

# Supporting Information

Robinson et al. 10.1073/pnas.1409204111

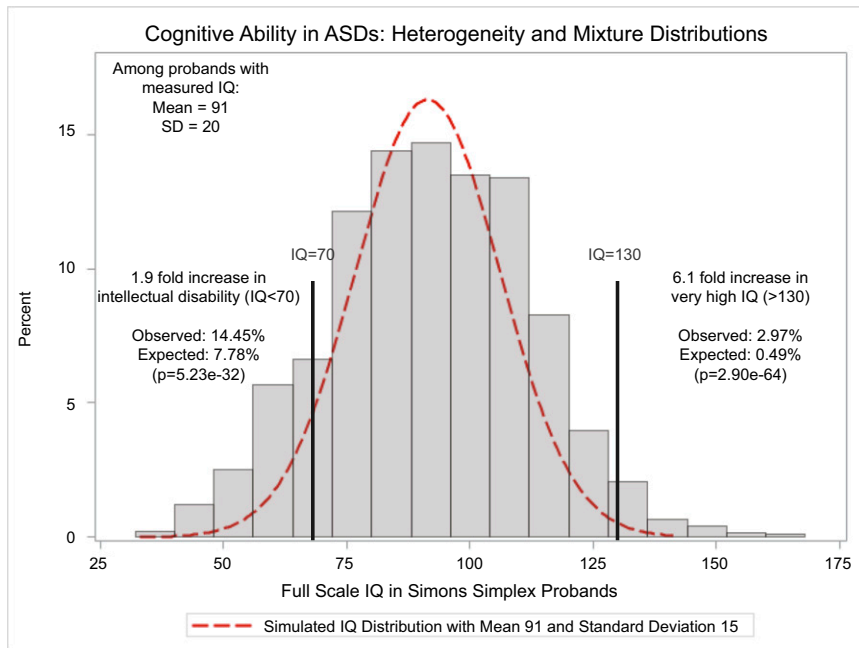


Fig. S1. Cognitive ability in autism spectrum disorders: heterogeneity and mixture distributions. Gray bars depict observed distribution of measured full-scale intelligence quotient (IQ) in the Simons Simplex Collection ( $n = 2,256$ ).

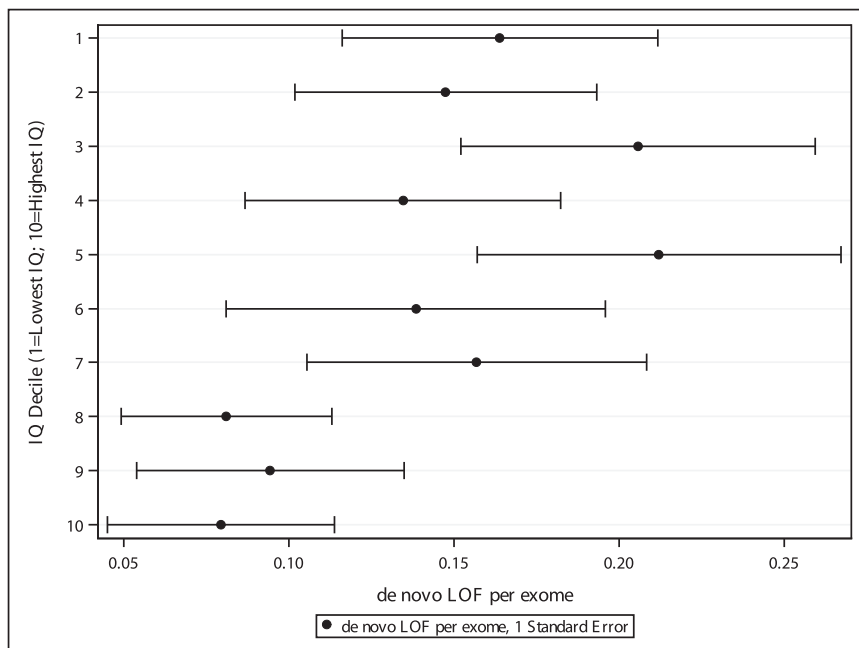


Fig. S2. Association between proband IQ decile and de novo loss of function (LOF) per exome.



**Table S1. Measures**

Construct	Measures	Subdomains/example items
Intelligence	(i) WISC-IV ( <i>n</i> = 48) (1); (ii) WASI ( <i>n</i> = 117) (2); (iii) DAS Early Years ( <i>n</i> = 1,123); DAS School Age ( <i>n</i> = 1,318) (3)	Summary scores (e.g., full scale, verbal, nonverbal IQ) were provided by the Simons Simplex Collection. Full-scale IQ was used in the primary analyses.
Behavior	Vineland Scales of Adaptive Behavior (4)	Subscales: social ability, communication ability, daily living skills ability
Language	Autism Diagnostic Interview-Revised (5)	—

1. Weschler D (2003) *Weschler Intelligence Scale for Children—4th Edition (WISC-IV)* (Harcourt Assessment, San Antonio, TX).
2. Weschler D (1997) *Weschler Abbreviated Scale of Intelligence (WASI)* (Harcourt Assessment, San Antonio, TX).
3. Elliott CD (1990) *DAS Administration and Scoring Manual* (The Psychological Corporation, San Antonio, TX).
4. Sparrow SS, Cicchetti DV, Balla DA (2005) *Vineland Adaptive Behavior Scales* (Pearson, Livonia, MN), 2nd Ed.
5. Le Couteur A, Lord C, Rutter M (2003) *Autism Diagnostic Interview—Revised (ADI-R)* (Western Psychological Services, Portland, OR).

**Table S2. Variation in case severity between IQ attempted but unscored group and individuals with measured intellectual disabilities (IQ <70 or IQ <55)**

Parameter	IQ attempted but unscored compared with IQ <70	IQ attempted but unscored compared with IQ <55
Vineland standard composite score	IAU ( <i>n</i> = 349), mean 61.06 (10.01) IQ <70 ( <i>n</i> = 326), mean 67.76 (7.75) <i>t</i> = 9.75, <i>df</i> = 650.87, <i>P</i> = 4.52E-21	IAU ( <i>n</i> = 349), mean 61.06 (10.01) IQ <55 ( <i>n</i> = 76), mean 64.00 (6.69) <i>t</i> = 3.14, <i>df</i> = 157.92, <i>P</i> = 0.002

Age at phrase speech development assessed in months. IAU, IQ attempted but unscored. Values in parentheses following the mean indicate standard deviation.

**Table S3. Association between *P*(LOF) and language/behavioral measures after IQ is regressed out**

Measure	<i>B</i>	<i>P</i>
Vineland Communication Standard Score	0.012	0.320
Vineland Daily Living Standard Score	−0.010	0.326
Vineland Social Standard Score	−0.002	0.851
Age at phrase speech development (ADI)	−0.002	0.820

All measures are normed and transformed as described for Fig. S3. ADI, Autism Diagnostic Interview.