. If the cancer involves only a small area of lymph nodes, the cure rate is over 90%. If it has spread to the bone marrow or spinal fluid, the cure rate drops to about 75%.

Source:

4 Disease list and descriptions: C

4.1 Carbohydrate transport and metabolism (Glycogen storage disease, Lactose intolerance, Galactosemia)

4.1.1 Glycogen storage disease

Glycogen storage disease (synonyms: glycogenosis, dextrinosis) is any one of several inborn errors of metabolism that result from enzyme defects that affect the processing of glycogen synthesis or breakdown within muscles, liver, and other cell types. There are nine diseases that are commonly considered to be glycogen storage diseases: GSD type I: glucose-6-phosphatase deficiency, von Gierke’s disease; GSD type II: acid maltase deficiency, Pompe’s disease; GSD type III: glycogen debrancher deficiency, Cori’s disease or Forbe’s disease; GSD type IV: glycogen branching enzyme deficiency, Andersen disease; GSD type V: muscle glycogen phosphorylase deficiency, McArdle disease; GSD type VI: liver phosphorylase deficiency, Hers’s disease; GSD type VII: muscle phosphfructokinase deficiency, Tarui’s disease; GSD type IX: phophorylase kinase deficiency; GSD type XI: glucose transporter deficiency, Fanconi-Bickel disease.

Source:
http://en.wikipedia.org/wiki/Glycogenosis

4.1.2 Lactose intolerance

Lactose intolerance is the condition in which lactase, an enzyme needed for proper metabolism of lactose, is not produced in adulthood. A hydrogen breath test is required for a clinical diagnosis. With lactose intolerance, the result of consuming lactose or a lactose-containing food is excess gas production and often diarrhea.

Source:
http://en.wikipedia.org/wiki/Lactose_intolerance
4.1.3 Galactosemia

Galactosemia is a rare genetic metabolic disorder which affects an individual’s ability to properly digest the sugar galactose. Lactose in food is broken down by the body into glucose and galactose. Normally, galactose is then converted into glucose by the enzyme GALT (galactose-1-phosphate uridylyltransferase). In individuals with galactosemia, GALT activity is severely diminished, leading to toxic levels of galactose to build up in the blood, resulting in hepatomegaly (an enlarged liver), renal failure, cataracts, and brain damage. Without treatment, mortality in infants with galactosemia is about 75%. Goppert first described the disease in 1917. Its incidence is about 1 per 47,000 births (classic type). It is much rarer in Japan.

Source:
http://en.wikipedia.org/wiki/Galactosemia

4.2 Carcinoma in situ

Carcinoma in situ (CIS) a an early form of carcinoma defined by the absence of invasion of surrounding tissues. CIS will usually not form a tumor. Rather, the lesion is flat (in the skin, cervix, etc) or follows the existing architecture of the organ (in the breast, lung, etc). Some CIS, however, form tumors, for example colon polyps or papillary cancer of the bladder. Many forms of cancer originate from a “carcinoma in situ” (CIS) lesion.

Source:
http://en.wikipedia.org/wiki/Carcinoma_in_situ

4.3 Cardiomyopathy

Cardiomyopathy is a weakening of the heart muscle or a change in heart muscle structure. It is often associated with inadequate heart pumping or other heart function abnormalities. While all types of cardiomyopathy can cause heart failure, each case requires specific strategies for recovery. Nonischemic cardiomyopathy is weakness in the muscle of the heart that is not due to coronary artery disease. To make a diagnosis of nonischemic cardiomyopathy, significant coronary artery disease should be ruled out.

Source:
http://heart-disease.health-cares.net/cardiomyopathy.php

4.4 Celiac sprue (celiac disease)

Celiac disease is a digestive disease that damages the small intestine and interferes with absorption of nutrients from food. People who have celiac disease cannot tolerate a protein called gluten, which is found in wheat, rye, barley, and possibly oats. When people with celiac disease eat foods containing gluten, their immune system responds by damaging the small intestine. Symptoms may or may not occur in the digestive system. For example, one person might have diarrhea and abdominal pain, while another person has irritability or depression. In fact, irritability is one of the most common symptoms in children.

Source:
http://digestive.niddk.nih.gov/ddiseases/pubs/celiac/
4.5  Cerebral palsy

Cerebral palsy (CP) is one of the most common conditions seen by pediatric neurologists. Many clinicians, however, would agree that CP is an abnormality of motor function (as opposed to mental function) that is acquired at an early age, usually less than a year of age, and is due to a brain lesion that is non-progressive. CP affects approximately 1 to 3 out of every thousand children born, making the condition not only a burden on the families, but also a significant financial burden on society. Up to 50% of patients with CP are mentally retarded. It is also just as important to note that many children with severe motor impairment due to CP are not intellectually impaired. Perhaps a third of all CP patients have seizures.

Source:

4.6  Cervical rib

A supernumerary (extra) rib which arises from the seventh cervical vertebra. It is located above the normal first rib. A cervical rib is present in only about 1 in 200 (0.5%) of people. It may cause nerve and artery problems.

Source:

4.7  Charcot-Marie-Tooth disease

Charcot-Marie-Tooth disease (CMT) is one of the most common inherited neurological disorders, affecting approximately 1 in 2,500 people in the United States. The disease is named for the three physicians who first identified it in 1886 — Jean-Martin Charcot and Pierre Marie in Paris, France, and Howard Henry Tooth in Cambridge, England. The neuropathy of CMT affects both motor and sensory nerves. A typical feature includes weakness of the foot and lower leg muscles, which may result in foot drop and a high-stepped gait with frequent tripping or falls. Later in the disease, weakness and muscle atrophy may occur in the hands, resulting in difficulty with fine motor skills. Onset of symptoms is most often in adolescence or early adulthood, however presentation may be delayed until mid-adulthood. The severity of symptoms is quite variable in different patients and some people may never realize they have the disorder. Progression of symptoms is very gradual. CMT is not fatal and people with most forms of CMT have a normal life expectancy. CMT is caused by mutations in genes that produce proteins involved in the structure and function of either the peripheral nerve axon or the myelin sheath. The gene mutations in CMT disease are usually inherited. Some forms of CMT are inherited in an autosomal dominant fashion, which means that only one copy of the abnormal gene is needed to cause the disease. Other forms of CMT are inherited in an autosomal recessive fashion, which means that both copies of the abnormal gene must be present to cause the disease. Still other forms of CMT are inherited in an X-linked fashion, which means that the abnormal gene is located on the X chromosome.

Source:
http://www.medicinenet.com/charcot-marie-tooth-disease/article.htm

4.8  Cholelithiasis (gallstones)

Gallstones are stones that form in the gall (bile). Gallstones usually form in the gallbladder; however, they also may form anywhere there is bile—in the intrahepatic, hepatic, common bile, and cystic ducts. Gallstones are common—they occur in approximately 20% of women in the US, Canada and Europe—but there is a large variation in prevalence among ethnic groups. These
differences probably are accounted for by genetic (hereditary) factors. First-degree relatives (parents, siblings, and children) of individuals with gallstones are $1\frac{1}{2}$ times more likely to have gallstones than if there were not a first-degree relative with gallstones. The majority of people with gallstones have no signs or symptoms and are unaware of their gallstones. (The gallstones are “silent.”)

Source: 
http://www.medicinenet.com/gallstones/article.htm

4.9 Cholera

Cholera is an acute, diarrheal illness caused by infection of the intestine with the bacterium Vibrio cholerae. The infection is often mild or without symptoms, but sometimes can be severe. Approximately 1 in 20 infected persons has severe disease characterized by: profuse watery diarrhea, vomiting, and leg cramps. In these persons, rapid loss of body fluids leads to dehydration and shock. Without treatment, death can occur within hours. Cholera can be simply and successfully treated by immediate replacement of the fluid and salts lost through diarrhea.

Source: 
http://www.medicinenet.com/cholera/article.htm

4.10 Chondrodystrophy (achondroplasia)

A disturbance that affects the development of the cartilage of the long bones and that especially involves the region of the epiphysial plates, resulting in arrested growth of the long bones. Achondroplasia is a genetic (inherited) condition that results in abnormally short stature. All persons with achondroplasia are little people. The average height of an adult with achondroplasia is 131 cm (52 inches, or 4 foot 4) in males and 124 cm (49 inches, or 4 foot 1) in females. The frequency of achondroplasia is estimated to range from about 1 in 10,000 births in Latin America to about 12 in 77,000 in Denmark. An average figure worldwide is approximately 1 in 25,000 births.

Source: 
http://www.answers.com/topic/chondrodystrophy

http://www.answers.com/main/ntquery;jsessionid=7irgrm00s4uad?method=4&dsid=501&dekey=achondroplasia&gwp=8&curtab=501_1&sbid=lc07a&linktext=achondroplasia

4.11 Congenital absence of vertebrae

Congenital absence of (sacrum and lumbar) vertebrae, is a rare and abnormal condition present at birth. It may be mild, involving the lack of the lower part of the tailbone (coccyx). In the severe form, it may involve the lack of the triangular bone that attaches to the pelvis (sacrum) and all the last five vertebrae. These cases display severe deformities and nerve disorders.

Source: 
http://www.highbeam.com/doc/1P1:28730117/f
congenital+absence+of+sacrum+and+lumbar+vertebrae.html?refid=ip_hf
4.12 Congenital spinal fusion (congenital spinal stenosis)

The congenital form of spinal stenosis is seen in individuals who are born with a narrow spinal canal. In these individuals, minimal changes in the structure of the spine can cause severe spinal stenosis. Spinal stenosis can cause a wide variety of symptoms throughout the body. The most common symptoms are generalized pain, weakness, and numbness in the affected region.

Source:
http://www.espineinstitute.com/Conditions_SpinalStenosis.htm#spinal
http://orthopedics.about.com/cs/spinalstenosis/a/spinalstenosis.htm

4.13 Cystic fibrosis

Cystic fibrosis (CF) is a life-threatening genetic disease affecting approximately 30,000 people in the United States. For people with the disease, a defective gene causes the body to produce a faulty protein that leads to abnormally thick, sticky mucus that clogs the lungs and can result in fatal lung infections. The mucus also obstructs the pancreas, causing difficulty for a person to absorb nutrients in food and can block the bile duct in the liver, eventually causing permanent damage in approximately six percent of people with CF.

In addition, more than 10 million Americans are genetic carriers. Carriers each have one copy of the defective CF gene, but do not have the disease and its symptoms. It takes two copies of the gene—one from each parent—for a child to be born with cystic fibrosis. Each time two carriers conceive, there is a 25 percent chance that their child will have CF; a 50 percent chance that the child will be a carrier of the CF gene but not have the disease; and a 25 percent chance that the child will not be a carrier and not have the disease.

CF occurs in approximately one of every 3,200 live Caucasian births (in one of every 3,500 live births of all Americans). About 1,000 new cases of CF are diagnosed each year. More than 80 percent of patients are diagnosed by age three; however, nearly 10 percent of newly diagnosed cases are age 18 or older.

The symptoms of CF vary from person to person due, in part, to the more than 1,000 mutations of the CF gene. Some of the most common symptoms can include: very salty-tasting skin; persistent coughing; frequent pneumonia, wheezing or shortness of breath; a failure to gain weight at the normal rate, perhaps with excessive appetite; and difficulty in having a bowel movement or frequent, abnormal bowel movements.

Adults with CF may experience health challenges such as CF-related diabetes and osteoporosis. CF also can cause reproductive problems—more than 95 percent of men with CF are sterile. But, with new technologies, some are becoming fathers. Although many women with CF are able to conceive, limited lung function and other health factors may make it difficult to carry a child to term.

Prompt, aggressive treatment of CF symptoms can extend the lives of those with the disease. The sweat test is the standard diagnostic test for CF. The CF Foundation recommends that the sweat test be performed at a CF Foundation-accredited care center, which has strict guidelines to ensure accurate results. This simple and painless procedure measures the amount of salt in the sweat. A high salt level indicates CF.

The ongoing treatment of CF depends upon the stage of the disease and the organs involved. Clearing mucus from the lungs is an important part of the daily CF treatment regimen. Chest physical therapy is one form of airway clearance, and it requires vigorous percussion (by using
(cupped hands) on the back and chest to dislodge the thick mucus from the lungs. Other forms of airway clearance can be done with the help of mechanical devices used to stimulate mucus clearance. Other types of treatments include: TOBI (tobramycin solution for inhalation), an aerosolized antibiotic used to treat lung infections; Pulmozyme, a mucus-thinning drug shown to reduce the number of lung infections and improve lung function; and azithromycin, an antibiotic proven to be effective in people with CF whose lungs are chronically infected with the common *Pseudomonas aeruginosa* bacteria.

When CF affects the pancreas, as it does in the majority of patients, the body does not absorb sufficient nutrients needed to grow and to thrive. Most people with CF eat a high-calorie diet and take replacement vitamins, and approximately 90 percent of people with CF take pancreatic enzyme replacements to help them absorb nutrients in digestion.

In 1955, children with CF usually did not live long enough to attend elementary school. Today, according to the CF Foundations National Patient Registry, the median age of survival for people with CF is in the late 30s and more than 40 percent of the CF population is age 18 and older. Many people with CF are living into their 30s, 40s, 50s and beyond. In the last four years alone, more than five years have been added to the median survival age of CF patients.

In 1955, there was no centralized care system for CF patients. Today, the CF Foundation accredits more than 115 care centers at major teaching and community hospitals, offering comprehensive, quality diagnosis and care nationwide — including 94 programs specifically for adults. Care center staff includes physicians, nurses, nutritionists, respiratory therapists, social workers, genetic counselors and other medical professionals.

In 1955, very little was understood about this disease. Today, the basic genetic defect that causes CF has been discovered, an arsenal of remedies have been developed to help treat it, and the CF Foundations “pipeline” is filled with more than 25 promising CF therapy candidates in clinical trials and six others in laboratory development.

Source:

http://www.cff.org/about_cystic_fibrosis/

5 Disease list and descriptions: D

5.1 Dejerine-Sottas syndrome

There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome, which is a severe degenerating neuropathy of the CMT1 type with onset by age 2 years. Features were nystagmus, distal muscular weakness, distal sensory change, pes cavus and exacerbations and remissions. The onset is usually with weakness and deformity of the feet and lower limbs. Autopsy showed the peripheral nerves to be increased in size, firm and gelatinous.

Source:


cmd=Retrieve&db=OMIM&dopt=Detailed&tmpl=dispomimTemplate&list_uids=145900

5.2 Depression

A depressive disorder is a syndrome that reflects a sad mood exceeding normal sadness or grief. Depression symptoms are characterized not only by negative thoughts, moods, and behaviors, but also by specific changes in bodily functions (e.g., eating, sleeping, and sexual activity). The
functional changes are often called neurovegetative signs. Certain people with depressive disorder, especially bipolar depression (manic depression), seem to have an inherited vulnerability to this condition. One in 10 people will have a depressive disorder in their lifetime, and in 1 of 10 cases, the depression is a fatal disease as a result of suicide.

Source:
http://www.medicinenet.com/depression/article.htm

5.3 Dermatomyositis-polymyositis

Inflammatory myopathies are acquired muscle diseases characterized by primary muscle weakness, endomysial inflammation, and elevated levels of serum muscle enzymes. Polymyositis and dermatomyositis, along with inclusion-body myositis, are the most common diseases of the striated muscle, skin, and surrounding connective tissue that clinicians observe. Each has unique clinical and histologic features. The pathology of both polymyositis and dermatomyositis have an underlying autoimmune basis, but the mechanisms for the 2 conditions differ. The incidence of polymyositis and dermatomyositis is 5-10 cases per 100,000 individuals. The active period of the disease is approximately 2-3 years in both children and adults. The duration is greater for patients with cardiac or pulmonary complications than for others; approximately 20% of the patients recover completely. The mortality rate after several years of the disease is approximately 15%; the rate is increased in patients with dermatomyositis with connective tissue diseases and malignancy. No racial predilection is observed. female preponderance has been reported in all age groups, with a female-to-male ratio of 2:1. Most patients present with polymyositis when aged 30-60 years, with a small peak in people aged 15 years. Dermatomyositis affects children and adults equally. The peak incidence is observed in individuals aged 45-64 years, with a small peak in children aged 5-14 years.

Source:
http://www.emedicine.com/neuro/topic85.htm

5.4 Diabetes mellitus

Better known just as “diabetes” — a chronic disease associated with abnormally high levels of the sugar glucose in the blood. The two main types of diabetes correspond to these two mechanisms and are called insulin dependent (type 1) and non-insulin dependent (type 2) diabetes. In type 1 diabetes, there is no insulin or not enough of it. In type 2 diabetes, there is generally enough insulin but the cells upon it should act are not normally sensitive to its action. The signs and symptoms of both types of diabetes include increased urine output and decreased appetite as well as fatigue. The major complications of diabetes include dangerously elevated blood sugar, abnormally low blood sugar due to diabetes medications, and disease of the blood vessels which can damage the eye, kidneys, nerves, and heart.

Source:

5.5 Diphtheria

An acute infectious disease that typically strikes the upper respiratory tract including the throat. It is caused by infection with the bacteria Corynebacterium diphtheriae. Symptoms include sore throat and mild fever at first. As the disease progresses, a membranous substance forms in the throat that makes it difficult to breathe and swallow. Diphtheria can be deadly. It is one of the diseases that the DTP (Diphtheria-Tetanus-Pertussis) and DTaP (Diphtheria-Tetanus-acellular Pertussis) vaccines are designed to prevent.
6 Disease list and descriptions: E

6.1 *E. coli* intestinal disease

*Escherichia coli* (*E. coli*) is a bacteria that normally lives in the intestines of people and animals. There are some kinds of *E. coli* that are capable of causing disease when within the colon. Enteroinvasive *E. coli* (EIEC) invades (passes into) the intestinal wall to produce severe diarrhea. Enterohemorrhagic *E. coli* (EHEC): A type of EHEC, *E. coli* 0157:H7, can cause bloody diarrhea and the hemolytic uremic syndrome (anemia and kidney failure). Enterotoxigenic *E. coli* (ETEC) produces a toxin that acts on the intestinal lining, and is the most common cause of travelers diarrhea. Enteropathogenic *E. coli* (EPEC) can causes diarrhea outbreaks in newborn nurseries. Enteroaggregative *E. coli* (EAggEC) can cause acute and chronic (long lasting) diarrhea in children.

Source:

http://www.medicinenet.com/enteroinvasive_e_coli/article.htm

6.2 Edward’s syndrome

Trisomy 18 syndrome. Children with the syndrome have an extra chromosome 18 with a characteristic pattern of multiple malformations and mental retardation. Features include low birth weight, small head (microcephaly), small jaw (micrognathia), malformations of the heart and kidneys, clenched fists with abnormal finger positioning, and malformed feet. The mental retardation is profound with an IQ too low to measure. Nineteen out of 20 of these children die before their first birthday. The condition is named after the British physician and geneticist John Edwards who discovered the extra chromosome in 1960.

Source:


6.3 Enzyme-deficiency (hemolytic anemia)

Hemolytic anemia is a condition of an inadequate number of circulating red blood cells (anemia) caused by premature destruction of red blood cells. Hemolytic anemia occurs when the bone marrow is unable to compensate for premature destruction of red blood cells by increasing their production. Causes of hemolytic anemia include infection, certain medications, autoimmune disorders, and inherited disorders. Types of hemolytic anemia include sickle-cell anemia, paroxysmal nocturnal hemoglobinuria, hemoglobin SC disease, hemolytic anemia due to G6PD deficiency, hereditary elliptocytosis, hereditary spherocytosis, hereditary ovalocytosis, idiopathic autoimmune hemolytic anemia, non-immune hemolytic anemia caused by chemical or physical agents, secondary immune hemolytic anemia, and thalassemia. Symptoms may include chills, fatigue, pale skin color, shortness of breath, rapid heart rate, yellow skin color, dark urine, and enlarged spleen.

Source:

6.4 Epilepsy (seizure disorder)

Epilepsy, a physical condition caused by sudden, brief changes in how the brain works, is estimated to affect one percent of the U.S. population, about 2.5 million people. In about half of all cases, no cause can be found, but head injuries, brain tumors, lead poisoning, problems in brain development before birth, and certain genetic and infectious illnesses can all cause epilepsy. Epilepsy occurs when nerve cells in the brain fire electrical impulses at a rate of up to four times higher than normal. This causes a sort of electrical storm in the brain, known as a seizure. A pattern of repeated seizure is referred to as epilepsy. Medication controls seizure for the majority of patients, who are otherwise healthy and able to live full and productive lives.

Source:
http://www.medicinenet.com/seizure/article.htm

6.5 Erythematousquamous dermatosis (seborrheic dermatitis)

Seborrheic dermatitis is a disease that causes flaking of the skin. It usually affects the scalp. In adolescents and adults, it is commonly called “dandruff.” In babies, it is known as “cradle cap.” Seborrheic dermatitis can also affect the skin on other parts of the body, such as the face and chest, and the creases of the arms, legs and groin. Seborrheic dermatitis usually causes the skin to look a little greasy and scaly or flaky. Seborrheic dermatitis has also been linked to neurologic disorders such as Parkinson’s disease and epilepsy. The reason for this relationship isn’t known.

Source:
http://familydoctor.org/157.xml

7 Disease list and descriptions: F

7.1 Food poisoning

A common flu-like illness typically characterized by nausea, vomiting and diarrhea, due to something the victim ate or drank that contained noxious bacteria, viruses, parasites, metals or toxins. The most prominent causes of food poisoning are Norwalk virus and Norwalk-like viruses, Campylobacter jejuni, Salmonella, Listeria monocytogenes, Vibrio vulnificus, and E. coli O157:H7.

Source:

7.2 Fragile X syndrome

One of the most common causes of inherited mental retardation and neuropsychiatric disease in human beings, affects as many as one in 2000 males and one in 4000 females. The syndrome is also known as FRAXA (the fragile X chromosome itself) and as the Martin-Bell syndrome. The characteristic features of the fragile X syndrome in boys include prominent or long ears, a long face, delayed speech, large testes (macroorchidism), hyperactivity, tactile defensiveness, gross motor delays, and autistic-like behaviors. Much less is known about girls with fragile X syndrome. Only about half of all females who carry the genetic mutation have symptoms themselves. Of those, half are of normal intelligence, and only one-fourth have an IQ under seventy. Few fragile X girls have autistic symptoms, although they tend to be shy and quiet. Fragile X syndrome is due to a dynamic mutation (a trinucleotide repeat) at an inherited fragile site on the X chromosome, and so is an X-linked disorder. Because the mutation is dynamic, it can change in length and
hence in severity from generation to generation, from person to person, and even within a given person. Approximately 15-25% of individuals with fragile X syndrome also are diagnosed with mild to moderate autism and autism spectrum disorders. Other clinical abnormalities associated with fragile X syndrome include attention deficit hyperactivity disorder (ADHD) and anxiety disorders. Variation in the cognitive and behavioral phenotype of the fragile X syndrome has been demonstrated in intellectual functioning, learning disability, executive function, attention, hyperactivity, depression, anxiety, and autistic behaviors. The explanation for this variation in phenotypic expression may depend on understanding the role of genetics and brain development in cognition and behavior.

Source:

7.3 Friedreich’s ataxia

Friedreich’s ataxia is one of the most common forms of autosomal recessive ataxia. The spinocerebellar tracts, dorsal columns, pyramidal tracts and, to a lesser extent, the cerebellum and medulla are involved. The disorder is usually manifest before adolescence and is generally characterized by incoordination of limb movements, dysarthria, nystagmus, diminished or absent tendon reflexes, Babinski sign, impairment of position and vibratory senses, scoliosis, pes cavus, and hammer toe. The triad of hypoactive knee and ankle jerks, signs of progressive cerebellar dysfunction, and preadolescent onset is commonly regarded as sufficient for diagnosis.

Source:

8 Disease list and descriptions: G

8.1 Giant cell arteritis

A serious disease characterized by inflammation of the walls of the blood vessels (vasculitis). The vessels affected by the inflammation are the arteries (hence the name “arteritis”). The age of affected patients is usually over 50 years of age. Giant cell arteritis can lead to blindness and/or stroke. It is detected by a biopsy of an artery. Giant cell arteritis is treated with high dose cortisone-related medications. Also called temporal arteritis or cranial arteritis.

Source:

8.2 Gram-negative folliculitis

Gram-negative folliculitis, first described by Fulton et al in 1968, is an infection caused by gram-negative organisms. The infection may occur as a complication in patients with acne vulgaris and rosacea and usually develops in patients who have received systemic antibiotics for prolonged periods. Gram-negative folliculitis may also occur in the setting of hot-tub immersion and in people infected with HIV. Gram-negative folliculitis is a relatively uncommon complication of prolonged antibiotic therapy. Gram-negative folliculitis has no associated increase in mortality. Morbidity is related to local pain and to the unwanted cosmetic effect of the folliculitis. Although gram-negative folliculitis is largely a complication of acne vulgaris and thus is expected to follow the age distribution of that entity, a slightly increased age at onset has been observed. The tendency for this disease to begin after the early teenage years is most likely because most patients who develop gram-negative folliculitis have undergone treatment of acne with a broad-spectrum antibacterial agent for a prolonged period.
8.3 Goiter

Goiter is an enlargement of the thyroid gland. It is not cancer. There are different kinds of goiters. A simple goiter usually occurs when the thyroid gland is not able to produce enough thyroid hormone to meet the body’s needs. A simple goiter may be classified as either an endemic (colloid) goiter or a sporadic (nontoxic) goiter. Endemic goiters occur within groups of people living in geographical areas with iodine-depleted soil, usually regions away from the sea coast. People in these communities might not get enough iodine in their diet (iodine is vital to the formation of thyroid hormone). The modern use of iodized table salt in the U.S. prevents this deficiency. However, inadequate iodine is still common in central Asia and central Africa. In most cases of sporadic goiter the cause is unknown. Hereditary factors may cause goiters. Risk factors for the development of a goiter include female sex, age over 40 years, inadequate dietary intake of iodine, residence in an endemic area, and a family history of goiter.

8.4 Good pastures syndrome

Goodpasture syndrome is an autoimmune disease of lung and kidney. Viral and streptococcal infections and exposure to hydrocarbon fumes have been suggested as possible causes. The host factor might be immune response genes.

8.5 Gout

Condition characterized by abnormally elevated levels of uric acid in the blood, recurring attacks of joint inflammation (arthritis), deposits of hard lumps of uric acid in and around the joints, and decreased kidney function and kidney stones. Uric acid is a breakdown product of purines, that are part of many foods we eat. The tendency to develop gout and elevated blood uric acid level (hyperuricemia) is often inherited and can be promoted by obesity, weight gain, alcohol intake, high blood pressure, abnormal kidney function, and drugs. The most reliable diagnostic test for gout is the identification of crystals in joints, body fluids and tissues.

9 Disease list and descriptions: H

9.1 Hemivertebra

A segmentation anomaly of the vertebral body due to failure of parts of the vertebra to develop and chondrify. The neural arch is often incomplete. Congenital scoliosis occurs as a result of the hemivertebra, and medial wedging of the hemivertebra occurs with time and weight-bearing.
9.2 *Helicobacter pylori* infection

*Helicobacter pylori* (*H. pylori*) is a bacterium that causes chronic inflammation of the inner lining of the stomach (gastritis) in humans. This bacterium also is the most common cause of ulcers worldwide. *H. pylori* infection is most likely acquired by ingesting contaminated food and water and through person to person contact. In the United States, 30% of the adult population is infected. (50% of infected persons are infected by the age of 60.) The infection is more common in crowded living conditions with poor sanitation. Infected individuals usually carry the infection indefinitely unless they are treated with medications to eradicate the bacterium. One out of every six patients with *H. pylori* infection will develop ulcers of the duodenum or stomach. *H. pylori* also is associated with stomach cancer and a rare type of lymphocytic tumor of the stomach called MALT lymphoma.

*Source:*

http://www.medicinenet.com/helicobacter_pylori/article.htm

9.3 Hepatitis A

Inflammation of the liver caused by the hepatitis A virus (HAV). HAV is usually transmitted from person to person by food or drink that has been contaminated with the stool of a person with hepatitis. In 2003 there were hepatitis A outbreaks in the US associated with eating raw or lightly cooked green onions (scallions). Hepatitis A develops within 2 months of exposure to the virus. The average incubation period for the virus is 28 days (range: 15-50 days) until symptoms appear. Hepatitis A can range greatly in severity. Older persons are more likely to have symptoms than children. In its most dire form, the disease can lead to liver failure and death. The symptoms and signs tend to appear abruptly and may include fatigue, loss of appetite, nausea, diarrhea, abdominal pain, fever, and jaundice (yellowing of the skin and eyes) and dark urine. Symptoms usually last less than 2 months. A few persons are ill for as long as 6 months. Recovery is usually complete.

*Source:*


9.4 Hepatitis B

Inflammation of the liver due to the hepatitis B virus (HBV), once thought to be passed only through blood products. It is now known that hepatitis B can also be transmitted via needle sticks, body piercing and tattooing using unsterilized instruments, the dialysis process, sexual and even less intimate close contact, and childbirth. Symptoms include fatigue, jaundice, nausea, vomiting, dark urine, light stools.

*Source:*


9.5 Hepatitis C

Inflammation of the liver due to the hepatitis C virus (HCV), which is usually spread by blood transfusion, hemodialysis, and needle sticks. HCV causes most transfusion-associated hepatitis, and the damage it does to the liver can lead to cirrhosis and cancer. At least half of HCV patients develop chronic hepatitis C infection. Chronic hepatitis C may be treated with interferon, sometimes in combination with anti-virals. There is no vaccine for hepatitis C.

*Source:*

9.6 Hepatitis D

Liver inflammation due to the hepatitis D virus (HDV), which only causes disease in patients who already have the hepatitis B virus. Transmission is via infected blood, needles, or sexual contact with an infected person. Symptoms are identical to those of hepatitis B. Chronic infection with HDV is currently treated with interferon, although it is not very successful. HDV infection can be prevented by the hepatitis B vaccine, and by avoiding activities that could lead to getting the virus.

Source:

9.7 Hepatitis E

A form of liver disease characterized by inflammation of the liver due to infection with the hepatitis E virus (HEV). Usually a mild disease, hepatitis E but can in rare cases prove fatal, particularly in pregnant women. HEV is transmitted via food or drink that has been handled by an infected person, or through infected water supplies in areas where fecal matter may get into the water. Consumption of uncooked deer meat is important risk factor for infection with HEV in Japan. Hepatitis E is rare in the US and Canada. It is more common in tropical and subtropical regions of the world.

Source:

9.8 Hereditary spastic paraplegia

Hereditary spastic paraplegia (HSP), also called familial spastic paraparesis (FSP), refers to a group of inherited disorders that are characterized by progressive weakness and stiffness of the legs. Though the primary feature of HSP is severe, progressive, lower extremity spasticity, in more complicated forms it can be accompanied by other neurological symptoms. These include optic neuropathy, retinopathy (diseases of the retina), dementia, ataxia (lack of muscle control), ichthyosis (a skin disorder resulting in dry, rough, scaly skin), mental retardation, peripheral neuropathy, and deafness.

Source:

9.9 HIV disease

HIV has also been called the human lymphotropic virus type III, the lymphadenopathy-associated virus and the lymphadenopathy virus. No matter what name is applied, it is a retrovirus. (A retrovirus has an RNA genome and a reverse transcriptase enzyme. Using the reverse transcriptase, the virus uses its RNA as a template for making complementary DNA which can integrate into the DNA of the host organism). Although the American research Robert Gallo at the National Institute of Health (NIH) believed he was the first to find HIV, it is now generally accepted that the French physician Luc Montagnier (1932–) and his team at the Pasteur Institute discovered HIV in 1983–1984.

Source:
9.10 **Hodgkin’s disease**

*Hodgkin’s disease*, sometimes called *Hodgkin’s lymphoma*, is a cancer that starts in lymphatic tissue. Lymphatic tissue includes the lymph nodes and related organs that are part of the body’s immune and blood-forming systems. The lymph nodes are small, bean-shaped organs found underneath the skin in the neck, underarm, and groin. They are also found in many other places in the body such as inside the chest, abdomen, and pelvis.

Because lymphatic tissue is present in many parts of the body, Hodgkin’s disease can start almost anywhere, but most often starts in lymph nodes in the upper part of the body. The most common sites are in the chest, neck, or under the arms. Hodgkin’s disease enlarges the lymphatic tissue, which can then cause pressure on important structures. It can spread through the lymphatic vessels to other lymph nodes. This is the major way it spreads. Most Hodgkin’s disease spreads to nearby lymph node sites in the body, not distant ones. It rarely gets into the blood vessels and can spread to almost any other site in the body, including the liver and lungs.

*Source:*
http://www.cancer.org/docroot/cri/content/cri_2_4_1x_what_is_hodgkins_disease_20.asp?sitearea=cri

9.11 **Hyperosmolality (hypernatremia)**

Hypernatremia is a medical condition in which there is excess sodium, urea, and other electrolytes in the body relative to the amount of water (electrolyte disturbance).

*Source:*
http://en.wikipedia.org/wiki/Hypernatremia

9.12 **Hypersensitivity angiitis**

Hypersensitivity angiitis is a disease in which patients present with palpable purpura dominant on the lower legs. As lesions evolve they become confluent, and sometimes hemorrhagic and ulcerate. Other organ systems may be involved, particularly the joints, gastrointestinal tract, and kidneys. Current evidence supports an immune complex pathogenesis in which a variety of antigens including bacteria, viruses, drugs, or chemicals are involved.

*Source:*

9.13 **Hypoglycemia**

Low blood sugar (glucose). The symptoms may include anxiety, sweating, tremor, palpitations, nausea, and pallor. Hypoglycemia also starves the brain of glucose energy, which is essential for proper brain function. Lack of glucose energy to the brain can cause symptoms ranging from headache, mild confusion, and abnormal behavior, to loss of consciousness, seizure, and coma. Severe hypoglycemia can cause death. The causes of hypoglycemia include drugs (such as insulin), liver disease, surgical absence of the stomach, tumors that release excess amounts of insulin, and pre-diabetes. In some patients, symptoms of hypoglycemia occur during fasting (fasting hypoglycemia). In others, symptoms of hypoglycemia occur after meals (reactive hypoglycemia). Immediate treatment of severe hypoglycemia consists of administering large amounts of glucose, and repeating this treatment at intervals if the symptoms persist.

*Source:*
9.14 Hypoosmolality (hyponatremia)

The electrolyte disturbance hyponatremia or hyponatraemia exists in humans when the sodium level in the plasma falls below 135 mmol/l. At lower levels water intoxication may result, an urgently dangerous condition. Hyponatremia is an abnormality that can occur in isolation or, as most often is the case, as a complication of other medical illnesses. Severe hyponatremia may cause osmotic shift of water from the plasma into the brain cells. Typical symptoms include nausea, vomiting, headache and malaise. As the hyponatremia worsens, confusion, stupor or coma may occur.

Source:
http://en.wikipedia.org/wiki/Hyponatremia

9.15 Hypertrophic obstructive cardiomyopathy

Cardiomyopathy is a condition in which the muscle of the heart is abnormal in the absence of an apparent cause. This terminology is purely descriptive and is based on the Latin derivation. There are four types of cardiomyopathy: Hypertrophic (HCM), Dilated (DCM), Restrictive (RCM) and Arrhythmogenic Right Ventricular (ARVC). The main feature of Hypertrophic Cardiomyopathy is an excessive thickening of the heart muscle (hypertrophy literally means to thicken). Heart muscle may thicken in normal individuals as a result of high blood pressure or prolonged athletic training. In Hypertrophic Cardiomyopathy, however, the muscle thickening occurs without an obvious cause. In addition, microscopic examination of the heart muscle in Hypertrophic Cardiomyopathy shows that it is abnormal.

Source:
http://www.cardiomyopathy.org/html/which_card_hcm.htm

10 Disease list and descriptions: I

10.1 Ichthyosis congenita

Ichthyosis group marked by great variability, most of the conditions being transmitted as autosomal recessive traits. The affected children present with an armour-like thickening of the skin, deep fissures and ectropion of the eyelids and the oral mucosa. On a clinical and histological basis, bullous (BULLOUS ICHTHYOTIC ERYTHRODERMA OF BROCC; ICHTHYOSIS BULLOSA OF SIEMENS) and non-bullous forms (HARLEQUIN ICHTHYOSIS; LAMELLAR ICHTHYOSES) can be distinguished.

Source:

Harlequin-type ichthyosis (also harlequin ichthyosis, ichthyosis congenita, or keratosis diffusa fetalis), the most severe form of congenital ichthyosis, is characterized by a thickening of the keratin layer in fetal human skin, appearing as massive, diamond-shaped scales. In addition, the eyes, ears, mouth, and other appendages may be abnormally contracted. The scaly keratin limits the child’s movement, and because it is cracked where normal skin would fold, it is easily pregnable by bacteria and other contaminants, resulting in a serious risk of fatal infection.

Sufferers are known as harlequin fetuses, harlequin babies, or harlequins.

The harlequin-type designation comes from both the baby’s apparent facial expression and the diamond-shape of the scales (resembling the costume of Arlecchino), which are caused by
severe hyperkeratosis. Seventeenth century entertainers known as jesters, or harlequins, wore
costumes with diamond patterns on them, as well as a particular style of face paint. The disease
can be diagnosed in the womb by way of fetal skin biopsy.

Source:

11 Disease list and descriptions: K

11.1 Kawasaki disease

A disease that has nothing to do with the motor bike of the same name but is a syndrome of
unknown origin that mainly affects young children, causing fever, reddening of the eyes (conjunc-
tivitis), lips and mucous membranes of the mouth, ulcerative gum disease (gingivitis), swollen
glands in the neck (cervical lymphadenopathy) and a rash that is raised and bright red (macu-
loerythematos) in a glove-and-sock fashion over the skin of the hands and feet which becomes
hard, swollen (edematous) and peels off. Also called the mucocutaneous lymph node syndrome.
Children with Kawasaki disease who are not treated within the first week to 10 days of the onset
of fever have five times the risk of developing coronary artery aneurysms. The syndrome was first
described in the late 1960's in Japan by the pediatrician Tomisaku Kawasaki. Kawasaki disease
affects the vascular system, and is now the main cause of acquired heart disease in children. It is
most common in people of Asian descent, and is both more common and more deadly in males.

Source:

11.2 Klippel-Feil syndrome

Short neck, low hairline at the nape of the neck and limited movement of the head. Klippel-
Feil sequence is due to a defect in the early development of the spinal column in the neck, resulting
in fusion of the cervical vertebrae. The condition is named for the French neurologists Maurice
Klippel and Andre Feil who in 1912 reported “A case of absence of the cervical vertebrae with
the thoracic cage going up to the base of the skull.”

Source:

12 Disease list and descriptions: L

12.1 Leprosy (Hansen disease)

Leprosy, a chronic granulomatous infection caused by a bacterium which affects various parts
of the body, including in particular the skin and nerves. (Granulomatous refers to the formation
of granulomas, inflammatory nodules that are usually small, granular, firm, and persistent.) The
bacterium responsible for leprosy is called Mycobacterium leprae or, for short, M. leprae. For
thousands of years, leprosy was one of the world’s most feared communicable diseases, because
the skin and nerve damage often led to terrible disfigurement and disability. India accounts
for almost four-fifths (nearly 80%) of all cases of leprosy in the world. Today leprosy can be
cured, particularly if treatment is begun early. The term Hansen disease instead of leprosy is
now preferred by some experts, because of it being less perjorative. Hansen disease was named
in honor of the Norwegian physician, Gerhard Armauer Henrik Hansen, who in 1873 discovered
the bacillus Mycobacterium leprae, the first microbe found to be the causative agent of a human
disease.
12.2 Lethal midline granuloma

Progressively destructive condition of uncertain aetiology, involving the nose and paranasal si-

nuses. It leads to erosion of the sinonasal and adjacent structures, when left untreated eventually
causing death by extension to the central nervous system, infection or haemorrhage. Radiolog-
ically, midfacial bone destruction with relatively little associated soft tissue thickening is seen.
This condition resembles the findings in Wegeners granulomatosis head and neck manifesta-
tion, although in the latter the changes are less pronounced.

Source:
http://www.medcyclopaedia.com/library/topics/volume_vi_2/l/
lethal_midline_granuloma.aspx?s=Lethal%20midline%20granuloma&scope=&mode=1

12.3 Leukodystrophy

A disorder of the white matter of the brain, the part of the brain that contains myelinated
nerve fibers. The white matter is white because it is the color of myelin, the insulation covering
the nerve fibers. The white matter is involved in the conduction of nerve impulses in the brain.

Source:

12.4 Lipid metabolism (Hypercholesterolemia, Hypertriglyceridemia, Hyper-

lipoproteinemia, Combined hyperlipidemia, Abetalipoproteinemia, Lipody-

trophy, Gaucher’s disease, Niemann-Pick disease, Carnitine-acylcarnitine

translocase deficiency, Mitochondrial trifunctional protein deficiency)

12.4.1 Hypercholesterolemia

Hypercholesterolemia (literally: high blood cholesterol) is the presence of high levels of choles-
terol in the blood. It is not a disease but a metabolic derangement that can be secondary to many
diseases and can contribute to many forms of disease, most notably cardiovascular disease. It is
closely related to the terms ”hyperlipidemia” (elevated levels of lipids) and ”hyperlipoproteinemia”
(elevated levels of lipoproteins). Elevated cholesterol does not lead to specific symptoms
unless it has been longstanding. Some types of hypercholesterolemia lead to specific physical
findings: xanthoma (thickening of tendons due to accumulation of cholesterol), xanthelasma
palpabrum(yellowish patches around the eyelids) and arcus senilis (white discoloration of the
peripheral cornea). Longstanding elevated hypercholesterolemia leads to accelerated atheroscle-
rosis; this can express itself in a number of cardiovascular diseases.

Source:
http://en.wikipedia.org/wiki/Hypercholesterolemia
12.4.2 Hypertriglyceridemia

In medicine, hypertriglyceridemia denotes high (hyper-) blood levels (-emia) of triglycerides, the most abundant fatty molecule in most organisms. It has been associated with atherosclerosis, even in the absence of hypercholesterolemia (high cholesterol levels). It can also lead to pancreatitis in excessive concentrations. Very high triglyceride levels may also interfere with blood tests; hyponatremia may be reported spuriously (pseudohyponatremia). A related term is "Hyperglyceridemia" or "Hyperglyceridaemia", which refers to a high level of all glycerides, including monoglycerides, diglycerides and triglycerides.

Source:
http://en.wikipedia.org/wiki/Hyperglyceridemia

12.4.3 Hyperlipoproteinemia

Hyperlipoproteinemia is the presence of elevated levels of lipoprotein in the blood. Lipids are transported in a protein capsule, and the density of the lipids and type of protein determines the fate of the particle and its influence on metabolism. Although the terms hyperlipoproteinemia and hypercholesterolemia are often used interchangeably, the former is more specific.

Source:
http://en.wikipedia.org/wiki/Hyperlipoproteinemia

12.4.4 Combined hyperlipidemia

In medicine, combined hyperlipidemia (or -aemia) is a commonly occurring form of hypercholesterolemia (elevated cholesterol levels) characterised by increased LDL and triglyceride concentrations, often accompanied by decreased HDL. The elevated triglyceride levels (>5mmol/l) are generally due to an increase in VLDL (very low density lipoprotein), a class of lipoprotein that is prone to cause atherosclerosis.

Source:
http://en.wikipedia.org/wiki/Combined_hyperlipidemia

12.4.5 Abetalipoproteinemia

Abetalipoproteinemia is a rare genetic disorder that interferes with the normal absorption of fat and fat soluble vitamins from food. This disorder leads to a multiple vitamin deficiency, affecting the fat soluble vitamins A, D, E, and K. However, many of the observed effects are due to vitamin E deficiency in particular.

Source:
http://en.wikipedia.org/wiki/Abetalipoproteinemia

12.4.6 Lipodystrophy

In medicine, lipodystrophy is a condition characterized by abnormal or degenerative conditions of the body’s adipose tissue. A more specific term, lipoatrophy is used when describing the loss of fat from one area (usually the face). A lipodystrophy can be a lump or small dent in the skin that forms when a person keeps performing injections in the same spot. These types of lipodystrophies are harmless. Lipodystrophies can be a possible side effect of HIV medication (mainly the protease inhibitors). Other lipodystrophies manifest as the excess or lack of fat in various regions of the body. These include but are not limited to having sunken cheeks, “humps” on the back or back of the neck. They are often seen as a symptom of AIDS or as side-effects from antiretroviral drugs. Lipodystrophy can be caused by metabolic abnormalities due to genetic issues.
12.4.7 Gaucher’s disease

Gaucher’s disease is the most common of the lipid storage diseases. It is caused by a deficiency of the enzyme glucocerebrosidase, leading to an accumulation of its substrate, the fatty substance glucocerebroside. Fatty material can collect in the spleen, liver, kidneys, lungs, brain and bone marrow. Symptoms may include enlarged spleen and liver, liver malfunction, skeletal disorders and bone lesions that may cause pain, severe neurologic complications, swelling of lymph nodes and (occasionally) adjacent joints, distended abdomen, a brownish tint to the skin, anemia, low blood platelets and yellow fatty deposits on the sclera. Persons affected most seriously may also be more susceptible to infection. The disease affects males and females equally. It is the most common lysosomal storage disease. It is named after the French doctor who originally described it in 1882.

12.4.8 Niemann-Pick disease

Niemann-Pick disease is an inherited condition involving lipid metabolism in which harmful amounts of lipids accumulate in the spleen, liver, lungs, bone marrow, and brain. There are four variants of Niemann-Pick disease based on the genetic cause and the symptoms exhibited by the patient. All variants are inherited in an autosomal recessive pattern. Mutations in NPC1, NPC2, and SMPD1 genes cause Niemann-Pick disease.

12.4.9 Carnitine-acylcarnitine translocase deficiency

Carnitine-acylcarnitine translocase deficiency is a rare condition that prevents the body from converting long-chain fatty acids into energy, particularly during periods without food. Carnitine, a natural substance acquired mostly through the diet, is used by cells to process fats and produce energy. People with this disorder have a faulty enzyme that prevents long-chain fatty acids from being transported into the innermost part of the mitochondria for processing. The signs of carnitine-acylcarnitine translocase deficiency usually begin within the first few hours of life. Seizures, an irregular heartbeat, and breathing problems are often the first signs of this disorder. This disorder may also cause extremely low levels of ketones (products of fat breakdown that are used for energy) and low blood sugar (hypoglycemia). Together, these two signs are called hypoketotic hypoglycemia. Other signs that are often present include ammonia in the blood (hyperammonemia), an enlarged liver (hepatomegaly), heart abnormalities (cardiomyopathy), and muscle weakness. This disorder can cause sudden infant death.

Sources:

http://en.wikipedia.org/wiki/Lipodystrophy
http://en.wikipedia.org/wiki/Gaucher%27s_disease
http://en.wikipedia.org/wiki/Niemann-Pick\_disease
http://en.wikipedia.org/wiki/Carnitine-acylcarnitine\_translocase\_deficiency
12.4.10 Mitochondrial trifunctional protein deficiency

Mitochondrial trifunctional protein deficiency is a rare inherited condition that prevents the body from converting certain fats to energy, particularly during periods without food. People with this disorder have inadequate levels of an enzyme that breaks down a certain group of fats called long-chain fatty acids. Onset of this disorder may begin during infancy or later in life. Signs and symptoms of the early onset form can include feeding difficulties, lack of energy (lethargy), low blood sugar (hypoglycemia), muscle weakness (hypotonia), liver problems, and a high risk for complications such as life-threatening heart and breathing problems, coma, and sudden unexpected death. The late-onset form is usually less severe; signs and symptoms can include hypotonia, muscle pain, a breakdown of muscle tissue, and abnormalities in the nervous system that affect arms and legs (peripheral neuropathy). Episodes of mitochondrial trifunctional protein deficiency can be triggered by periods of fasting or by illnesses such as viral infections.

Source:
http://en.wikipedia.org/wiki/Mitochondrial\_trifunctional\_protein\_deficiency

12.5 Lown-Ganong-Levine syndrome

Lown–Ganong–Levine syndrome is the acceleration of the conduction of the cardiac impulse with a normal QRS complex. In this condition, there are short PR intervals and atrial arrhythmias, no delta waves. This occurs in the presence of a fast-conducting pathway within the atroventricular node which causes a short PR interval but permits normal ventricular depolarisation. The patient may give a history of palpitations.

Source:
http://www.gpnotebook.co.uk/cache/630849546.htm

12.6 Lumbosacral spondylolysis

Lumbar spondylolysis is a unilateral or bilateral defect of the pars interarticularis affecting one or more of the lumbar vertebrae. The term is derived from the Greek words spondylos, meaning vertebra, and lysis, meaning break or defect. In the United States, a reported difference exists between the sexes and races, with an incidence of spondylolysis of 6.4% in white men, 2.8% in black men, 2.3% in white women, and 1.1% in black women. Pars defect is twice as common in boys than in girls, although high-grade slippage is 4 times more common in girls than in boys. Alaskan Eskimos (26%) have the highest incidence, with the highest rate in Eskimos from north of the Yukon River (Lonstein, 1999).

Source:
http://www.emedicine.com/sports/topic71.htm

12.7 Lymphosarcoma (lymphoma)

Lymphoma is any of a variety of cancer that begins in the lymphatic system. In technical terms, lymphoma denotes malignancies of lymphocytes or, more rarely, of histiocytes. Just as there are many types of lymphocytes, so there are many types of lymphoma. Lymphomas are part of the broad group of diseases called hematological neoplasms. Traditionally, Lymphoma is classified as Hodgkins lymphoma, discovered by Thomas Hodgkin in 1832, and non-Hodgkins lymphoma (all other types of lymphoma). According to the U.S. National Institutes of Health, lymphomas account for about five percent of all cases of cancer in the United States, and Hodgkins disease in particular accounts for less than one percent of all cases of cancer in the United States. Because the lymphatic system is part of the bodys immune system, patients with weakened immune system, such as from HIV infection or from certain drugs or medication, also have a higher incidence of lymphoma.
13 Disease list and descriptions: M

13.1 Migraine

Usually, periodic attacks of headaches on one or both sides of the head. These may be accom-
panied by nausea, vomiting, increased sensitivity of the eyes to light (photophobia), increased
sensitivity to sound (phonophobia), dizziness, blurred vision, cognitive disturbances, and other
symptoms. Some migraines do not include headache, and migraines may or may not be preceded
by an aura.

Source:

13.2 Meningococcal infection

Infection with the bacterium Neisseria meningitidis. Persons with a meningococcal infec-
tion may have any of several acute illnesses, including: meningitis and sepsis (blood infection),
meningitis in the absence of sepsis, sepsis in the absence of meningitis, a predominantly cuta-
neous (skin) condition called purpura fulminans, or some combination of these conditions. Other
illnesses that can occur from meningococcal infection in the absence of sepsis include pharyngitis
(throat infection), conjunctivitis (eye infection), pneumonia, and arthritis.

Source:

13.3 Mineral metabolism (Fe) (Haemochromatosis)

Haemochromatosis, also spelled hemochromatosis, is a hereditary disease characterized by
improper processing by the body of dietary iron which causes iron to accumulate in a number of
body tissues, eventually causing organ dysfunction. It is the main iron overload disorder. Males
are usually diagnosed after their forties, and women about a decade later, owing to regular iron
loss by menstruation, but cases have been found in young children as well. Hemochromatosis is
one of the most common inheritable genetic defects, especially in people of northern European
extraction, with about 1 in 10 people carrying the defective gene. The prevalence of haemochro-
matosis varies in different populations.

Source:
http://en.wikipedia.org/wiki/Hemochromatosis

13.4 Mineral metabolism (Cu) (Wilson's disease, Menkes' disease)

13.4.1 Wilson's disease

Wilson's disease or lentigohepatic degeneration is an autosomal recessive hereditary disease,
with an incidence of about 1 in 30,000 in most parts of the world. However it is much more
common in Central America, especially in El Salvador. 1 in 186 Salvadorians are born with
the disease. Its main feature is accumulation of copper in tissues, which manifests itself with
neurological symptoms and liver disease. The estimated heterozygous carrier rate is about 1 in
9,000, meaning that 1 in 9,000 people are unaffected carriers of this mutation. The disease affects
men more than women and occurs mostly in hispanic races. Symptoms usually appear around
the ages of 18 to 21 years, but sometimes not until the age of 30, and in rare instances up to age 50. The most classical sign are the Kayser-Fleischer rings (brown rings around the cornea in the eye) that result from copper deposition in Descemet’s membrane of the cornea.

Source:
http://en.wikipedia.org/wiki/Wilson%27s_disease

13.4.2 Menkes' disease

Menkes Disease is caused by a defective gene that regulates the metabolism of copper in the body. Because it is an X-linked gene, the disease primarily affects male infants. Copper accumulates at abnormally low levels in the liver and brain, but at higher than normal levels in the kidney and intestinal lining. Affected infants may be born prematurely. Symptoms appear during infancy. Normal or slightly slowed development may proceed for 2 to 3 months, and then there will be severe developmental delay and a loss of early developmental skills. Menkes Disease is also characterized by seizures, failure to thrive, subnormal body temperature, and strikingly peculiar hair, which is kinky, colorless or steel-colored, and easily broken. There can be extensive neurodegeneration in the gray matter of the brain. Arteries in the brain can also be twisted with frayed and split inner walls. This can lead to rupture or blockage of the arteries. Weakened bones (osteoporosis) may result in fractures.

Source:
http://www.ninds.nih.gov/disorders/menkes/menkes.htm

13.5 Mineral metabolism (Mg) (Hypermagnesemia, Hypomagnesemia)

13.5.1 Hypermagnesemia

Hypermagnesemia is an electrolyte disturbance in which there is an abnormally elevated level of magnesium in the blood. Usually this results in excess of magnesium in the body. Hypermagnesemia occurs rarely because the kidney is very effective in excreting excess magnesium. It usually develops only in people with kidney failure who are given magnesium salts or who take drugs that contain magnesium. Symptoms may include weakness, nausea and vomiting, hypotension, impaired breathing, and arrhythmia and asystole, which are most prominent cardiac symptoms due to conduction delays, since magnesium acts as physiologic calcium blocker.

Source:
http://en.wikipedia.org/wiki/Hypermagnesemia

13.5.2 Hypomagnesemia

Hypomagnesemia is an electrolyte disturbance in which there is an abnormally low level of magnesium. Usually a serum level less than 0.6 mmol/l is used as reference. It must be noted that hypomagnesemia is not equal to magnesium deficiency. Deficiency of magnesium causes among others cardiac arrhythmia and increased irritability of the nervous system with tetany. It may result from a number of conditions including inadequate intake of magnesium, chronic diarrhea, malabsorption, alcoholism, diuretic use and other disorders.

Source:
http://en.wikipedia.org/wiki/Hypomagnesemia
13.6 Mineral metabolism (Ca) (Hypocalcaemia, Hypercalcaemia, Pseudohypoparathyroidism)

13.6.1 Hypocalcaemia

In medicine, hypocalcaemia is the presence of low serum calcium levels in the blood, usually taken as less than 2.2 mmol/L or 9mg/dl or an ionized calcium level of less than 1.1 mmol/L (4.5 mg/dL). It is a type of electrolyte disturbance. It mainly occurs due to a deficient parathormone, ineffective parathormone or deficiency of Vitamin D. It may be seen alongside hypomagnesemia.

Source:
http://en.wikipedia.org/wiki/Hypocalcemia

13.6.2 Hypercalcaemia

Hypercalcaemia (or Hypercalcemia) is an elevated calcium level in the blood. It can be an asymptomatic laboratory finding, but because an elevated calcium level is often a clue to other serious disease, a diagnosis should be undertaken if it persists. It can be due to excessive skeletal calcium release, increased intestinal calcium absorption, or decreased renal calcium excretion. Hypercalcemia per se can result in fatigue, depression, confusion, anorexia, nausea, vomiting, constipation, or increased urination; if it is chronic it can result in urinary calculi (renal stones or bladder stones). Abnormal heart rhythms can result, and an EKG finding of a short QT interval suggests hypercalcemia. Symptoms are more common at high calcium levels (12.0 mg/dL or 3 mmol/l). Severe hypercalcemia (above 15-16 mg/dL or 3.75-4 mmol/l) is considered a medical emergency: at these levels, coma and cardiac arrest can result.

Source:
http://en.wikipedia.org/wiki/Hypercalcemia

13.6.3 Pseudohypoparathyroidism

Pseudohypoparathyroidism is a condition that mimics hypoparathyroidism, but is due to a resistance to parathyroid hormone, rather than a lack of the hormone (akin to the distinction between Type1 diabetes and Type2 diabetes.) Hyperparathyroidism is overactivity of the parathyroid glands resulting in excess production of parathyroid hormone (PTH). Increased PTH consequently leads to increased serum calcium (hypercalcemia) due to 1) increased bone resorption, allowing flow of calcium from bone to blood, 2) reduces renal clearance of calcium, and 3) increases intestinal calcium absorption.

Source:
http://en.wikipedia.org/wiki/Pseudohypoparathyroidism
http://en.wikipedia.org/wiki/Hyperparathyroidism

13.7 Mitochondrial disease

Mutations in the mitochondrial chromosome are responsible for a number of disorders including, for example: an eye disease called Leber’s hereditary optic atrophy; a type of epilepsy called MERRF which stands for Myoclonus Epilepsy with Ragged Red Fibers; and a form of dementia called MELAS for Mitochondrial Encephalopathy, Lactic Acidosis and Stroke-like episodes. All mitochondrial diseases were entirely enigmatic before it was discovered that they were due to mutations not in regular chromosomes but in the chromosome of mitochondria.

Source:
13.8 **Moyamoya**

A pattern of progressive obstructive and occlusive cerebral arteritis (inflammation of the cerebral arteries that obstructs and occludes them), predominantly in children and young adults. Symptoms include headaches, behavioral abnormalities, and recurrent attacks of paralysis.

*Source:*

13.9 **Multiple epiphyseal dysplasia**

Multiple epiphyseal dysplasia (MED) is a rare inherited spectrum of disorders characterized by malformation (dysplasia) of the “growing portion” or head of the long bones (epiphyses). Affected individuals may have an abnormally short thighbone (femur), unusually short hands and fingers, mind short stature, a waddling gait, and/or pain in the hips and knees. In some cases, painful swelling and inflammation of certain joints (arthritis) may be present as early as five years of age. Most cases of multiple epiphyseal dysplasia are inherited as autosomal dominant traits; rare cases are inherited as autosomal recessive traits.

*Source:*
http://www.webmd.com/hw/raising_a_family/nord359.asp

13.10 **Multiple sclerosis**

A disease of the central nervous system (CNS) marked by numbness, weakness, loss of muscle coordination, and problems with vision, speech, and bladder control. MS is an autoimmune disease in which the body’s immune system attacks myelin, a key substance that serves as a nerve insulator and helps in the transmission of nerve signals. The progress, severity and specific symptoms in MS are unpredictable. Most people with MS are between the ages of 20 and 40 at the time of diagnosis.

*Source:*

13.11 **Mumps**

An acute viral illness that usually presents with inflammation of the salivary glands, particularly the parotid glands. A child with mumps often looks like a chipmunk with a full mouth due to the swelling of the parotids. Mumps can also cause inflammation of other tissues, most frequently the covering and substance of the central nervous system (meningoencephalitis), the pancreas (pancreatitis) and, after adolescence, the ovary (oophoritis) and the testis (orchitis). The testis is particularly susceptible to damage from mumps; the damage can lead to infertility. Together with the likes of measles and chickenpox, mumps was once considered one of the inevitable infectious diseases of childhood. Since a mumps vaccine became available in 1967, the incidence of mumps has declined in the U.S., but there are still many underimmunized populations.

*Source:*
13.12 Muscular dystrophy

One of a group of genetic diseases characterized by progressive weakness and degeneration of the skeletal or voluntary muscles which control movement. The muscles of the heart and some other involuntary muscles are also affected in some forms of muscular dystrophy, and a few forms involve other organs as well. Muscular dystrophy can affect people of all ages. Although some forms first become apparent in infancy or childhood, others may not appear until middle age or later. Duchenne muscular dystrophy is the most common kind of muscular dystrophy affecting children. Myotonic dystrophy is the most common of these diseases in adults. There is no specific treatment for any of the forms of muscular dystrophy. The prognosis with muscular dystrophy varies according to the type of muscular dystrophy and the progression of the disorder. Some cases may be mild and very slowly progressive with normal lifespan, while other cases may have more marked progression of muscle weakness, functional disability and loss of ambulation. Life expectancy depends on the degree of progression and late respiratory deficit. In Duchenne muscular dystrophy, death usually occurs in the late teens to early 20s.

Source:

13.13 Mycoplasma

*Mycoplasma hominis* and *Mycoplasma pneumoniae* are among the dozen types of mycoplasma that occur in humans. *Mycoplasma hominis* is a common inhabitant of the vagina and can cause infections of the female and male genital tracts. *Mycoplasma pneumoniae* can infect the upper respiratory tract and the lungs. It is a major cause of respiratory infection in children of school age and young adults. It is also a common cause of pneumonia in persons with HIV.

Source:

13.14 Myotonic disorders (myotonia)

Myotonia is a neuromuscular disorder charaterized by the slow relaxation of the muscles after voluntary contraction or electrical stimulation. Generally, repeated effort is needed to relax the muscles, and the condition improves after the muscles have warmed-up. Individuals with the disorder may have trouble releasing their grip on objects or may have difficulty rising from a sitting position and a stiff, awkward gait. The disorder can affect all muscle groups. It may be acquired or inherited, and is caused by an abnormality in the muscle membrane. Myotonia is a symptom commonly seen in patients with myotonic muscular dystrophy and in a group of disorders called channelopathies (hereditary diseases that are caused by mutations in the chloride, sodium or potassium ion transport channels in the muscle membrane).

Source:
http://en.wikipedia.org/wiki/Myotonia

14 Disease list and descriptions: N

14.1 Neurofibromatosis

A genetic disorder of the nervous system that primarily affects the development and growth of neural (nerve) cell tissues, causes tumors to grow on nerves, and may produce other abnormalities. Neurofibromatosis (NF) consists of two very different disorders: Neurofibromatosis type1(NF1) and Neurofibromatosis type2(NF2).
14.2 Neuromyelitis optica (NMO, Devic’s disease)

Neuromyelitis optica is an inflammatory disease of the central nervous system in which there are episodes of inflammation and damage to myelin (fatty, protective covering of nerves) that almost exclusively affect the optic (eye) nerves and spinal cord. It usually causes temporary blindness, occasionally permanent, in one of both eyes. It can also lead to varying degrees of weakness or paralysis in the legs or arms, loss of sensation, and/or bladder and bowel dysfunction from spinal cord damage.

Source:
http://mayoclinic.org/devics-disease/index.html

15 Disease list and descriptions: O

15.1 Ornithosis (psittacosis)

Ornithosis is an illness characterized by fever, chills, headache, photophobia (the avoidance of light), cough, and muscle aches. It is caused by an infection with bacteria known as *Chlamydia psittaci*. Humans become infected with psittacosis when they inhale *C. psittaci* bacteria that are present in dried bird droppings, feather dust or other secretions of infected birds. Person-to-person spread of psittacosis is very unlikely. The symptoms include fever, chills, headache, rash, photophobia, muscle aches and either upper or lower respiratory tract disease. Pneumonia is common with psittacosis.

Source:
http://health.utah.gov/epi/fact_sheets/psittac.html

15.2 Osteogenesis imperfecta

Brittle bone disease. Osteogenesis imperfecta (OI) is not one but a group of genetic diseases, all of which affect collagen in connective tissue in the body and all of which result in fragile bones.

Source:

15.3 Osteopetrosis

Thickening of the bones which become abnormally dense due an inherited defect in bone resorption — the process in which old bone is broken down and removed so that new bone can be added to the skeleton. Osteoclasts are the cells responsible for bone resorption. In osteopetrosis, the osteoclasts do not perform normally. This flaw in bone resorption results in bones that are abnormally dense, yet are fragile and easily broken. Men and women are equally affected by the disease. Anemia, infection, and bleeding are just some of the symptoms that individuals with osteopetrosis can experience — blindness, deafness and even stroke can occur when the skeleton is so dense that blood vessels and nerves cannot pass through the bones. Osteopetrosis is also known as marble bone disease and Albers-Schonberg disease.

Source:
16 Disease list and descriptions: P

16.1 Parkinson’s disease

Parkinson’s disease is a chronic, progressive neurodegenerative movement disorder. Tremors, rigidity, slow movement (bradykinesia), poor balance, and difficulty walking (called parkinsonian gait) are characteristic primary symptoms of Parkinson’s disease.

Idiopathic Parkinson’s disease is the most common form of parkinsonism, a group of movement disorders that have similar features and symptoms. Parkinson’s disease is called idiopathic Parkinson’s because the cause is unknown. In the other forms of parkinsonism, a cause is known or suspected.

Parkinson’s results from the degeneration of dopamine-producing nerve cells in the brain, specifically in the substantia nigra and the locus coeruleus. Dopamine is a neurotransmitter that stimulates motor neurons, those nerve cells that control the muscles. When dopamine production is depleted, the motor system nerves are unable to control movement and coordination. Parkinson’s disease patients have lost 80% or more of their dopamine-producing cells by the time symptoms appear.

Parkinson’s disease afflicts 1 to 1 1/2 million people in the United States. The disorder occurs in all races but is somewhat more prevalent among Caucasians. Men are affected slightly more often than women.

Symptoms of Parkinson’s disease may appear at any age, but the average age of onset is 60. It is rare in people younger than 30 and risk increases with age. It is estimated that 5% to 10% of patients experience symptoms before the age of 40.

Source:
http://www.neurologychannel.com/parkinsonsdisease/

16.2 Patau’s syndrome

Patau’s syndrome occurs when the baby has an extra copy of chromosome 13. This is also a trisomy disorder. Most Patau’s syndrome babies do not survive more than a few days. The majority of pregnancies with this chromosomal abnormality results in a miscarriage. The odds of a live birth having Patau’s syndrome is 1 in 12,000. Babies born with Patau’s syndrome experience many complications. They are mentally challenged and have multiple physical abnormalities such as malformed feet, hands and facial features. They may also be deaf and have difficulties seeing and smelling. This is usually a result of incomplete brain development.

Source:
http://www.pregnancy-info.net/chromosomal_patau.html

16.3 Pertussis (whooping cough)

Whooping cough, a communicable, potentially deadly illness characterized by fits of coughing followed by a noisy, “whooping” indrawn breath. It is caused by the bacteria Bordetella pertussis. The illness is most likely to affect young children, but sometimes appears in teenagers and adults, even those who have been previously immunized. Immunization with DPT (diphtheria-pertussis-tetanus) vaccine provides protection, although that immunity may wear off with age.

Source:
16.4 **Pervasive development disorders**

The term “pervasive development disorders” (PDDs) refers to a group of conditions that involve delays in the development of many basic skills, most notably the ability to socialize with others, to communicate and to use imagination. Children with these conditions often are confused in their thinking and generally have problems understanding the world around them. There are five types of PDDs: Autism, Asperger’s syndrome, Childhood disintegrative disorder, Rett’s syndrome, Pervasive development disorder not otherwise specified (PDDNOS). The cause of these illnesses is not known. Some studies suggest that PDDs are caused by a problem with the nervous system (brain and spinal cord). It is estimated that PDDs occur in about 5 to 15 children per 10,000 births. In general, PDDs are more common in boys than in girls, with the exception of Rett’s syndrome, which occurs almost always in girls.


16.5 **Phenylketonuria**

The inherited inability to metabolize (process) the essential amino acid phenylalanine due to complete or near-complete deficiency of the enzyme phenylalanine hydroxylase. Newborns are screened for phenylketonuria (PKU) by a blood test, usually with the Guthrie card bloodspot obtained from a heelprick. Treatment is with a special diet low in phenylalanine. The goal is to normalize the levels of phenylalanine and tyrosine in the blood to prevent brain damage. Failure of treatment results in profound irreversible mental retardation, microcephaly (an abnormally small head), epilepsy, and behavior problems. It is clear that if the diet is not followed closely, especially during childhood, some impairment is inevitable. Maternal phenylketonuria requires a diet low in phenylalanine. Phenylketonuria is inherited in an autosomal recessive manner, as are lesser degrees of phenylalanine hydroxylase deficiency.


16.6 **Pick’s disease**

A form of dementia characterized by a slowly progressive deterioration of social skills and changes in personality leading to impairment of intellect, memory, and language.


16.7 **Plasma cell leukemia**

Plasma cell leukemia is the most advanced form of multiple myeloma. It is a rare disease representing 1 to 5% of cases of multiple myeloma. It is generally an aggressive disease with survivals ranging in months. As part of the multiple myeloma spectrum, it involves cyogenetic abnormalities commonly seen in multiple myeloma and virtually always translocations of the heavy chain immunoglobulin locus. The treatment is mostly ineffective but the most promising results have been obtained with the vincristine, doxorubicin, dexamethasone (VAD) chemotherapy regimen or with high dose cyclophosphamide and etoposide. High dose chemotherapy with autologous bone marrow support shows also promise in the few cases reported.

16.8 Poliomyelitis

An acute and sometimes devastating disease caused by a virus. The virus enters the mouth and multiplies in lymphoid tissues in the throat and intestine. Polio can be a minor illness, as it is in 80–90% of clinical infections, chiefly in young children, and not involve the CNS. Symptoms are slight fever, malaise, headache, sore throat, and vomiting 3–5 days after exposure. Recovery occurs in 24–72 hours. As a major illness, polio may or may not be paralytic. Symptoms usually appear without prior illness, particularly in older children and adults, 7–14 days after exposure. Symptoms are fever, severe headache, stiff neck and back, deep muscle pain, and sometimes areas of hyperesthesia (increased sensation) and paresthesia (altered sensation). Recovery is complete in the abortive and nonparalytic forms of polio. In paralytic polio, about 50% of patients recover with no residual paralysis, about 25% are left with mild disabilities, and the remaining patients have severe permanent disability. The greatest return of muscle function occurs in the first 6 months, but improvement may continue for up to 2 years. Polio(myelitis) is also called infantile paralysis.

Source:

Poliomyelitis, often called polio or infantile paralysis, is a viral paralytic disease. The causative agent, a virus called poliovirus (PV), enters the body orally, infecting the intestinal wall. It may proceed to the blood stream and into the central nervous system causing muscle weakness and often paralysis. An ancient disease, it was first recognized as a medical entity by Jakob Heine in 1840.

Source:
http://en.wikipedia.org/wiki/Acute_poliomyelitis

16.9 Plague

The plague is an infectious disease due to a bacteria called Yersinia pestis. Transmission of the plague to people can also occur from eating infected animals such as squirrels. Once someone has the plague, they can transmit it to another person via aerosol droplets.

Source:

16.10 Pneumonia

Inflammation of one or both lungs with consolidation. Pneumonia is frequently but not always due to infection. The infection may be bacterial, viral, fungal or parasitic. Symptoms may include fever, chills, cough with sputum production, chest pain, and shortness of breath.

Source:

16.11 Polyarteritis nodosa

An autoimmune disease characterized by spontaneous inflammation of the arteries (arteritis) of the body. Because arteries are involved, the disease can affect any organ of the body, most commonly muscles, joints, intestines, nerves, kidneys, and skin.

Source:
16.12 Polyostotic fibrous dysplasia

A genetic disorder of bones, skin pigmentation and hormonal problems with premature sexual development. Also called McCune-Albright syndrome or the Albright syndrome. In the syndrome, there is bone disease with fractures and deformity of the legs, arms and skull; pigment patches of the skin; and endocrine (hormonal) disease with early puberty (early menstrual bleeding, development of breasts and pubic hair) and an increased rate of growth. Polyostotic fibrous dysplasia is usually caused by mosaicism for a mutation in a gene called GNAS1 (Guanine Nucleotide binding protein, Alpha Stimulating activity polypeptide 1). The syndrome shows a broad spectrum of severity.

Source:

16.13 Prader-Willi syndrome

A syndrome characterized by severe hypotonia (floppiness), poor suck and feeding problems in early infancy followed later in infancy by excessive eating that, if unchecked, leads gradually to huge obesity. All children with Prader-Willi syndrome (PWS) show developmental delay and mild-to-moderate mental retardation with multiple learning disabilities. Hypogonadism is present in both females (with small labia minora and clitoris) and males (with underdeveloped scrotum and nondescent of the testes). Short stature and small hands and feet are common. It is due to absence of the paternally contributed region on chromosome 15q11–q13. What is missing is not just any chromosome 15q11–13 region but specifically that from the father. There is currently no specific treatment or cure for PWS. Parents are advised to limit consumption of high-calorie foods, and to use techniques such as special education, speech therapy, and physical therapy to maximize the child’s potential. Severe psychiatric illness is common in PWS adults. Those with psychotic illness have a double maternal copy of 15q11–13, suggesting that genes in this region are important in causing psychotic illness.

Source:

16.14 Prion-related disorders

A disease due to a prion, a proteinaceous infectious particle that lacks nucleic acids. Prions are composed largely, if not entirely, of an altered formal (an abnormal isoform) of a normal cellular protein. The known prion diseases of humans and other mammals are: bovine spongiform encephalopathy (BSE), Creutzfeldt-Jakob disease (CJD), Gerstmann-Straussler-Scheinker syndrome (GSS), fatal familial insomnia (FFI), kuru, scrapie, transmissible mink encephalopathy (TME), chronic wasting disease (CWD), feline spongiform encephalopathy (FSE), exotic ungulate encephalopathy (EUE).

Source:

16.15 Primary cerebellar degeneration

A heterogenous group of degenerative syndromes marked by progressive cerebellar dysfunction either in isolation or combined with other neurologic manifestations. Sporadic and inherited subtypes occur. Inheritance patterns include autosomal dominant, autosomal recessive, and X-linked.

Source:
16.16 Progeria

A rare genetic disorder that causes children to age prematurely. The classic type of childhood progeria is Hutchinson-Gilford syndrome, which is commonly referred to as progeria. It is characterized by dwarfism, baldness, pinched nose, small face and small jaw relative to the head size, delayed tooth formation, aged-looking skin, diminution of fat beneath the skin, stiff joints, and premature arteriosclerosis. Children with the progeria syndrome usually appear normal at birth. However, within a year, their growth rate slows and their appearance begins to change and age prematurely. They often suffer from symptoms typically seen in elderly people, especially severe cardiovascular disease. Death occurs on average at age 13, usually from heart attack or stroke. There currently are no diagnostic tests or treatments for progeria which remains relentlessly progressive and fatal.

Source:

16.17 Psoriasis

Psoriasis is an autoimmune disease affecting the skin and joints. When it affects the skin it commonly appears as red scaly elevated patches called plaques. Psoriasis plaques frequently occur on the elbows and knees, but can affect any area of skin including the scalp and genital area. Fingernails and toenails are often affected (psoriatic nail dystrophy). Psoriasis can also cause inflammation of the joints (psoriatic arthritis). Psoriatic arthritis can affect the hips, knees and spine (spondylitis). The prevalence of psoriasis in Western populations is estimated to be around 2-3%. It affects both sexes equally and occurs at all ages. Several factors are thought to be affravate psoriasis. These include stress and excessive alcohol consumption. Individuals with psoriasis may also suffer from depression and loss of self-esteem.

Source:
http://en.wikipedia.org/wiki/Psoriasis

17 Disease list and descriptions: R

17.1 Reiter’s syndrome

A chronic form of inflammatory arthritis wherein the following three conditions are combined: (1) arthritis; (2) inflammation of the eyes (conjunctivitis); and (3) inflammation of the genital, urinary or gastrointestinal systems. Reiter syndrome is a systemic rheumatic disease, meaning that it can and does affect organs as well as the joints. It can cause inflammation in many areas, including the eyes, mouth, lungs, kidneys, heart, and skin.

Source:
17.2 Renal glycosuria

Renal glycosuria, also known as renal glucosuria, is a rare condition in which the simple sugar glucose is excreted in the urine despite normal or low blood glucose levels. With normal kidney (renal) function, glucose is excreted in the urine only when there are abnormally elevated levels of glucose in the blood. However, in those with renal glycosuria, glucose is abnormally eliminated in the urine due to improper functioning of the renal tubules, which are primary components of nephrons, the filtering units of the kidneys. In most affected individuals, the condition causes no apparent symptoms or serious effects. When renal glycosuria occurs as an isolated finding with otherwise normal kidney function, the condition is thought to be inherited as an autosomal recessive trait.

Source:
http://en.wikipedia.org/wiki/Glycosuria

17.3 Reticulosarcoma (large cell lymphoma)

Reticulosarcoma is the most common aggressive form of non-Hodgkin lymphoma. It occurs in both diffuse and nodular form. The large cells may have cleaved and non-cleaved nuclei. The median age is 57, with a range of 10–88 years. The specific cause of most forms of non-Hodgkin lymphoma is unclear. It is possible that genetics and exposure to viral infections may increase the risk for developing this malignancy. Children and adults with other hereditary abnormalities have an increased risk of developing non-Hodgkin lymphoma, including patients with ataxia telangiectasia, X-linked lymphoproliferative disease, or the Wiskott-Aldrich syndrome.

Source:
http://ncimeta.nci.nih.gov/MetaServlet/servlet/ResultServlet2?conceptID=C0024302
http://www.lymphomainfo.net/nhl/largecell.html

17.4 Rheumatoid arthritis

Rheumatoid arthritis is an autoimmune disease that causes chronic inflammation of the joints. Rheumatoid arthritis can also cause inflammation of the tissue around the joints, as well as other organs in the body. Autoimmune diseases are illnesses which occur when the body tissues are mistakenly attacked by its own immune system. Because it can affect multiple other organs of the body, rheumatoid arthritis is referred to as a systemic illness and is sometimes called rheumatoid disease. While rheumatoid arthritis is a chronic illness, meaning it can last for years, patients may experience long periods without symptoms. Typically, however, rheumatoid arthritis is a progressive illness that has the potential to cause joint destruction and functional disability. In some patients with rheumatoid arthritis, chronic inflammation leads to the destruction of the cartilage, bone and ligaments causing deformity of the joints. Damage to the joints can occur early in the disease and be progressive. Rheumatoid arthritis is a common rheumatic disease, affecting more than two million people in the United States. The disease is three times more common in women as in men. It afflicts people of all races equally. The disease can begin at any age, but most often starts after age forty and before sixty. In some families, multiple members can be affected, suggesting a genetic basis for the disorder.

Source:
http://www.medicinenet.com/rheumatoid\_arthritis/article.htm
18 Disease list and descriptions: S

18.1 Salmonellosis

Infection with bacteria belonging to the genus *Salmonella*. Salmonellosis is a common cause of food poisoning as, for example, from raw eggs. The symptoms of salmonellosis usually begin within 12 to 24 hours of exposure to the bacteria and include stomach cramps, diarrhea, fever, and sometimes vomiting. Most people exposed to *Salmonella* feel well within a few days and do not require treatment other than extra fluids. Some people need antibiotics. And a few need hospitalization for diarrhea and dehydration. Salmonellosis is particularly dangerous in people with immunodeficiency and in people with sickle cell anemia.

*Source:*


18.2 Schilder’s disease

Schilder’s disease is a rare progressive demyelinating disorder which usually begins in childhood. Symptoms may include dementia, aphasia, seizures, personality changes, poor attention, tremors, balance instability, incontinence, muscle weakness, headache, vomiting, and vision and speech impairment. The disorder is a variant of multiple sclerosis. As with multiple sclerosis, the course and prognosis of Schilder’s disease are unpredictable. In some cases, Schilder’s disease is fatal.

*Source:*


18.3 Schizophrenia

One of several brain diseases whose symptoms that may include loss of personality (flat affect), agitation, catatonia, confusion, psychosis, unusual behavior, and withdrawal. The illness usually begins in early adulthood. The causes of schizophrenia are not yet fully known. Schizophrenia is not caused by poor parenting practices. A variant version of a gene called COMT has been found to increase the risk for developing schizophrenia. Other genes and environmental factors may well be involved in schizophrenia.

*Source:*


18.4 Scleroderma

A disease of connective tissue with the formation of scar tissue (fibrosis) in the skin and sometimes also in other organs of the body. The cause of scleroderma is not known. The disease is more frequent in females than in males.

*Source:*

18.5 **Shigellosis**

Epidemic and opportunistic bacillary dysentery due to infection with the *Shigella* bacteria. Shigellosis causes intestinal pain and diarrhea with mucus and blood in the stool. It is especially common in tropical countries, but frequently occurs elsewhere. It is a particular hazard for people with AIDS or other immunodeficiency states. Named for the Japanese bacteriologist Kiyoshi Shiga (1870-1957).

*Source:*

18.6 **Sjogren’s syndrome**

Sjogren’s syndrome classically features a combination of dry eyes, dry mouth, and another disease of the connective tissues, most commonly rheumatoid arthritis. Sjogren’s syndrome is an autoimmune disease, characterized by the abnormal production of extra antibodies in the blood that are directed against various tissues of the body. This particular autoimmune illness is caused by inflammation in the glands of the body. It is also found more commonly in families that have members with other autoimmune illnesses, such as systemic lupus erythematosus, autoimmune thyroid disease, juvenile diabetes, etc. 90% of Sjogren’s syndrome patients are female.

*Source:*
http://www.medicinenet.com/sjogrens_syndrome/article.htm

18.7 **SLE (systemic lupus erythematosus)**

A chronic inflammatory condition caused by an autoimmune disease. Lupus can cause disease of the skin, heart, lungs, kidneys, joints, and nervous system. When only the skin is involved, the condition is called discoid lupus. When internal organs are involved, the condition is called systemic lupus erythematosus (SLE). Up to 10% of persons with discoid lupus (lupus limited to the skin) eventually develop the systemic form of lupus (SLE). SLE is eight times more common in women than men. The causes of SLE are unknown. However, heredity, viruses, ultraviolet light, and drugs may all play a role.

*Source:*

18.8 **Spina bifida occulta**

A bony defect in the vertebral column that causes a cleft in that column. The cleft remains covered by skin. Treatment is usually not required.

*Source:*

18.9 **Spondylolisthesis**

Spondylolisthesis differs in children and adults. Spondylolisthesis can also be due to stress fracture (common in gymnasts), traumatic fracture, and bone disease. Symptoms and signs can include lordosis (swayback), pain in the lower back, thighs and buttocks, stiffness, muscle tightness, and tenderness in the slipped area. Pressure on nerve roots may cause changes in sensation and pain radiating down the legs.

*Source:*
18.10  Staph infection

*Staphylococcus* (staph) infection can be simple and localized, such as with impetigo of the skin. It can, however, become widespread, by infecting the blood. It can thereby seed to various areas of the body, such as the bone, kidneys, or heart. This spreading occurs more commonly in persons with abnormally suppressed immune systems.

*Source:*
http://www.medicinenet.com/staph_infection/article.htm

18.11  Strep infection

*Group A Streptococcus* (GAS) is a bacterium often found in the throat and on the skin. Most GAS infections are relatively mild illnesses such as “strep throat,” or impetigo. On rare occasions, these bacteria can cause other severe and even life-threatening diseases (necrotizing faciitis, streptococcal toxic shock syndrome). *Group B Streptococcus* (GBS) is a type of bacteria that causes illness in newborn babies, pregnant women, the elderly, and adults with other illness, such as diabetes or liver disease. GBS is the most common cause of life-threatening infections in newborns.

*Source:*
http://www.cdc.gov/ncidod/dbmd/diseaseinfo/groupastreptococcal_g.htm

19  Disease list and descriptions: T

19.1  Takayasu disease

A chronic inflammatory disease of the aorta and its branch arteries. The cause is unknown. The disease is most common in young women of Asian descent and usually begins between 10 and 30 years of age. Symptoms include painful, cool or blanched extremities, dizziness, headaches, chest and abdominal pain, and low-grade fever. The blood pressure is often high. The sedimentation rate (sed rate) may be elevated, reflecting inflammation. Anemia is frequent.

*Source:*

19.2  TTP (thrombotic thrombocytopenic purpura)

A life-threatening disease involving embolism and thrombosis (plugging) of the small blood vessels in the brain. TTP is characterized by platelet microthrombi (tiny traveling clots composed of platelets, the clotting cells in the blood), thrombocytopenia (lack of platelets), hemolytic anemia (from the breakup of red blood cells), fever, renal (kidney) abnormalities and neurologic changes such as neurological signs such as aphasia, blindness, and convulsions. It occurs at a rate of 3.7 cases per year per million persons. The mortality (death) rate for promptly treated cases ranges from 10 to 20 percent.

*Source:*
19.3 Tuberculosis (TB)

Tuberculosis (TB) is an infectious disease caused by bacteria whose scientific name is *Mycobacterium tuberculosis*. TB most commonly affects the lungs but also can involve most any organ of the body. Tuberculosis is spread (transmitted) primarily from person to person during close contact by breathing infected air. Over 8 million new cases of TB occur each year worldwide. In the United States, it is estimated that 10–15 million people are infected with the TB bacteria and 22,000 new cases of TB occur each year. The usual symptoms that occur with an active TB infection are a generalized tiredness or weakness, weight loss, fever, and night sweats. If the infection in the lung worsens, then further symptoms can include coughing, chest pain, coughing up of sputum (material from the lungs) and/or blood, and shortness of breath.

*Source:*

http://www.medicinenet.com/tuberculosis/article.htm

19.4 Tuberous sclerosis

A genetic disorder characterized by abnormalities of the skin, brain, kidney, and heart. Tuberous sclerosis is inherited in an autosomal dominant manner and results from mutation of either one of two genes: the TSC1 gene on chromosome 9 or the TSC2 gene on chromosome 16. Two-thirds of cases of tuberous sclerosis are due to new mutations and the other third are inherited from a parent.

*Source:*


19.5 Tularemia

A bacterial disease caused by infection with a bacterium called *Francisella tularensis* that usually occurs in wild and domestic animals, most often rabbits, and can be transmitted to humans by contact with animal tissues or ticks and fleas. Also called rabbit fever and deerfly fever. Symptoms appear 2–10 days after exposure. Most often there is a red spot on the skin which enlarges and ulcerates together with enlarged lymph nodes (swollen glands) in the axilla (armpit) or groin. Ingestion of the organism may produce a throat infection, intestinal pain, diarrhea and vomiting. Inhalation of the organism may produce a fever or a pneumonia-like illness. Tularemia is a dangerous disease. It is fatal in about 5% of untreated cases but less than 1% of treated cases.

*Source:*


19.6 Typhoid fever

An acute illness with fever caused by infection with the Salmonella typhi bacteria contracted from contaminated water and food. The disease has an insidious onset characterized by fever, headache, constipation, malaise, chills, and myalgia (muscle pain). Diarrhea is uncommon, and vomiting is not usually severe. Confusion, delirium, intestinal perforation, and death may occur in severe cases. Without therapy, the illness may last for 3 to 4 weeks and death rates range between 12% and 30%. About 16 million cases of typhoid fever and 600,000 deaths occur yearly worldwide. There are about 400 cases a year in the US, mostly among travelers.

*Source:*

20 Disease list and descriptions: U

20.1 Urea cycle metabolism (urea cycle disorder)

An urea cycle disorder is a genetic disorder caused by a deficiency of one of the enzymes in the urea cycle which is responsible for removing ammonia from the blood stream. The urea cycle involves a series of biochemical steps in which nitrogen, a waste product of protein metabolism, is removed from the blood and converted to urea. Normally, the urea is transferred into the urine and removed from the body. In urea cycle disorders, the nitrogen accumulates in the form of ammonia, a highly toxic substance, and is not removed from the body. Ammonia then reaches the brain through the blood, where it causes irreversible brain damage and/or death. Urea cycle disorders are included in the category of inborn errors of metabolism. There is no cure. Because many cases of urea cycle disorders remain undiagnosed and/or infants born with the disorders die without a definitive diagnosis, the exact incidence of these cases is unknown and underestimated. It is believed that up to 20% of Sudden infant Death Syndrome cases may be attributed to an undiagnosed inborn error of metabolism such a urea cycle disorder. In April 2000, research experts at the Urea Cycle Consensus Conference estimated the incidence of the disorders at 1 in 10000 births.

Source:
http://en.wikipedia.org/wiki/Urea_cycle_disorder#The_six_urea_cycle_disorders

21 Disease list and descriptions: V

21.1 Viral infection

Infection caused by the presence of a virus in the body. Depending on the virus and the persons state of health, various viruses can infect almost any type of body tissue, from the brain to the skin. Viral infections cannot be treated with antibiotics; in fact, in some cases the use of antibiotics makes the infection worse. The vast majority of human viral infections can be effectively fought by the body’s own immune system, with a little help in the form of proper diet, hydration, and rest. As for the rest, treatment depends on the type and location of the virus, and may include anti-viral or other drugs.

Source:

21.2 Viral infections of the central nervous system (CNS)

21.2.1 Dawson’s inclusion body encephalitis

Rare chronic progressive encephalitis caused by the measles virus and occurring primarily in children and young adults; death usually occurs within three years; characterized by primary measles infection before the age of two years.

Source:
http://www.wrongdiagnosis.com/medical/dawson_s_inclusion_body_encephalitis.htm

21.2.2 Van Bogaert’s sclerosing leukoencephalitis

A rare, chronic and progressive encephalitis in children and adolescents, involving the white matter of the cerebrum, brain stem, cerebral cortex, thalamus, and spinal cord.

Source:
Van Bogaert’s sclerosing leukoencephalitis: rare chronic progressive encephalitis caused by the measles virus and occurring primarily in children and young adults; death usually occurs within three years; characterized by primary measles infection before the age of two years.

Source:
http://www.wrongdiagnosis.com/medical/van_bogaert_s_sclerosing_leukoencephalitis.htm

21.2.3 Progressive multifocal leukoencephalopathy

Progressive multifocal leukoencephalopathy (PML) is an infrequent disorder of the nervous system that primarily affects individuals with suppressed immune systems (including, allograft recipients such as kidney transplant patients; patients with cancers such as leukemia or lymphoma; and nearly 10% of patients with acquired immune deficiency syndrome — AIDS. The disorder, which is caused by a common human polyomavirus, JC virus, is characterized by demyelination or destruction of the myelin sheath that covers nerve cells. The myelin sheath is the fatty covering — which acts as an insulator — on nerve fibers in the brain. Symptoms of PML include mental deterioration, vision loss, speech disturbances, ataxia (inability to coordinate movements), paralysis, and, ultimately, coma reflecting the multifocal distribution of brain lesions. In rare cases, seizures may occur.

Source:

21.2.4 Unspecified slow virus infection of central nervous system

21.3 Vitamin deficiency

If the intake of vitamin(s) is insufficient due to poor nutrition, restricted diets, or inadequate intestinal absorption of the vitamins, diseases can occur. Examples of diseases caused by vitamin deficiencies include anemia (due to deficiencies of folic acid and vitamin B12), nerve and brain damage (due to deficiencies of thiamin and vitamin B12), easy and excessive bleeding (due to deficiency of vitamin K), impaired night vision and blindness (due to deficiency of vitamin A), bone diseases (due to deficiency of vitamin D or calcium), and scurvy (due to deficiency of vitamin C). These vitamin deficiency diseases are rare in the western societies, and occur mainly in areas of the world where people have very poor diets.

Source:
http://www.medicinenet.com/vitamins_and_calcium_supplements/article.htm

22 Disease list and descriptions: W

22.1 Wegener’s granulomatosis

An uncommon type of inflammation of small arteries and veins (vasculitis) that classically involves the vessels supplying the tissues of the lungs, nasal passages (sinuses), and kidneys. Wegener’s granulomatosis usually affects young or middle-aged adults. Symptoms include fatigue, weight loss, fever, shortness of breath, bloody sputum, joint pains, and sinus inflammation, sometimes with nasal ulcerations and bloody nasal discharge. Wegener’s granulomatosis is a serious disease. Without treatment, it can be fatal within months.

Source:
22.2  WPW syndrome

WPW is an abbreviation for the Wolff–Parkinson–White syndrome, a condition caused by an abnormality in the electrical system of the heart which normally tells the heart muscle when to contract. In the WPW syndrome, there is an extra electrical connection inside the heart that acts as a short circuit, causing the heart to beat too rapidly and sometimes in an irregular manner. The syndrome can be life-threatening although this is unusual.

Source:

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Number of records</th>
<th>Raw %</th>
<th>Adjusted %</th>
</tr>
</thead>
<tbody>
<tr>
<td>Amino acid (AA) metabolism (aromatic)</td>
<td>79</td>
<td>0.005</td>
<td>0.008</td>
</tr>
<tr>
<td>Amino acid (AA) metabolism (branched)</td>
<td>16</td>
<td>0.001</td>
<td>0.002</td>
</tr>
<tr>
<td>Amino acid (AA) metabolism (Lowe)</td>
<td>24</td>
<td>0.002</td>
<td>0.002</td>
</tr>
<tr>
<td>Amino acid (AA) metabolism (sulfur-bearing)</td>
<td>826</td>
<td>0.055</td>
<td>0.082</td>
</tr>
<tr>
<td>Amino acid (AA) metabolism (straight-chain)</td>
<td>55</td>
<td>0.004</td>
<td>0.005</td>
</tr>
<tr>
<td>Amino acid (AA) transport</td>
<td>159</td>
<td>0.011</td>
<td>0.016</td>
</tr>
<tr>
<td>Acanthosis nigricans</td>
<td>431</td>
<td>0.029</td>
<td>0.043</td>
</tr>
<tr>
<td>Acidosis</td>
<td>6,239</td>
<td>0.414</td>
<td>0.620</td>
</tr>
<tr>
<td>Aciduria</td>
<td>25</td>
<td>0.002</td>
<td>0.002</td>
</tr>
<tr>
<td>Actinomycosis</td>
<td>1,467</td>
<td>0.097</td>
<td>0.146</td>
</tr>
<tr>
<td>Acute leukemia</td>
<td>132</td>
<td>0.009</td>
<td>0.013</td>
</tr>
<tr>
<td>Acute promyelocytic leukemia</td>
<td>172</td>
<td>0.011</td>
<td>0.017</td>
</tr>
<tr>
<td>Ainhum</td>
<td>26</td>
<td>0.002</td>
<td>0.003</td>
</tr>
<tr>
<td>Albright-Sternberg syndrome</td>
<td>196</td>
<td>0.013</td>
<td>0.016</td>
</tr>
<tr>
<td>Alcoholism</td>
<td>27,638</td>
<td>1.835</td>
<td>2.748</td>
</tr>
<tr>
<td>Allergic rhinitis</td>
<td>19,216</td>
<td>1.276</td>
<td>1.910</td>
</tr>
<tr>
<td>Alkalosis</td>
<td>1,605</td>
<td>0.107</td>
<td>0.160</td>
</tr>
<tr>
<td>Alopecia</td>
<td>3,979</td>
<td>0.264</td>
<td>0.396</td>
</tr>
<tr>
<td>Alopecia areata</td>
<td>821</td>
<td>0.055</td>
<td>0.082</td>
</tr>
<tr>
<td>Alzheimer's disease</td>
<td>9,073</td>
<td>0.603</td>
<td>0.902</td>
</tr>
<tr>
<td>Amebiasis</td>
<td>110</td>
<td>0.007</td>
<td>0.011</td>
</tr>
<tr>
<td>Amyotrophic lateral sclerosis</td>
<td>2,182</td>
<td>0.145</td>
<td>0.217</td>
</tr>
<tr>
<td>Anaerobes</td>
<td>709</td>
<td>0.047</td>
<td>0.070</td>
</tr>
<tr>
<td>Aniridia</td>
<td>11</td>
<td>0.001</td>
<td>0.001</td>
</tr>
<tr>
<td>Ankylosing spondylitis</td>
<td>510</td>
<td>0.034</td>
<td>0.051</td>
</tr>
<tr>
<td>Anthrax</td>
<td>2,704</td>
<td>0.180</td>
<td>0.269</td>
</tr>
<tr>
<td>Aortic aneurysm</td>
<td>2,990</td>
<td>0.199</td>
<td>0.297</td>
</tr>
<tr>
<td>Aplastic anemia</td>
<td>5,191</td>
<td>0.345</td>
<td>0.516</td>
</tr>
<tr>
<td>Attention deficit</td>
<td>6,964</td>
<td>0.462</td>
<td>0.692</td>
</tr>
<tr>
<td>Autism</td>
<td>481</td>
<td>0.032</td>
<td>0.048</td>
</tr>
<tr>
<td>Behcet’s syndrome</td>
<td>53</td>
<td>0.004</td>
<td>0.005</td>
</tr>
<tr>
<td>Benign neoplasms</td>
<td>77,272</td>
<td>5.132</td>
<td>7.682</td>
</tr>
<tr>
<td>Bipolar disorder</td>
<td>12,373</td>
<td>0.822</td>
<td>1.230</td>
</tr>
<tr>
<td>Breast cancer (female)</td>
<td>15,361</td>
<td>1.020</td>
<td>1.527</td>
</tr>
<tr>
<td>Breast cancer (male)</td>
<td>700</td>
<td>0.046</td>
<td>0.070</td>
</tr>
<tr>
<td>Brucellos</td>
<td>298</td>
<td>0.020</td>
<td>0.030</td>
</tr>
<tr>
<td>Budd-Chiari syndrome</td>
<td>152</td>
<td>0.010</td>
<td>0.015</td>
</tr>
<tr>
<td>Bundle branch block</td>
<td>11,853</td>
<td>0.787</td>
<td>1.178</td>
</tr>
<tr>
<td>Buphthalmnos</td>
<td>311</td>
<td>0.021</td>
<td>0.031</td>
</tr>
<tr>
<td>Burkitt’s lymphoma</td>
<td>104</td>
<td>0.007</td>
<td>0.010</td>
</tr>
<tr>
<td>Carcinoma in situ</td>
<td>31,151</td>
<td>2.069</td>
<td>3.097</td>
</tr>
<tr>
<td>Carbohydrate transport and metabolism</td>
<td>778</td>
<td>0.052</td>
<td>0.077</td>
</tr>
<tr>
<td>Cardiomyopathy</td>
<td>11,457</td>
<td>0.761</td>
<td>1.139</td>
</tr>
<tr>
<td>Celiac sprue</td>
<td>1,954</td>
<td>0.130</td>
<td>0.194</td>
</tr>
<tr>
<td>Cerebral palsy</td>
<td>2,968</td>
<td>0.197</td>
<td>0.295</td>
</tr>
<tr>
<td>Cervical rib</td>
<td>44</td>
<td>0.003</td>
<td>0.004</td>
</tr>
<tr>
<td>Charcot-Marie-Tooth syndrome</td>
<td>247</td>
<td>0.016</td>
<td>0.025</td>
</tr>
<tr>
<td>Cholelithiasis</td>
<td>15,353</td>
<td>1.020</td>
<td>1.526</td>
</tr>
<tr>
<td>Cholera</td>
<td>29</td>
<td>0.002</td>
<td>0.003</td>
</tr>
<tr>
<td>Chondrodystrophy</td>
<td>207</td>
<td>0.014</td>
<td>0.021</td>
</tr>
</tbody>
</table>

Table 1: Disorders, record counts, and the raw and adjusted prevalence.
<table>
<thead>
<tr>
<th>Disorder</th>
<th>Number of records</th>
<th>Raw %</th>
<th>Adjusted %</th>
</tr>
</thead>
<tbody>
<tr>
<td>Congenital absence of vertebra</td>
<td>30</td>
<td>0.002</td>
<td>0.003</td>
</tr>
<tr>
<td>Congenital spinal fusion</td>
<td>44</td>
<td>0.003</td>
<td>0.004</td>
</tr>
<tr>
<td>Cystic fibrosis</td>
<td>1,740</td>
<td>0.116</td>
<td>0.173</td>
</tr>
<tr>
<td>Dejerine-Sottas disease</td>
<td>479</td>
<td>0.032</td>
<td>0.048</td>
</tr>
<tr>
<td>Depression</td>
<td>27,085</td>
<td>1.799</td>
<td>2.693</td>
</tr>
<tr>
<td>Dermatomyositis-polymyositis</td>
<td>642</td>
<td>0.043</td>
<td>0.064</td>
</tr>
<tr>
<td>Diabetes mellitus type 1</td>
<td>19,372</td>
<td>1.286</td>
<td>1.926</td>
</tr>
<tr>
<td>Diabetes mellitus type 2</td>
<td>60,815</td>
<td>4.039</td>
<td>6.046</td>
</tr>
<tr>
<td>Diphtheria</td>
<td>52</td>
<td>0.003</td>
<td>0.005</td>
</tr>
<tr>
<td>E. coli intestinal</td>
<td>102</td>
<td>0.007</td>
<td>0.010</td>
</tr>
<tr>
<td>Edward’s syndrome</td>
<td>74</td>
<td>0.005</td>
<td>0.007</td>
</tr>
<tr>
<td>Enzyme-deficiency (hemolytic anemia)</td>
<td>57</td>
<td>0.004</td>
<td>0.006</td>
</tr>
<tr>
<td>Epilepsy</td>
<td>12,099</td>
<td>0.803</td>
<td>1.203</td>
</tr>
<tr>
<td>Erythematousquamous dermatosis</td>
<td>2,496</td>
<td>0.166</td>
<td>0.248</td>
</tr>
<tr>
<td>Food poisoning</td>
<td>1,184</td>
<td>0.079</td>
<td>0.118</td>
</tr>
<tr>
<td>Friedreich’s ataxia</td>
<td>46</td>
<td>0.003</td>
<td>0.005</td>
</tr>
<tr>
<td>Fragile X syndrome</td>
<td>9</td>
<td>0.001</td>
<td>0.001</td>
</tr>
<tr>
<td>Giant cell arteritis</td>
<td>828</td>
<td>0.055</td>
<td>0.082</td>
</tr>
<tr>
<td>Gram-negative bacteria</td>
<td>17,560</td>
<td>1.166</td>
<td>1.746</td>
</tr>
<tr>
<td>Goiter</td>
<td>10,820</td>
<td>0.719</td>
<td>1.076</td>
</tr>
<tr>
<td>Goodpasture’s syndrome</td>
<td>21</td>
<td>0.001</td>
<td>0.002</td>
</tr>
<tr>
<td>Gout</td>
<td>192</td>
<td>0.013</td>
<td>0.019</td>
</tr>
<tr>
<td>HIV</td>
<td>6,138</td>
<td>0.408</td>
<td>0.610</td>
</tr>
<tr>
<td>Helicobacter pilori</td>
<td>4,718</td>
<td>0.313</td>
<td>0.469</td>
</tr>
<tr>
<td>Hemivertebra</td>
<td>60</td>
<td>0.004</td>
<td>0.006</td>
</tr>
<tr>
<td>Hepatitis A</td>
<td>1,131</td>
<td>0.075</td>
<td>0.112</td>
</tr>
<tr>
<td>Hepatitis B</td>
<td>5,757</td>
<td>0.382</td>
<td>0.572</td>
</tr>
<tr>
<td>Hepatitis C</td>
<td>18,421</td>
<td>1.223</td>
<td>1.831</td>
</tr>
<tr>
<td>Hepatitis D</td>
<td>46</td>
<td>0.003</td>
<td>0.005</td>
</tr>
<tr>
<td>Hepatitis E</td>
<td>32</td>
<td>0.002</td>
<td>0.003</td>
</tr>
<tr>
<td>Hereditary spastic paraplegia</td>
<td>53</td>
<td>0.004</td>
<td>0.005</td>
</tr>
<tr>
<td>Hodgkin’s disease</td>
<td>176</td>
<td>0.012</td>
<td>0.017</td>
</tr>
<tr>
<td>Hyperosmolality</td>
<td>2,910</td>
<td>0.193</td>
<td>0.289</td>
</tr>
<tr>
<td>Hypoosmolality</td>
<td>12,942</td>
<td>0.859</td>
<td>1.287</td>
</tr>
<tr>
<td>Hypersensitivity angiitis</td>
<td>1,492</td>
<td>0.099</td>
<td>0.148</td>
</tr>
<tr>
<td>Hypertrophic cardiomyopathy</td>
<td>603</td>
<td>0.040</td>
<td>0.060</td>
</tr>
<tr>
<td>Hypoglycemia</td>
<td>373</td>
<td>0.025</td>
<td>0.037</td>
</tr>
<tr>
<td>Ichthyosis congenita</td>
<td>170</td>
<td>0.011</td>
<td>0.017</td>
</tr>
<tr>
<td>Kawasaki’s disease</td>
<td>495</td>
<td>0.033</td>
<td>0.049</td>
</tr>
<tr>
<td>Keratoderma</td>
<td>670</td>
<td>0.044</td>
<td>0.067</td>
</tr>
<tr>
<td>Klippel-Feil syndrome</td>
<td>51</td>
<td>0.003</td>
<td>0.005</td>
</tr>
<tr>
<td>Leprosy</td>
<td>479</td>
<td>0.032</td>
<td>0.048</td>
</tr>
<tr>
<td>Lethal midline granuloma</td>
<td>29</td>
<td>0.002</td>
<td>0.003</td>
</tr>
<tr>
<td>Leukodystrophy</td>
<td>223</td>
<td>0.015</td>
<td>0.022</td>
</tr>
<tr>
<td>Lown-Ganong-Levine syndrome</td>
<td>20</td>
<td>0.001</td>
<td>0.002</td>
</tr>
<tr>
<td>Lumbosacral spondylolysis</td>
<td>415</td>
<td>0.028</td>
<td>0.041</td>
</tr>
<tr>
<td>Lymphosarcoma</td>
<td>403</td>
<td>0.027</td>
<td>0.040</td>
</tr>
<tr>
<td>Lipid metabolism disease</td>
<td>57,308</td>
<td>3.806</td>
<td>5.698</td>
</tr>
<tr>
<td>Meningococcus</td>
<td>4,733</td>
<td>0.314</td>
<td>0.471</td>
</tr>
<tr>
<td>Migraine</td>
<td>8,049</td>
<td>0.535</td>
<td>0.800</td>
</tr>
</tbody>
</table>

Table 2: Disorders, record counts, and the raw and adjusted prevalence.
<table>
<thead>
<tr>
<th>Disorder</th>
<th>Number of records</th>
<th>Raw %</th>
<th>Adjusted %</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mineral metabolism (Fe)</td>
<td>460</td>
<td>0.031</td>
<td>0.046</td>
</tr>
<tr>
<td>Mineral metabolism (Cu)</td>
<td>53</td>
<td>0.004</td>
<td>0.005</td>
</tr>
<tr>
<td>Mineral metabolism (Mg)</td>
<td>1,133</td>
<td>0.075</td>
<td>0.113</td>
</tr>
<tr>
<td>Mineral metabolism (Ca)</td>
<td>3,933</td>
<td>0.261</td>
<td>0.391</td>
</tr>
<tr>
<td>Mitochondrial disease</td>
<td>31</td>
<td>0.002</td>
<td>0.003</td>
</tr>
<tr>
<td>Moyamoya</td>
<td>130</td>
<td>0.009</td>
<td>0.013</td>
</tr>
<tr>
<td>Multiple epiphyseal dysplasia</td>
<td>14</td>
<td>0.001</td>
<td>0.001</td>
</tr>
<tr>
<td>Multiple sclerosis</td>
<td>14,979</td>
<td>0.995</td>
<td>1.489</td>
</tr>
<tr>
<td>Mumps</td>
<td>3,901</td>
<td>0.259</td>
<td>0.388</td>
</tr>
<tr>
<td>Muscular distrophy</td>
<td>746</td>
<td>0.050</td>
<td>0.074</td>
</tr>
<tr>
<td>Mycoplasma</td>
<td>16</td>
<td>0.001</td>
<td>0.002</td>
</tr>
<tr>
<td>Myotonic disorders</td>
<td>201</td>
<td>0.013</td>
<td>0.020</td>
</tr>
<tr>
<td>Neuromyelitis optica</td>
<td>47</td>
<td>0.003</td>
<td>0.005</td>
</tr>
<tr>
<td>Neurofibromatosis</td>
<td>638</td>
<td>0.042</td>
<td>0.063</td>
</tr>
<tr>
<td>Ornithosis</td>
<td>44</td>
<td>0.003</td>
<td>0.004</td>
</tr>
<tr>
<td>Osteogenesis imperfecta</td>
<td>125</td>
<td>0.008</td>
<td>0.012</td>
</tr>
<tr>
<td>Osteopetrosis</td>
<td>64</td>
<td>0.004</td>
<td>0.006</td>
</tr>
<tr>
<td>Parkinson’s disease</td>
<td>6,116</td>
<td>0.406</td>
<td>0.608</td>
</tr>
<tr>
<td>Patau’s syndrome</td>
<td>75</td>
<td>0.005</td>
<td>0.007</td>
</tr>
<tr>
<td>Pertussis</td>
<td>425</td>
<td>0.028</td>
<td>0.042</td>
</tr>
<tr>
<td>Pervasive developmental disorder</td>
<td>1,162</td>
<td>0.077</td>
<td>0.116</td>
</tr>
<tr>
<td>Phenyktonurina</td>
<td>15</td>
<td>0.001</td>
<td>0.001</td>
</tr>
<tr>
<td>Pick’s disease</td>
<td>11</td>
<td>0.001</td>
<td>0.001</td>
</tr>
<tr>
<td>Plague</td>
<td>118</td>
<td>0.008</td>
<td>0.012</td>
</tr>
<tr>
<td>Plasma cell leukemia</td>
<td>7</td>
<td>0.000</td>
<td>0.001</td>
</tr>
<tr>
<td>Pneumonia</td>
<td>13,192</td>
<td>0.876</td>
<td>1.312</td>
</tr>
<tr>
<td>Poliomyelitis</td>
<td>3,581</td>
<td>0.238</td>
<td>0.356</td>
</tr>
<tr>
<td>Polyarteritis nodosa</td>
<td>161</td>
<td>0.011</td>
<td>0.016</td>
</tr>
<tr>
<td>Polyostatic fibrous dysplasia of bone</td>
<td>10</td>
<td>0.001</td>
<td>0.001</td>
</tr>
<tr>
<td>Prader-Willi syndrome</td>
<td>56</td>
<td>0.004</td>
<td>0.006</td>
</tr>
<tr>
<td>Primary cerebellar degeneration</td>
<td>91</td>
<td>0.006</td>
<td>0.009</td>
</tr>
<tr>
<td>Prion-related disease</td>
<td>2,325</td>
<td>0.154</td>
<td>0.231</td>
</tr>
<tr>
<td>CNS viral disease</td>
<td>76,835</td>
<td>5.103</td>
<td>7.639</td>
</tr>
<tr>
<td>Progeria</td>
<td>89</td>
<td>0.006</td>
<td>0.009</td>
</tr>
<tr>
<td>Psoriasis</td>
<td>4,577</td>
<td>0.304</td>
<td>0.455</td>
</tr>
<tr>
<td>Reiter’s syndrome</td>
<td>136</td>
<td>0.009</td>
<td>0.014</td>
</tr>
<tr>
<td>Renal glycosuria</td>
<td>29</td>
<td>0.002</td>
<td>0.003</td>
</tr>
<tr>
<td>Reticulosarcoma</td>
<td>611</td>
<td>0.041</td>
<td>0.061</td>
</tr>
<tr>
<td>Rheumatoid arthritis</td>
<td>7,333</td>
<td>0.487</td>
<td>0.729</td>
</tr>
<tr>
<td>Systemic lupus erythematosus</td>
<td>3,194</td>
<td>0.212</td>
<td>0.318</td>
</tr>
<tr>
<td>Salmonella</td>
<td>695</td>
<td>0.046</td>
<td>0.069</td>
</tr>
<tr>
<td>Schilder’s syndrome</td>
<td>13</td>
<td>0.001</td>
<td>0.001</td>
</tr>
<tr>
<td>Scleroderma</td>
<td>11,256</td>
<td>0.747</td>
<td>1.119</td>
</tr>
<tr>
<td>Scleroderma</td>
<td>680</td>
<td>0.045</td>
<td>0.068</td>
</tr>
<tr>
<td>Shigella</td>
<td>6,315</td>
<td>0.419</td>
<td>0.628</td>
</tr>
<tr>
<td>Sjogren’s syndrome</td>
<td>348</td>
<td>0.023</td>
<td>0.035</td>
</tr>
<tr>
<td>Spina bifida occulta</td>
<td>40</td>
<td>0.003</td>
<td>0.004</td>
</tr>
<tr>
<td>Spondylolisthesis</td>
<td>1,024</td>
<td>0.068</td>
<td>0.102</td>
</tr>
<tr>
<td>Staphilococcus</td>
<td>23,592</td>
<td>1.567</td>
<td>2.346</td>
</tr>
<tr>
<td>Streptococcus</td>
<td>27,682</td>
<td>1.838</td>
<td>2.752</td>
</tr>
</tbody>
</table>

Table 3: Disorders, record counts, and the raw and adjusted prevalence.
<table>
<thead>
<tr>
<th>Disorder</th>
<th>Number of records</th>
<th>Raw %</th>
<th>Adjusted %</th>
</tr>
</thead>
<tbody>
<tr>
<td>Takayasu’s arteritis</td>
<td>74</td>
<td>0.005</td>
<td>0.007</td>
</tr>
<tr>
<td>Thrombotic thrombocytopenic purpura</td>
<td>112</td>
<td>0.007</td>
<td>0.011</td>
</tr>
<tr>
<td>Tuberculosis</td>
<td>66,569</td>
<td>4.421</td>
<td>6.618</td>
</tr>
<tr>
<td>Tuberculosis</td>
<td>202</td>
<td>0.013</td>
<td>0.020</td>
</tr>
<tr>
<td>Tuberous sclerosis</td>
<td>256</td>
<td>0.017</td>
<td>0.025</td>
</tr>
<tr>
<td>Typhoid</td>
<td>54</td>
<td>0.004</td>
<td>0.005</td>
</tr>
<tr>
<td>Urca cycle metabolism</td>
<td>74</td>
<td>0.005</td>
<td>0.007</td>
</tr>
<tr>
<td>Virus</td>
<td>135,833</td>
<td>9.021</td>
<td>13.505</td>
</tr>
<tr>
<td>Vitamin deficiency</td>
<td>776</td>
<td>0.052</td>
<td>0.077</td>
</tr>
<tr>
<td>WPW syndrome</td>
<td>739</td>
<td>0.049</td>
<td>0.073</td>
</tr>
<tr>
<td>Wegener’s granulomatosis</td>
<td>191</td>
<td>0.013</td>
<td>0.019</td>
</tr>
</tbody>
</table>

Table 4: Disorders, record counts, and the raw and adjusted prevalence.