

| SFARI Score | Gene Symbol | Description | Log2 Fold-Change | Adjusted P-value |
|-------------|-------------|---|------------------|------------------|
| S | AHI1 | Joubert Syndrome, role in schizophrenia | 0.109 | 0.0359 |
| S | BRAF | cardiofaciocutaneous syndrome with frequent autistic features like social impairment, internalizing/externalizing problems | 0.100 | 0.0409 |
| S | CACNA1C | Timothy Syndrome, no evidence in idiopathic autism | 0.143 | 0.0187 |
| S | DMD | Muscular dystrophy with reported symptoms of autism. Little evidence in idiopathic autism. | 0.096 | 0.0112 |
| S | MECP2 | Mutations of MECP2 causes Rett syndrome, associated with ASD (DSM-IV). Autism and neuropsychiatric symptoms prevalent in MECP2 duplication. | 0.097 | 0.0253 |
| S | NF1 | Neurofibromatosis is a Mendelian disease associated with autism. | 0.112 | 0.0488 |
| S | NFIX | Sotos syndrome 2 (Malan syndrome) and Marshall-Smith Syndrome | 0.161 | 0.0024 |
| S | OCRL | Lowe Syndrome, X-linked multisystem disorder, maladaptive behaviors | 0.084 | 0.0166 |
| S | PACS1 | Syndromic ID from de novo missense variant in PACS1 presented with ASD or showed behavior associated with ASD | 0.095 | 0.0164 |
| S | TCF4 | Pitt-Hopkins Syndrome, ID, severe communication and language impairment, severe social interaction impairment, intronic SNP associated with schizophrenia | -0.127 | 0.0244 |
| S | TSC1 | Mutations in TSC1 cause autosomal dominant tuberous sclerosis (TSC). 25% of TSC individuals have autism | 0.104 | 0.0005 |
| 1S | ARID1B | Associated with Coffin-Siris Syndrome, ASC candidate gene for autism (FDR < 0.01). | 0.080 | 0.0407 |
| 1S | CHD8 | Variants identified in ASD probands from SSC. ASC candidate gene for autism (FDR < 0.01). DD, dysmorphic facial features | 0.079 | 0.0358 |
| 1S | DYRK1A | Variants identified in ASD probands from SSC. ASC candidate gene for autism (FDR < 0.01). Identified with ASD, ID, microcephaly | 0.060 | 0.0205 |
| 1S | POGZ | Variants identified in ASD probands from SSC. ASC candidate gene for autism (FDR < 0.01). DD, ID, hypotonia, facial features | 0.126 | 0.0007 |
| 1S | SETD5 | Variants identified in ASD probands from SSC. ASC candidate gene for ASD (FDR < 0.1). ID | 0.111 | 0.0030 |
| 1S | SYNGAP1 | ASC candidate gene for ASD (FDR < 0.01). ID | 0.104 | 0.0432 |
| 2S | BCL11A | Variants identified in ASD proband from SSC and ASC. ASC candidate gene for ASD (FDR < 0.05). DD, ID | 0.097 | 0.0446 |
| 2S | DEAF1 | ID, motor delay, speech development, hyperactive behavior, compulsive behavior, aggressive behavior, mood swings, poor eye contact | 0.092 | 0.0433 |
| 2S | KMT2A | Wiedemann-Steiner Syndrome. Variants identified in ASD probands from ASC, SSC, and DDDS. ID, hypertrichosis cubiti, short stature, distinct facial | 0.149 | 0.0312 |
| 3S | AHDC1 | Xia-Gibbs Syndrome. Variants identified in ASD probands from SSC. ID, DD, expressive language delay, hypotonia, sleep apnea, dysmorphic features, brain abnormalities, failure to thrive, ataxia | 0.131 | 0.0489 |
| 3S | KAT6A | Variants identified in ASD probands from SSC. ID, epilepsy | 0.097 | 0.0331 |
| 3S | MBD5 | Mutations associated with ID, epilepsy, and ASD | 0.110 | 0.0058 |
| 3S | STXBP1 | Variants responsible for ID, early-onset epileptic encephalopathy. Some individuals present with ASD with or without ID/epilepsy. | 0.088 | 0.0173 |
| 4S | NR2F1 | Bosch-Boonstra-Schaaf optic atrophy syndrome. Variants identified in ASD probands from SSC and ASC. DD, ID, optic atrophy, hypotonia, seizures, repetitive behavior, ASD, thinning of corpus callosum, hearing defects. | 0.143 | 0.0015 |