Supplementary Information

***CA2*:** We identified a female individual with ASD and a homozygous canonical splice site mutation (chr8:86377699G>A; IVS2+1G>A) in intron 2 of the *CA2* gene, encoding carbonic anhydrase II. Her parents were of Middle Eastern descent and known to be first cousins. Loss of function mutations in *CA2* cause a rare autosomal recessive syndrome of intellectual disability with osteopetrosis and renal tubular acidosis, with variable intellectual disability (Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, MIM #259730). The allele in this patient is recognized as a clinically pathogenic variant[1] (ClinVar RCV000000966.2). In addition to ASD, this individual was noted to have osteopetrosis, mild corpus callosal hypogenesis, congenital hypothyroidism, and failure to thrive, which together match reported phenotypes for carbonic anhydrase II deficiency syndrome.

***DDHD1*:** We identified a male individual with ASD with compound heterozygosity for two canonical splice site mutations (exon6:c.1311-2A>T and exon13:c.2459-2A>T in the *DDHD1* gene, encoding a brain-expressed phosphatidic acid phospholipase involved in lipid metabolism and mitochondrial function. Recessive loss-of-function mutations in *DDHD1* have been reported to cause hereditary spastic paraplegia (Spastic paraplegia 28, autosomal recessive, MIM #609340) in five pedigrees.[2-4] Clinical features include progressive spastic gait with or without other neurologic complications, such as cerebellar impairment, axonal neuropathy, distal sensory loss, and/or mitochondrial impairment. To date, ASD and cognitive disturbances have not been described, although recessive loss-of-function mutations in the closely related phospholipase *DDHD2* cause spastic paraplegia, intellectual disability, short stature, and dysgenesis of the corpus callosum.[5] The patient was originally recruited through the Autism Genetic Resource Exchange (AGRE). No additional clinical details were available.

***NSUN2*:** A homozygous stop-gain mutation (c.C1708T:p.Q570X) in *NSUN2* was identified in a girl with ASD and ID. Her parents were of Turkish descent and were first cousins once removed. *NSUN2* encodes a 767 amino acid methyltransferase that is responsible for a critical enzymatic modification of the first position of the anticodon of tRNA-leu(CAA) to 5-methylcytosine. This modification is essential for proper anticodon pairing of tRNAs and the translation of mRNA. *NSUN2* is highly expressed during brain development, and mutations in *NSUN2* have been shown to cause autosomal recessive intellectual disability in several families (Autosomal Recessive Mental Retardation 5, MIM #611091), with autistic features reported in at least one family.[6]

***PAH*:** We re-identified homozygous nonsense mutations in *PAH* (Phenylketonuria, MIM #261600) in two unrelated ASD individuals from consanguineous families. These findings were previously described.[7]

***RARB*:** One male individual with ASD carried a homozygous p.C26X nonsense mutation in *RARB*. He was of Turkish descent and his parents were known to be consanguineous. *RARB* encodes a retinoic acid receptor downstream of the *FOXP2* transcription factor, and recessive LOF mutations (R119X and frameshift I403Sfs15X) and dominant *de novo* GOF mutations (R387C and R387S) have been recently described in patients with PDAC syndrome (microphthalmia, pulmonary hypoplasia, diaphragmatic hernia, and cardiac defects, Microphthalmia, syndromic 12, MIM #615524). All but one reported case ended in fetal demise, but one patient (with a *de novo* R387C mutation) survived to age 16 and suffered from intellectual disability (Srour et al, *Am J Hum Genet* 2013). The patient with a *de novo* R387 mutation lacked full features of PDAC syndrome, exhibiting microophthlamia and diaphragmatic hernia but no pulmonary or cardiac defects, raising the possibility of an expanded phenotypic spectrum. The ASC individual had, in addition to ASD, bilateral microphthalmia, and a brain MRI demonstrating hypoplastic optic nerves and optic chiasm.

***RFX5*:** We identified a female individual with ASD who carried homozygous knockout mutations in *RFX5*, a known cause of bare lymphocyte syndrome, a recessively inherited severe primary immunodeficiency associated with failure of major histocompatibility class II gene expression[8] (MIM #209920). The patient was of Middle Eastern ancestry and parents were second cousins. No additional clinical details regarding this ASC patient were available.

***ROGDI*:** We found a homozygous splice-site mutation in *ROGDI* in a male individual with ASD, ID, and epilepsy. This phenotype was consistent with known recessive mutations in *ROGDI* which cause Kohlschutter-Tonz syndrome (MIM #226750), a recessively inherited brain disorder which is marked by early-onset intractable seizures, severe global developmental delay, spasticity, and amelogenesis imperfecta[9,10]. This individual was of Turkish ancestry, and parents were first cousins.

***SLC1A1*:** As described in the main text, we identified an autistic individual from Germany, bearing a homozygous LOF mutation in the gene *SLC1A1* (Dicarboxylic aminoaciduria, MIM #222730). This affected individual bore a homozygous c.G142T:p.E48X change that was inherited from heterozygous parents. No further clinical details were available.

***USH2A*:** We identified a homozygous nonsense mutation in *USH2A* (Usher syndrome, type 2A, MIM #276901) in one individual. These findings were previously described[11] and the individual was found to suffer from hearing loss.

***FEV*:** As described in the main text, homozygous nonsense mutations in *FEV* (p.E89X) were found in two siblings. Parents were second cousins, and mother had a history of stillbirths. The elder brother was diagnosed clinically with autism and intellectual disability (IQ 69). He suffered

from severe stereotyped, aggressive and self-injurious behaviors. He had no history of seizures. At age 10, head circumference was 55 cm (73.5%), height was 134 cm (11%), weight 46 kg (92%) with a BMI of 25.6 (99%). Exam was notable for a squarish face, high forehead, slight hypertelorism, epicanthal folds, short philtrum, broad lower lip, small teeth, prominent, low-set ears, skin hyperpigmentation, 5th finger clinodactyly of both hands, and brachydactyly of his toes. An EEG showed rolandic focus bilaterally with generalization and EKG and cardiac ultrasound were normal. The younger brother carried a clinical diagnosis of PDD-NOS and intellectual disability (IQ not measured). Head circumference at age 4 was 51 cm (29.8%), height was 101 cm (24%), weight was 18 kg (70%), and BMI was 17.7 (91%). He exhibited muscular hypotonia. Skin showed general hypopigmentation but he had two café-au-lait patches as well. No abnormal facial features were noted. An EEG was normal, as were EKG and cardiac ultrasound studies. Both brothers scored in the Autism range on ADOS testing. Vineland Adaptive Behavior Composite scores were 39 and 69, respectively.

Analysis of biallelic mutations of established medical relevance

We examined how often these biallelic missense mutations involved alleles established in ClinVar as Pathogenic or Likely Pathogenic. Of 5,852 unaffected individuals, only three harbored biallelic damaging missense mutations meeting these criteria: one unaffected individual with a homozygous p.E99Q variant in *MCCC2*, and two unaffected individuals with homozygous p.A300S mutations in *PAH.* Both *MCCC2* p.E99Q and *PAH* p.A300S are notable, despite being classified as Pathogenic, for exhibiting variable penetrance and expressivity, and both have been reported in neurotypical individuals before (**Table 3, Supplementary Table 13**). In contrast, of 2343 affected individuals, thirteen had biallelic damaging mutations with Pathogenic/Likely Pathogenic alleles in ClinVar (**Table 3, Supplementary Table 13**). All thirteen cases were diagnostic of genetic disorders with known neurodevelopmental consequences including classic metabolic diseases, mitochondrial depletion syndrome, and other syndromic conditions. These

thirteen cases, combined with the previously described eight cases involving biallelic loss of function mutations, constituted ~1% of our cohort (21 out of 2,343 affected individuals), underscoring the importance of clinical screening for monogenic recessive conditions in this patient population.

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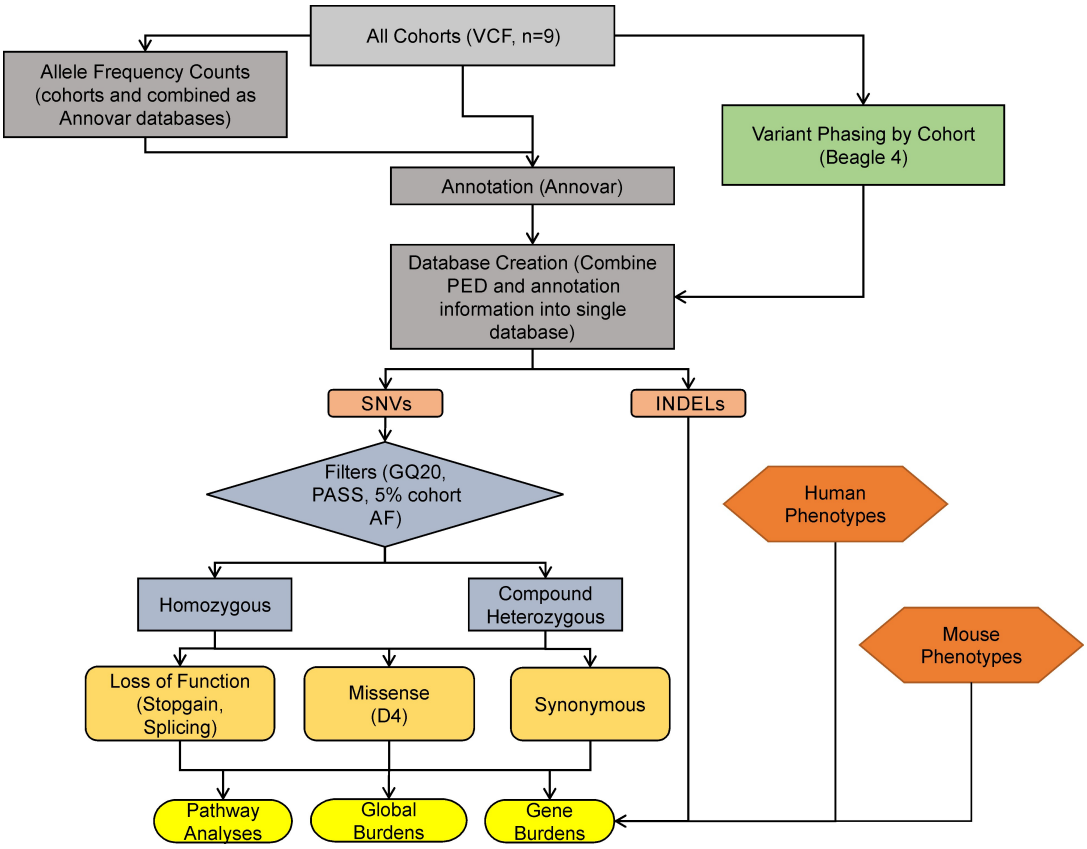
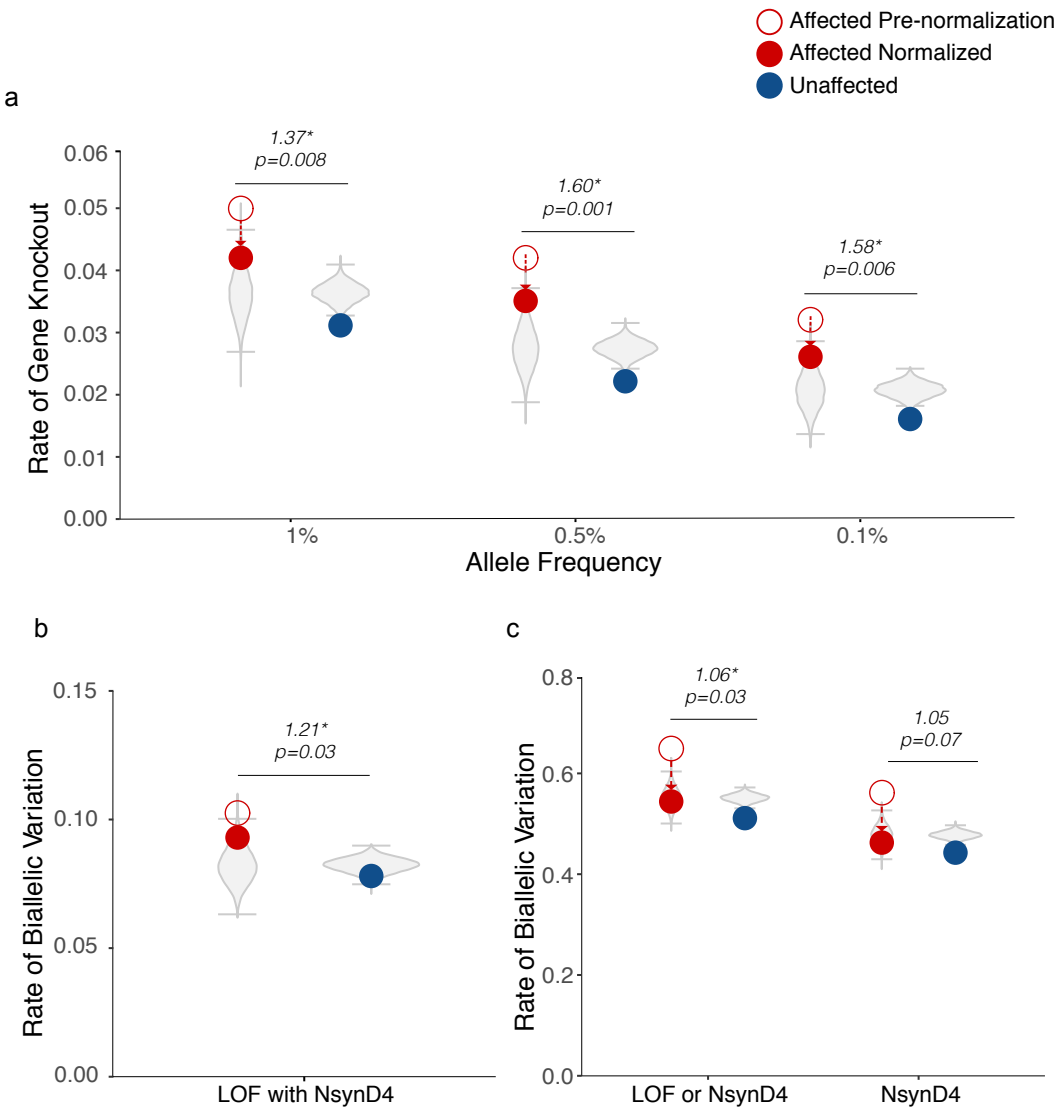
LIST OF SUPPLEMENTARY TABLES

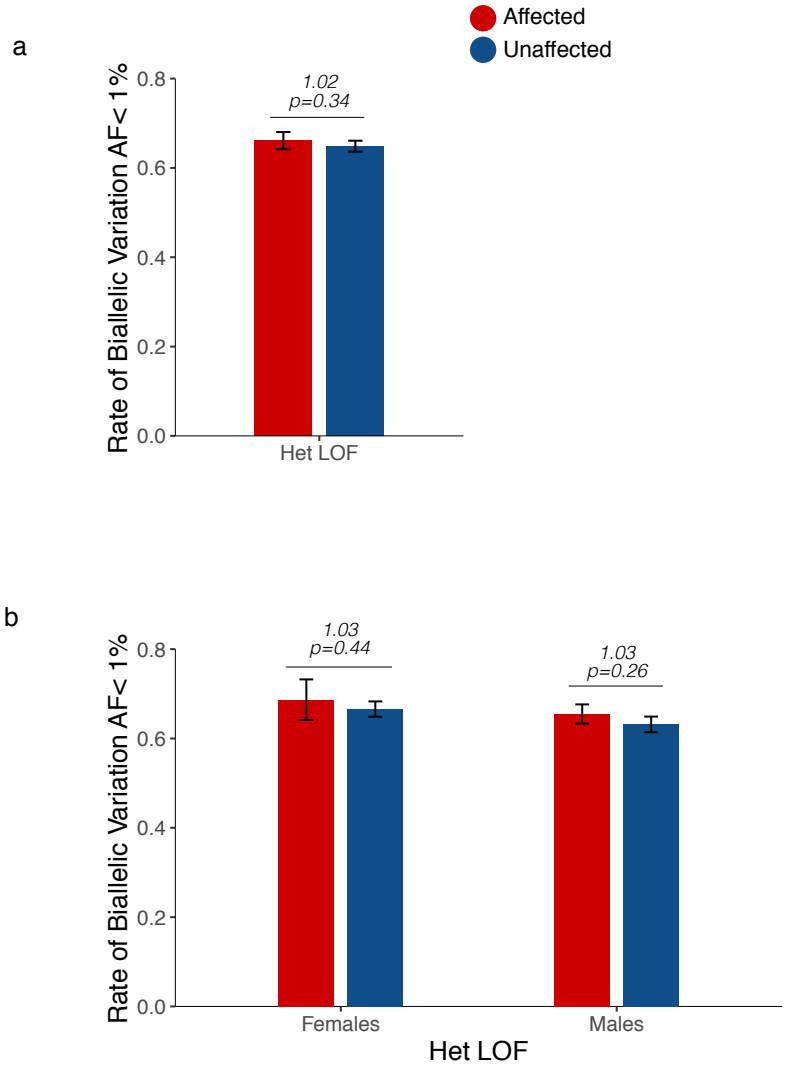
SUPPLEMENTARY FIGURES



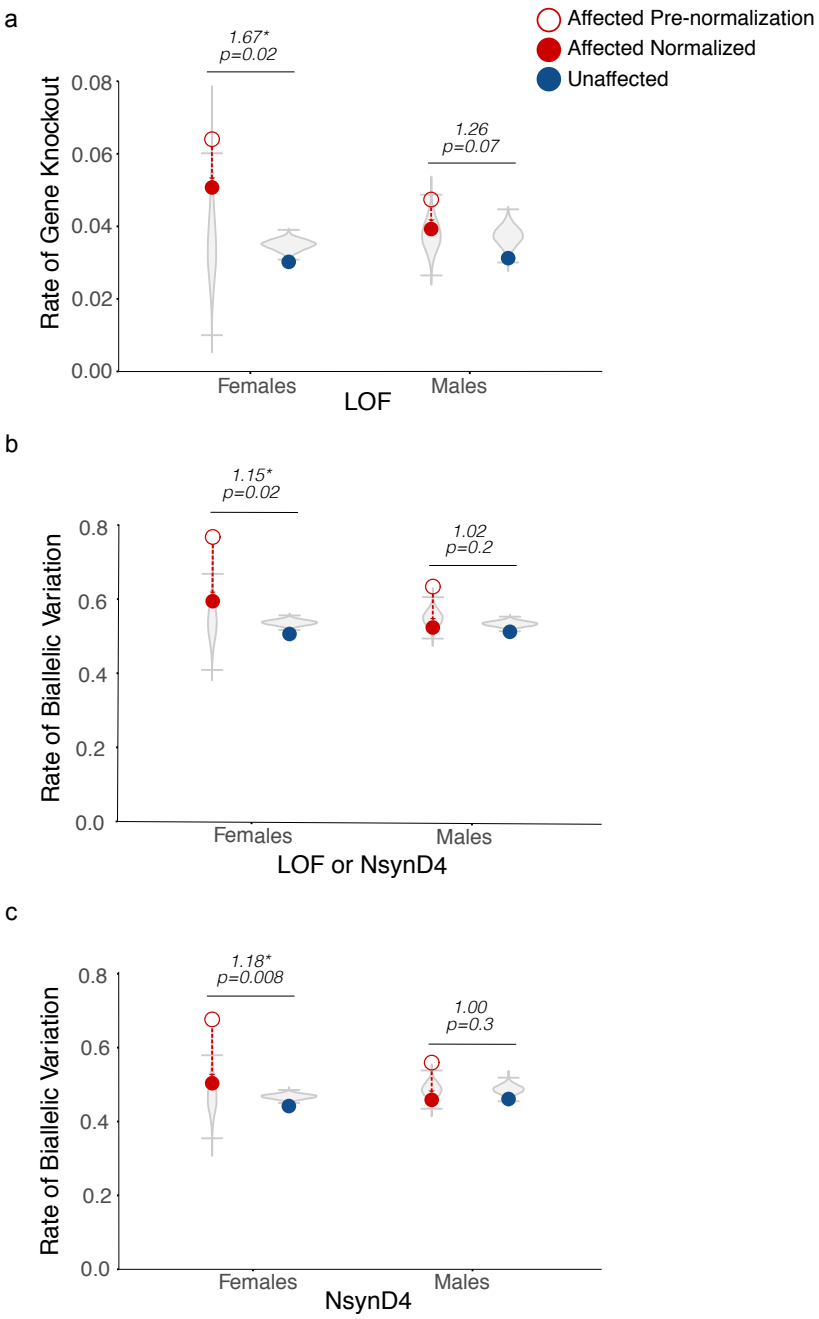
Supplementary Figure 1. Analytical pipeline for potentially damaging biallelic variations in the ASC



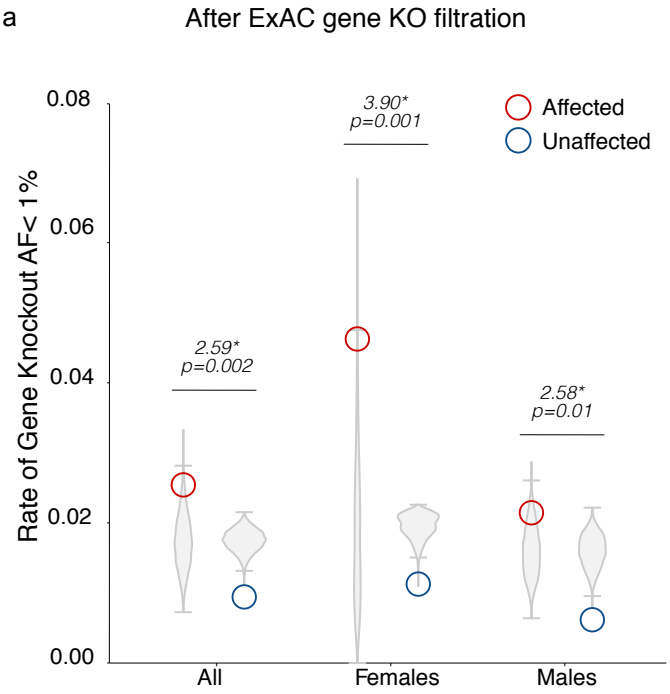
Supplementary Figure 2. Excess of biallelic mutation in ASD overlaid with distribution of rates assessed by random sampling of cases and controls. Solid dots represent rates post-normalization for potential effects of population stratification (**Methods**) while open circles represent actual measured rates prior to normalization. Distribution of rates from 10,000 random assignments of affected status displayed as violin plots in gray. Statistical testing was performed on the post-normalization rates. **(a)** Rates of biallelic gene knockout (strict LOF) in the ASC, stratified by diagnosis and allele frequency. Rates of biallelic variation, considering **(b)** LOF variants paired with a damaging missense variant (NsynD4, predicted to be deleterious by at least 4 algorithms), **(c)** LOF or NsynD4 variants, or NsynD4 variants alone.



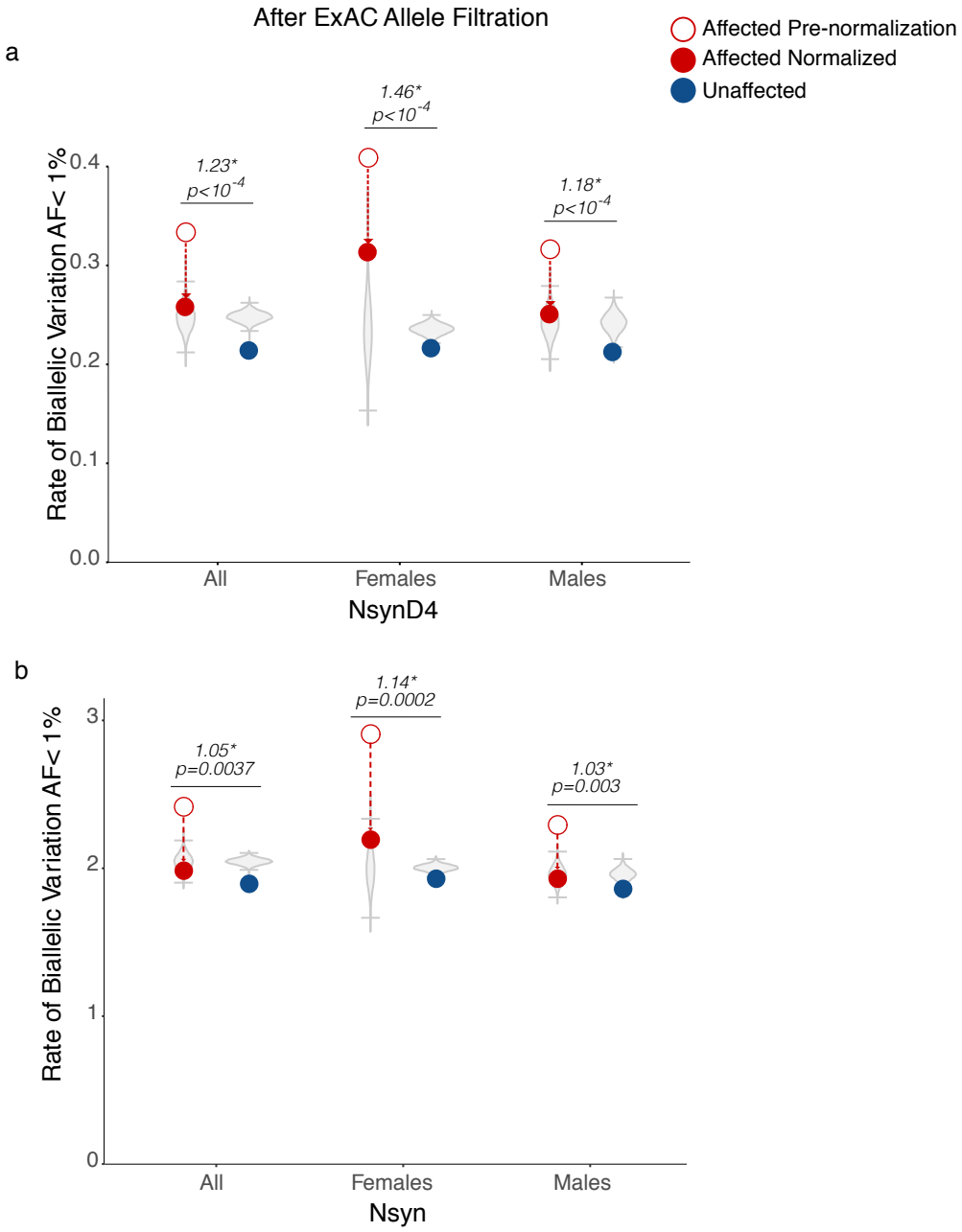
Supplementary Figure 3. Alleles that contribute to rare biallelic LOF events are not found in excess in the heterozygous / monoallelic state, either (a) in all samples from the ASC, or (b) when stratified by sex. This is consistent with a recessive model of disease risk (as opposed to an additive dominant/codominant model). We estimate that given our sample sizes, we are powered to detect an effect size of ~OR1.1 (under a dominant model) for this class of variant; e.g., these results suggest that if there is categorical risk for these variants under an additive/dominant model, the effect size is <1.1. Error bars represent the 95% confidence intervals.



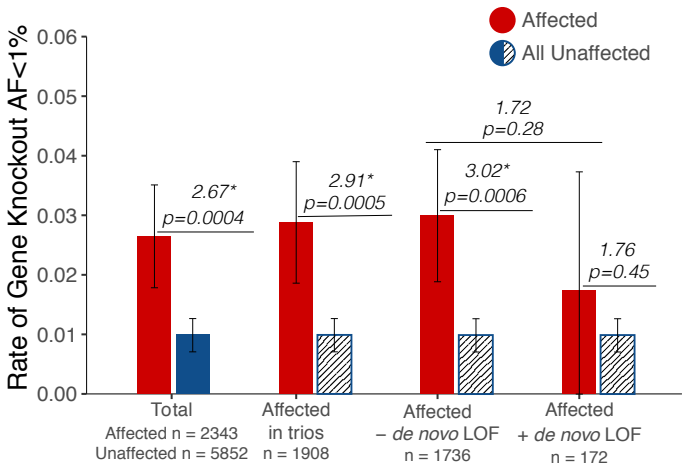
Supplementary Figure 4. Effects of sex on excess of biallelic mutation in ASD overlaid with distribution of rates assessed by random sampling of cases and controls. Solid dots represent rates post-normalization for potential effects population stratification while open circles represent actual measured rates prior to normalization. Distribution of rates from 10,000 random assignments of affected status displayed as violin plots in gray. Statistical testing was performed on the post-normalization rates. **(a)** Rates of biallelic gene knockout (strict LOF) in the ASC, stratified by diagnosis and sex. **(b, c**) Rates of biallelic variation stratified by gender, considering **(b)** LOF or damaging missense (LOF or NsynD4) variants or **(c)** damaging missense variants alone (NsynD4).



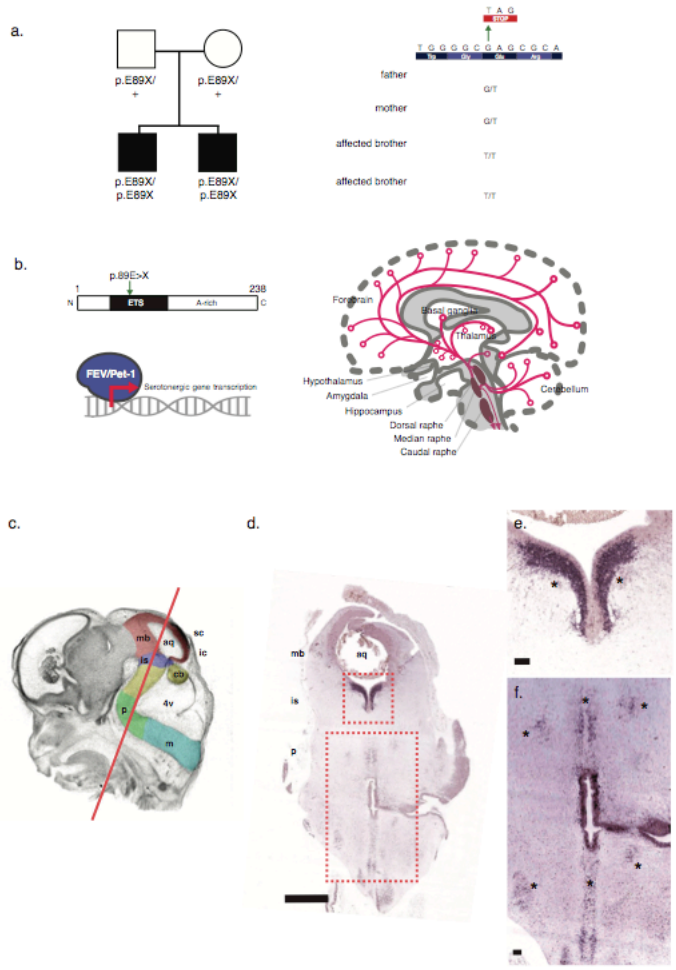
Supplementary Figure 5. Comparison of excess biallelic gene knockout (strict LOF) after filtration of commonly inactivated genes in ExAC with rates from random sampling. Solid dots represent rates post-normalization for potential effects of population stratification while open circles represent actual measured rates prior to normalization. Distribution of rates from 10,000 random assignments of affected status displayed as violin plots in gray.



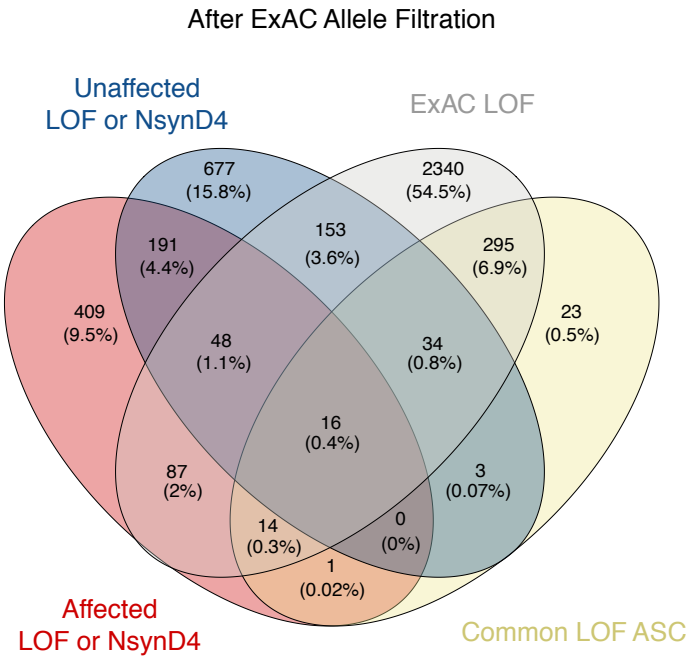
Supplementary Figure 6. Reduction of background rates through filtration of homozygous alleles in ExAC **(a)** increases excess of rare biallelic damaging missense (NsynD4) mutation in sex-stratified individuals with ASD and **(b)** allows or small detectable enrichment for all missense alleles (Nysn). Solid dots represent rates post-normalization for potential effects population stratification while open circles represent actual measured rates prior to normalization. Distribution of rates from 10,000 random assignments of affected status displayed as violin plots in gray. Statistical testing was performed on the post-normalization rates.



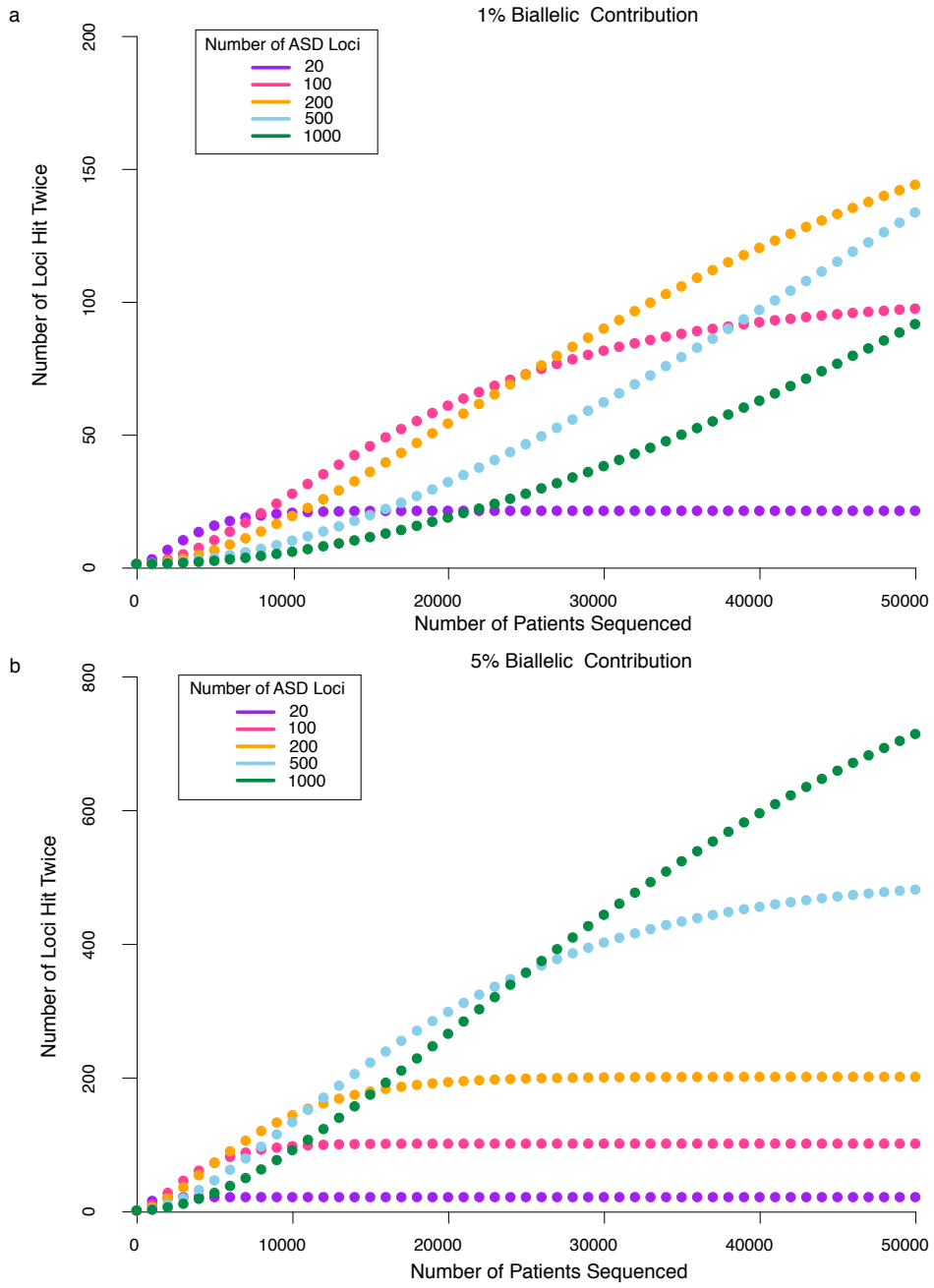
Supplementary Figure 7: Comparison of burdens of biallelic LOF mutations in the ASC following ExAC knockout filtering, stratified by the presence or absence of detectable *de novo* LOF mutations as reported in de Rubeis *et al*, 2014. Each comparison is against all unaffected individuals in the ASC. Error bars represent the 95% confidence intervals.



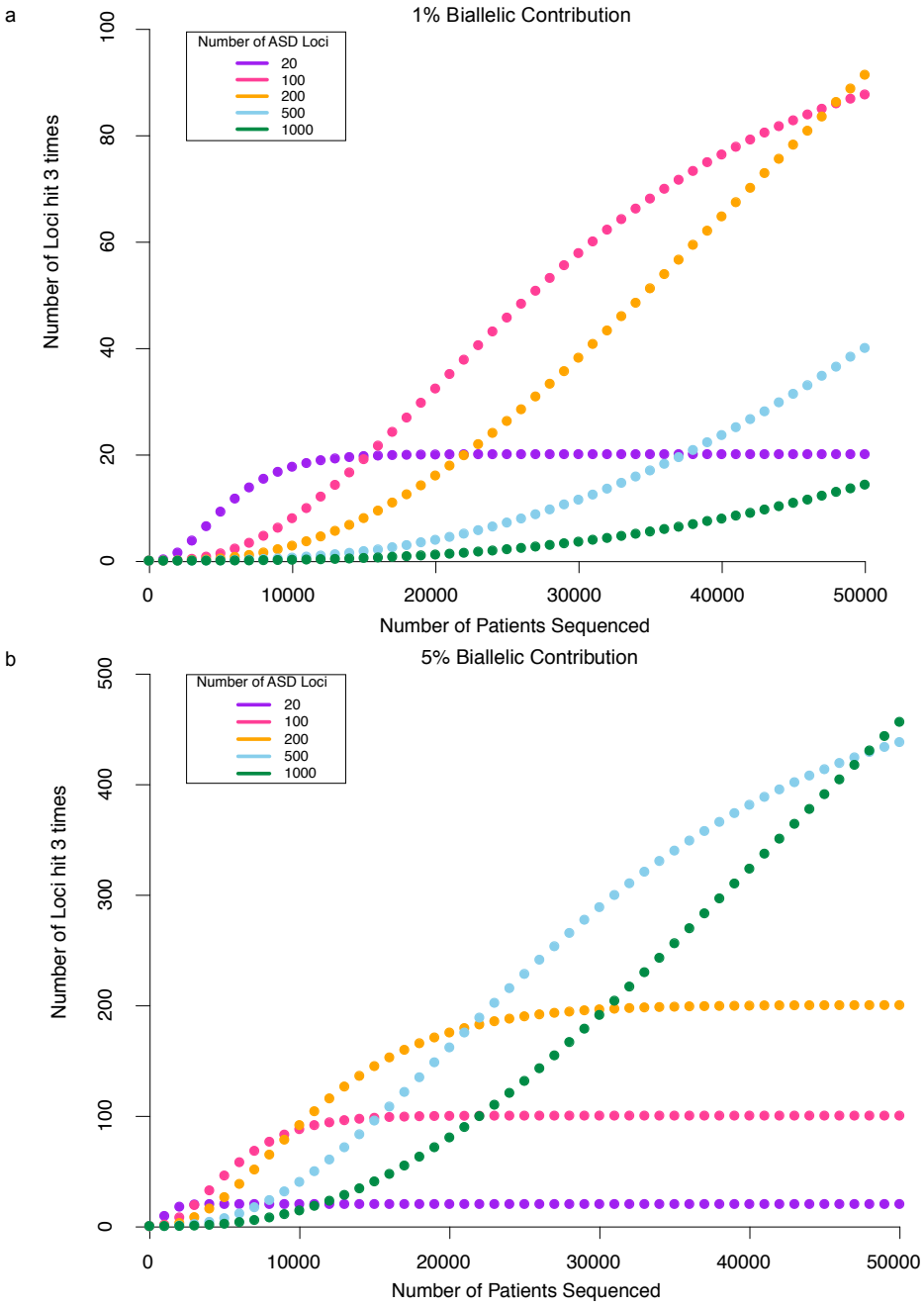
Supplementary Figure 8: Knockout of the serotonergic transcription factor *FEV*/Pet-1 in two brothers with autism from the ASC. **(a)** Family pedigree and sequencing results showing two children with ASD bearing biallelic stopgain mutations in *FEV*. **(b)** The stopgain mutation in *FEV*/Pet-1 lies in a highly conserved ETS DNA-binding domain. *FEV* is thought to be responsible for transcriptional regulation of serotonergic genes in the serotonergic raphe neurons, which send projections broadly throughout the human brain to regulate emotion, cognition, and motor activity. **(c-g)** *In situ* hybridization analysis demonstrates specific expression of *FEV* in developing serotonergic raphe nuclei in the developing human embryonic brain (Carnegie Stage 23). Coronal plane of section is illustrated in (**c**). **(d)** 252-bp sense *FEV* probe (negative control). (**e**) 252-bp antisense *FEV* probe (corresponding to NM_017521 nt. 1453-1704). Dotted boxes indicate regions presented at higher magnification in (**f**) and (**g**), with *FEV* staining in clusters of developing serotonergic raphe nuclei (asterisks). Intense staining in the isthmus denotes expression in the developing dorsal raphe nucleus, the largest of the serotonergic nuclei. Approximate levels corresponding to midbrain, isthmus, and pons are as indicated. Abbreviations: *4v*, 4th ventricle; *aq*, aqueduct; *cb,* cerebellum; *ic,* inferior colliculus; *is,* isthmus; *m,* medulla; *mb,* midbrain, *p,* pons; *sc,* superior colliculus. Scale bars: (**d**,**e**), 1mm; (**f**,**g**), 100µm.



Supplementary Figure 9. Candidate ASD genes impacted by biallelic LOF or NsynD4 mutation identified by exclusion of genes with knockouts in ExAC or the ASC, and those with similar mutations in unaffected individuals of the ASC.



Supplementary Figure 10: Increased power to detect novel genes with at least 2 independent mutations through increased sample size. Power calculations demonstrating the ability to detect genes with biallelic events in at least 2 families assuming **a)** a 1% biallelic contribution similar to our LOF estimates and b) a 5% biallelic contribution similar to our missense estimates.



Supplementary Figure 11: Increased power to detect novel genes with at least 3 independent mutations through increased sample size. Power calculations demonstrating the ability to detect genes with biallelic events in at least 3 families assuming **a)** a 1% biallelic contribution similar to our LOF estimates and b) a 5% biallelic contribution similar to our missense estimates.

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