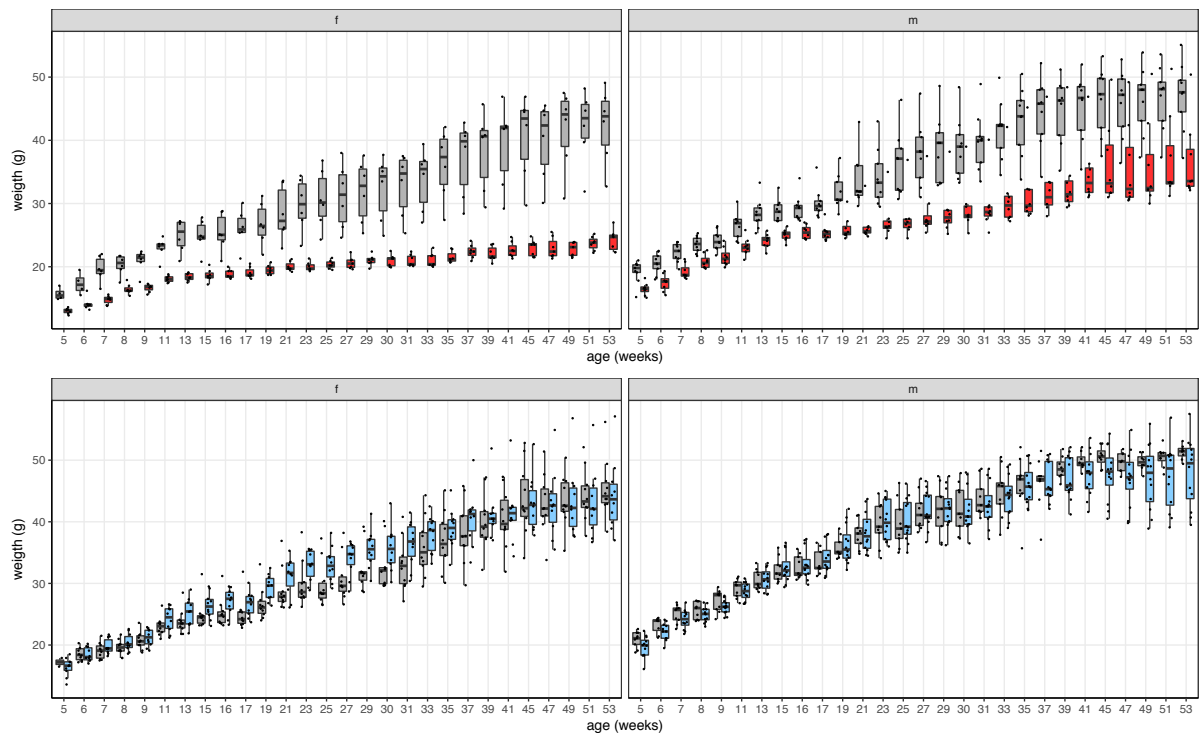


Supplemental Information

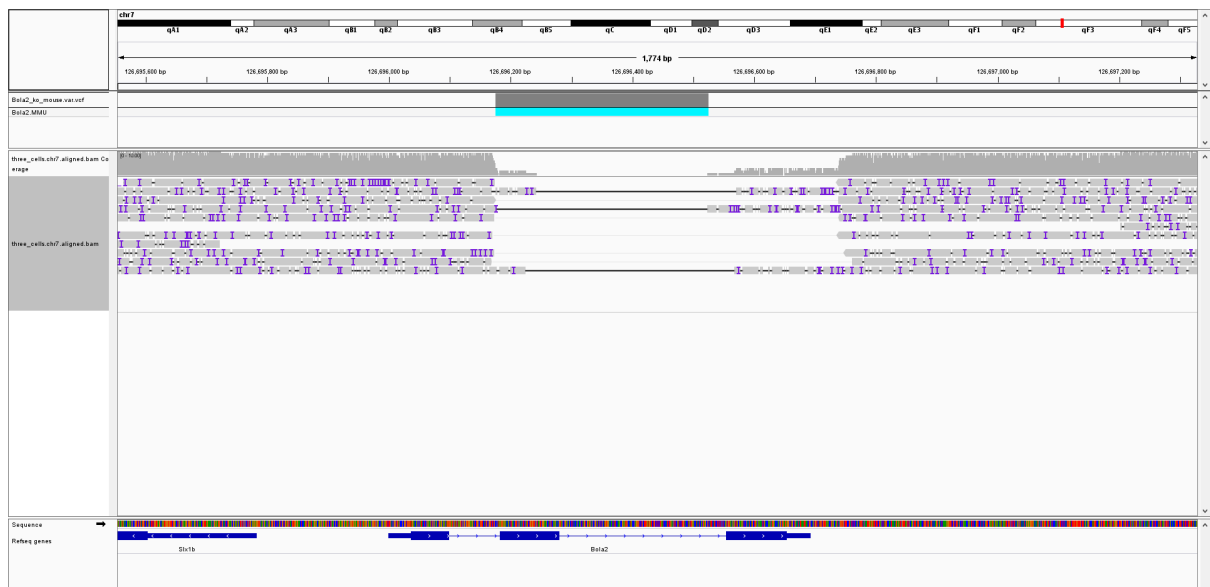
The human-specific *BOLA2* duplication modifies iron homeostasis and anemia predisposition in chromosome 16p11.2 autism patients

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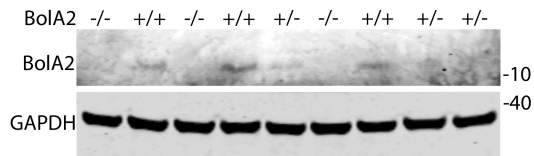
Supplemental Figures



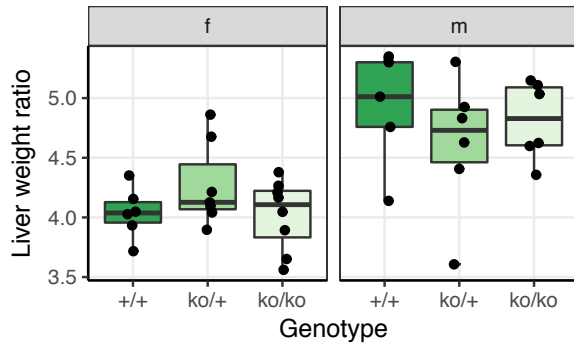
Supplemental Figure S1. Longitudinal body weight profiles of 16p11.2^{Del/+} (top) and 16p11.2^{Dup/+} (bottom) female (left) and male (right) mouse models and wild-type littermates. The genotype color code is: Del/+ (red), Dup/+ (light blue), wild-type (grey). Note that the x-axis scale is not continuous.



Supplemental Figure S2. SMRT sequencing platform reads of a neomycin cassette-excised *Bola2*^{-/-} male were aligned to mm10. Within the *Bola2* locus on Chromosome 7 shown here please note the nine spanning reads, all indicating the deletion. PBSV calls a homozygous deletion of 351 bp (cyan box).



Supplemental Figure S3. Western blot of liver samples from *Bola2*^{+/+}, *Bola2*^{+/-} and *Bola2*^{-/-} mice with an anti-BOLA2 antibody performed as described in [17].



Supplemental Figure S4. Liver weight normalized to body weight (mg/g) of *Bola2*^{-/-}, *Bola2*^{+/-} and wild-type littermates from the Swiss cohort at 31 weeks of age.

Supplemental Tables

Supplemental Table S1. Hematological parameters of 16p11.2 CNV carriers and controls of the UK Biobank.

Supplemental Table S2. *BOLA2* copy number and anemia status of 16p11.2 deletion carriers from the SVIP cohort.

Supplemental Table S3. Hematological and blood iron quantifications of 16p11.2 deletion carriers from the European cohort with genotyped *BOLA2* copy number. Values in red are below the minimum value of the reference range. Individuals not considered in the statistics are grey-shaded (related family members).

Supplemental Table S4. Heparin-plasma iron level of 16p11.2^{Del/+} and 16p11.2^{Dup/+} mouse models and their wild-type littermates.

Supplemental Table S5. Hematological parameters of 16p11.2^{Del/+} mouse models and their wild-type littermates.

Supplemental Table S6. Hematological parameters of 16p11.2^{Dup/+} mouse models and their wild-type littermates.

Supplemental Table S7. Heparin-plasma iron level of *Bola2*^{+/+}, *Bola2*^{+/-}, and *Bola2*^{-/-} mice (European cohort).

Supplemental Table S8. Hematological parameters of *Bola2*^{+/+}, *Bola2*^{+/-}, and *Bola2*^{-/-} mice (European cohort).

Supplemental Table S9. Serum iron, TIBC, Tf sat, ZPP, liver and spleen iron levels of *Bola2*^{+/+}, *Bola2*^{+/-}, and *Bola2*^{-/-} mice (neo-in mice, US cohort).

Supplemental Table S10. Hematological parameters of *Bola2*^{+/+}, *Bola2*^{+/-}, and *Bola2*^{-/-} mice (neo-in mice, US cohort).

Supplemental Table S11. Serum iron, TIBC, Tf sat, and ZPP levels of *Bola2*^{+/+} and *Bola2*^{-/-} mice (neo-excised mice, US cohort).

Supplemental Table S12. Hematological parameters of *Bola2*^{+/+} and *Bola2*^{-/-} mice (neo-excised mice, US cohort).

Supplemental Table S13. Normal hemoglobin and iron levels in the blood of adult humans, great apes, and mice.

Supplemental Table S14. Members of the 16p11.2 Consortium.