PRENATALAUTISM

Elenco dei geni e delle malattie associate a disturbi dello spettro autistico

PrenatalAutism - list of genes screened and genetic diseases associated to ASD

Gene	Chromosome	Associated genetic disorder	Phenotype OMIM	Inheritance	OMIM gene	SFARI Gene
ABCA13	7	autism spectrum disorders			607807	https://gene.sfari.org/database/human-gene/ABCA13
ACTB	7	Baraitser-Winter neurodevelopmental syndrome 1	243310	Autosomal dominant	102630	https://gene.sfari.org/database/human-gene/ACTB
ACY1	3	Aminoacylase 1 deficiency	609924	Autosomal recessive	104620	https://gene.sfari.org/database/human-gene/ACY1
ADCY5	3	ADCY5-related dyskinesia; Familial dyskinesia with facial myokymia	606703	Autosomal dominant;	600293	https://gene.sfari.org/database/human-gene/ADCY5
ADNP	20	Helsmoortel-Van der Aa syndrome	615873	Autosomal dominant	611386	https://gene.sfari.org/database/human-gene/ADNP
ADSL	22	Adenylosuccinate lyase deficiency	103050	Autosomal recessive	608222	https://gene.sfari.org/database/human-gene/ADSL
AFF2	X	FRAXE intellectual disability	309548	X-linked	300806	https://gene.sfari.org/database/human-gene/AFF2
AGO1	1	Neurodevelopmental disorder with speech impairment and dysmorphism	620292	Autosomal dominant	606228	https://gene.sfari.org/database/human-gene/AGO1
AGO2	8	Lessel-Kreienkamp syndrome	619149	Autosomal dominant	606229	https://gene.sfari.org/database/human-gene/AGO2
AHI1	6	Joubert syndrome 3	608629	Autosomal recessive	608894	https://gene.sfari.org/database/human-gene/AHI1
ALDH5A1	6	Succinic semialdehyde dehydrogenase deficiency	271980	Autosomal recessive	610045	https://gene.sfari.org/database/human-gene/ALDH5A1
ALG6	1	Congenital disorder of glycosylation, type Ic	603147	Autosomal recessive	605947	https://gene.sfari.org/database/human-gene/ALG6
ANK2	4	Long QT syndrome 4 (Ankyrin-B syndrome)	600919	Autosomal dominant	600919	https://gene.sfari.org/database/human-gene/ANK2
ANK3	10	Intellectual developmental disorder, autosomal recessive 37	615493	Autosomal recessive	600465	https://gene.sfari.org/database/human-gene/ANK3
ANKRD11	16	KBG syndrome	148050	Autosomal dominant	611192	https://gene.sfari.org/database/human-gene/ANKRD11
AP1S2	X	X-linked intellectual disability, Fried type	300486	X-linked	300629	https://gene.sfari.org/database/human-gene/AP1S2
ARHGEF9	X	Developmental and epileptic encephalopathy 8; X-linked intellectual disability	300067	X-linked	300429	https://gene.sfari.org/database/human-gene/ARHGEF9
ARID1B	6	Coffin-Siris syndrome 1	135900	Autosomal dominant	614556	https://gene.sfari.org/database/human-gene/ARID1B
ARX	X	Early infantile epileptic encephalopathy 1; Partington syndrome; Lissencephaly, X-linked	308350	X-linked	300382	https://gene.sfari.org/database/human-gene/ARX
ASPM	1	Primary microcephaly 5	608716	Autosomal recessive	605481	https://gene.sfari.org/database/human-gene/ASPM
ASTN2	9	autism spectrum disorders			612856	https://gene.sfari.org/database/human-gene/ASTN2
ATP1A3	19	Alternating hemiplegia of childhood 2; Rapid-onset dystonia-parkinsonism; CAPOS syndrome	614820; 128235; 601338	Autosomal dominant;	182350	https://gene.sfari.org/database/human-gene/ATP1A3
ATP2B2	3	Deafness, autosomal dominant 78	619804	Autosomal dominant	108733	https://gene.sfari.org/database/human-gene/ATP2B2
ATRX	X	Alpha-thalassemia/mental retardation syndrome, X-linked (ATR-X)	301040	X-linked	300032	https://gene.sfari.org/database/human-gene/ATRX
AVPR1A	12	autism spectrum disorders			600821	https://gene.sfari.org/database/human-gene/AVPR1A
AUTS2	7	AUTS2 syndrome	615834	Autosomal dominant	607270	https://gene.sfari.org/database/human-gene/AUTS2
BCL11A	2	Dias–Logan syndrome	617101	Autosomal dominant	606557	https://gene.sfari.org/database/human-gene/BCL11A
BCORL1	X	X-linked syndromic intellectual disability, BCORL1-related	301029	X-linked recessive	300688	https://gene.sfari.org/database/human-gene/BCORL1
BRAF	7	Cardio-facio-cutaneous syndrome	115150	Autosomal dominant	164757	https://gene.sfari.org/database/human-gene/BRAF
BRSK2	11	autism spectrum disorders			608414	https://gene.sfari.org/database/human-gene/BRSK2
BRWD3	X	X-linked intellectual disability, Cabezas type	300659	X-linked recessive	300553	https://gene.sfari.org/database/human-gene/BRWD3
C12ORF57	12	Temtamy syndrome	218340	Autosomal recessive	615140	https://gene.sfari.org/database/human-gene/C12ORF57
CACNA1A	19	Episodic ataxia 2; Spinocerebellar ataxia 6; Familial hemiplegic migraine 1	108500; ; 141500	Autosomal dominant	601011	https://gene.sfari.org/database/human-gene/CACNA1A
CACNA1C	12	Timothy syndrome; Long QT syndrome 8; Brugada syndrome 3; Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures	601005; 618447; 611875; 620029	Autosomal dominant	114205	https://gene.sfari.org/database/human-gene/CACNA1C
CACNA1D	3	Neurodevelopmental disorder with hypotonia and autistic features; Primary aldosteronism, seizures, and neurologic abnormalities	615474	Autosomal dominant	114208	https://gene.sfari.org/database/human-gene/CACNA1D
CACNA1E	1	Developmental and epileptic encephalopathy-69	618285	Autosomal dominant	601013	https://gene.sfari.org/database/human-gene/CACNA1E
CACNA1F	X	Congenital stationary night blindness, X-linked; Incomplete congenital stationary night blindness	300071	X-linked	300110	https://gene.sfari.org/database/human-gene/CACNA1F
CACNA1G	17	Spinocerebellar ataxia 42	618087; 616795	Autosomal dominant	604065	https://gene.sfari.org/database/human-gene/CACNA1G
CACNA1H	16	{Epilepsy, childhood absence, susceptibility to, 6}; Hyperaldosteronism, familial, type IV	611942; 617027	Autosomal dominant	607904	https://gene.sfari.org/database/human-gene/CACNA1H
CACNA2D3	3	autism spectrum disorders			607081	https://gene.sfari.org/database/human-gene/CACNA2D3
CACNB2	10	Brugada syndrome 4	611876	Autosomal dominant	600003	https://gene.sfari.org/database/human-gene/CACNB2
CAMK4	5	Neurodevelopmental disorder with intellectual disability and spasticity			114080	https://gene.sfari.org/database/human-gene/CAMK4
CASK	X	Microcephaly with pontine and cerebellar hypoplasia; X-linked intellectual disability; FG syndrome 4	300422; 300749	X-linked recessive	300172	https://gene.sfari.org/database/human-gene/CASK
CC2D1A	19	Intellectual developmental disorder, autosomal recessive 3	608443	Autosomal recessive	610055	https://gene.sfari.org/database/human-gene/CC2D1A
CDC42BPB	14	Chilton-Okur-Chung neurodevelopmental syndrome	619841	Autosomal dominant	614062	https://gene.sfari.org/database/human-gene/CDC42BPB
CDH8	16	autism spectrum disorders			603008	https://gene.sfari.org/database/human-gene/CDH8
CDKL5	X	Developmental and epileptic encephalopathy 2	300672	X-linked	300203	https://gene.sfari.org/database/human-gene/CDKL5
CEP290	12	Joubert syndrome 5; Leber congenital amaurosis 10	611755	Autosomal recessive	610142	https://gene.sfari.org/database/human-gene/CEP290
CHD2	15	Developmental and epileptic encephalopathy 94	615369	Autosomal dominant	602119	https://gene.sfari.org/database/human-gene/CHD2
CHD3	17	Snijders Blok–Campeau syndrome	618622	Autosomal dominant	602120	https://gene.sfari.org/database/human-gene/CHD3
CHD7	8	CHARGE syndrome	214800	Autosomal dominant	608892	https://gene.sfari.org/database/human-gene/CHD7
CHD8	14	Neurodevelopmental disorder with macrocephaly and facial dysmorphism	615032	Autosomal dominant	610528	https://gene.sfari.org/database/human-gene/CHD8
CHKB	22	Muscular dystrophy, congenital, with mental retardation	602541	Autosomal recessive	612395	https://gene.sfari.org/database/human-gene/CHKB
CHRNA7	15	{Schizophrenia, susceptibility to; Epilepsy			118511	https://gene.sfari.org/database/human-gene/CHRNA7
CIC	19	Intellectual developmental disorder with speech delay and facial dysmorphism	617600	Autosomal dominant	612199	https://gene.sfari.org/database/human-gene/CIC
CNKSR2	X	X-linked intellectual disability with seizures		X-linked	300724	https://gene.sfari.org/database/human-gene/CNKSR2
CNOT3	19	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies	618672	Autosomal dominant	604910	https://gene.sfari.org/database/human-gene/CNOT3
CNTN4	3	autism spectrum disorders			607280	https://gene.sfari.org/database/human-gene/CNTN4

CNTNAP2	7	{Autism susceptibility 15}; Pitt-Hopkins like syndrome 1	612100; 610042	Autosomal recessive	604569	https://gene.sfari.org/database/human-gene/CNTNAP2
CNTNAP4	16	autism spectrum disorders			610518	https://gene.sfari.org/database/human-gene/CNTNAP4
CNTNAP5	2	autism spectrum disorders			610519	https://gene.sfari.org/database/human-gene/CNTNAP5
CORO1A	16	Immunodeficiency 8	615401	Autosomal recessive	605000	https://gene.sfari.org/database/human-gene/CORO1A
CREBBP	16	Rubinstein–Taybi syndrome 1; Menke-Hennekam syndrome 1	180849; 618332	Autosomal dominant	600140	https://gene.sfari.org/database/human-gene/CREBBP
CSDE1	1	autism spectrum disorders			602171	https://gene.sfari.org/database/human-gene/CSDE1
CSMD1	8	autism spectrum disorders			608397	https://gene.sfari.org/database/human-gene/CSMD1
CTNNA3	10	Arrhythmogenic right ventricular dysplasia 13	615616	Autosomal dominant	607667	https://gene.sfari.org/database/human-gene/CTNNA3
CTNNB1	3	Neurodevelopmental disorder with spastic diplegia and visual defects	615075	Autosomal dominant	116806	https://gene.sfari.org/database/human-gene/CTNNB1
CTTNBP2	7	autism spectrum disorders			609772	https://gene.sfari.org/database/human-gene/CTTNBP2
CUL3	2	Neurodevelopmental disorder with or without autism or seizures	619239	Autosomal dominant	603136	https://gene.sfari.org/database/human-gene/CUL3
CYP27A1	2	Cerebrotendinous xanthomatosis	213700	Autosomal recessive	606530	https://gene.sfari.org/database/human-gene/CYP27A1
DEAF1	11	Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures; Vulto-van Silfout-de Vries syndrome	617171, 615828;	Autosomal recessive; Autosomal dominant	602635	https://gene.sfari.org/database/human-gene/DEAF1
DHCR7	11	Smith-Lemli-Opitz syndrome	270400	Autosomal recessive	602858	https://gene.sfari.org/database/human-gene/DHCR7
DIP2A	21	autism spectrum disorders			611689	https://gene.sfari.org/database/human-gene/DIP2A
DISC1	1	{Schizophrenia 9, susceptibility to}	604906		605210	https://gene.sfari.org/database/human-gene/DISC1
DLGAP2	8	autism spectrum disorders			605445	https://gene.sfari.org/database/human-gene/DLGAP2
DLL1	6	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures	618709	Autosomal dominant	606582	https://gene.sfari.org/database/human-gene/DLL1
DNMT3A	2	Heyn-Sproul-Jackson syndrome; Tatton-Brown-Rahman syndrome	618724; 615879	Autosomal dominant	602769	https://gene.sfari.org/database/human-gene/DNMT3A
DOCK4	7	autism spectrum disorders			607679	https://gene.sfari.org/database/human-gene/DOCK4
DOCK8	9	Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections	243700	Autosomal recessive	611432	https://gene.sfari.org/database/human-gene/DOCK8
DOLK	9	Congenital disorder of glycosylation, type Im	610768	Autosomal recessive	610746	https://gene.sfari.org/database/human-gene/DOLK
DPP10	2	autism spectrum disorders			608209	https://gene.sfari.org/database/human-gene/DPP10
DPP6	7	Intellectual developmental disorder, autosomal dominant 33	616311	Autosomal dominant	126141	https://gene.sfari.org/database/human-gene/DPP6
DPYD	1	Dihydropyrimidine dehydrogenase deficiency	274270	Autosomal recessive	612779	https://gene.sfari.org/database/human-gene/DPYD
DPYSL2	8	autism spectrum disorders			602463	https://gene.sfari.org/database/human-gene/DPYSL2
DSCAM	21	autism spectrum disorders			602523	https://gene.sfari.org/database/human-gene/DSCAM
DST	6	Epidermolysis bullosa simplex with muscular dystrophy	226670	Autosomal recessive	113810	https://gene.sfari.org/database/human-gene/DST
DYNC1H1	14	Spinal muscular atrophy, lower extremity-predominant 1; Cortical dysplasia, complex, with other brain malformations 13; Charcot-Marie-Tooth disease, axonal, type 2O	158600; 614563; 614228	Autosomal dominant	600112	https://gene.sfari.org/database/human-gene/DYNC1H1
DYRK1A	21	Intellectual developmental disorder, autosomal dominant 7	614104	Autosomal dominant	600855	https://gene.sfari.org/database/human-gene/DYRK1A
EHMT1	9	Kleefstra syndrome	610253	Autosomal dominant	607001	https://gene.sfari.org/database/human-gene/EHMT1
EIF4E	4	{Autism, susceptibility to, 19}	615091		133440	https://gene.sfari.org/database/human-gene/EIF4E
ELP2	18	Intellectual developmental disorder, autosomal recessive 58	617270	Autosomal recessive	616054	https://gene.sfari.org/database/human-gene/ELP2
EN2	7	autism spectrum disorders			131310	https://gene.sfari.org/database/human-gene/EN2
EP300	22	Rubinstein–Taybi syndrome 2	613684	Autosomal dominant	602700	https://gene.sfari.org/database/human-gene/EP300
FBN1	15	Marfan syndrome	154700	Autosomal dominant	134797	https://gene.sfari.org/database/human-gene/FBN1
FEZF2	3	autism spectrum disorders			613301	https://gene.sfari.org/database/human-gene/FEZF2
FOXG1	14	FOXG1 syndrome	613454	Autosomal dominant	164874	https://gene.sfari.org/database/human-gene/FOXG1
FOXP1	3	FOXP1 syndrome (neurodevelopmental disorder with language impairment)	613670	Autosomal dominant	605515	https://gene.sfari.org/database/human-gene/FOXP1
FOXP2	7	Speech and language disorder 1; Childhood apraxia of speech	602081	Autosomal dominant	605317	https://gene.sfari.org/database/human-gene/FOXP2
FRMPD4	X	Intellectual developmental disorder, X-linked 104	300983	X-linked	300781	https://gene.sfari.org/database/human-gene/FRMPD4
GABRB3	15	Epileptic encephalopathy, childhood absence epilepsy; Autism susceptibility	617113	Autosomal dominant	137192	https://gene.sfari.org/database/human-gene/GABRB3
GABRG3	15	autism spectrum disorders			137166	https://gene.sfari.org/database/human-gene/GABRG3
GALNT2	1	Congenital disorder of glycosylation, type Iit	618885	Autosomal recessive	602274	https://gene.sfari.org/database/human-gene/GALNT2
GATM	15	Cerebral creatine deficiency syndrome 3; Fanconi renotubular syndrome 1	612718; 134600	Autosomal recessive; Autosomal dominant	602360	https://gene.sfari.org/database/human-gene/GATM
GFAP	17	Alexander disease	203450	Autosomal dominant	137780	https://gene.sfari.org/database/human-gene/GFAP
GIGYF2	2	Parkinsonism susceptibility; Neurodevelopmental susceptibility	607688		612003	https://gene.sfari.org/database/human-gene/GIGYF2
GPHN	14	Molybdenum cofactor deficiency-like; Epileptic encephalopathy	615501	Autosomal recessive	603930	https://gene.sfari.org/database/human-gene/GPHN
GRIA3	X	X-linked intellectual disability with behavioral abnormalities	300699	X-linked	305915	https://gene.sfari.org/database/human-gene/GRIA3
GRIK2	6	Intellectual developmental disorder, autosomal recessive 6; Neurodevelopmental disorder with impaired language and ataxia and with or without seizures	611092; 619580	Autosomal recessive; Autosomal dominant	138244	https://gene.sfari.org/database/human-gene/GRIK2
GRIN1	9	Developmental and epileptic encephalopathy-29; Neurodevelopmental disorder with hypotonia	617171	Autosomal dominant	138249	https://gene.sfari.org/database/human-gene/GRIN1
GRIN2A	16	Epilepsy, focal, with speech disorder and rolandic discharges; Developmental and epileptic encephalopathy-27	616214	Autosomal dominant	138253	https://gene.sfari.org/database/human-gene/GRIN2A
GRIN2B	12	Developmental and epileptic encephalopathy 27; Intellectual developmental disorder, autosomal dominant 6, with or without seizures	616139; 613970	Autosomal dominant	138252	https://gene.sfari.org/database/human-gene/GRIN2B
GRIP1	12	Fraser syndrome 3	617667	Autosomal recessive	604597	https://gene.sfari.org/database/human-gene/GRIP1
GRM5	11	autism spectrum disorders			602666	https://gene.sfari.org/database/human-gene/GRM5
GRM7	3	Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities	618922	Autosomal recessive	604101	https://gene.sfari.org/database/human-gene/GRM7

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HDAC4	2	Neurodevelopmental disorder with central hypotonia and dysmorphic facies	619797	Autosomal dominant	605314	https://gene.sfari.org/database/human-gene/HDAC4
HDAC8	X	Cornelia de Lange syndrome 5	300882	X-linked Dominant	300269	https://gene.sfari.org/database/human-gene/HDAC8
HDLBP	2	autism spectrum disorders			602283	https://gene.sfari.org/database/human-gene/HDLBP
HEPACAM	11	Megalencephalic leukoencephalopathy with subcortical cysts 2A; Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development	613925; 613926	Autosomal recessive; Autosomal dominant	611642	https://gene.sfari.org/database/human-gene/HEPACAM
HERC2	15	Intellectual developmental disorder, autosomal recessive 38	615516	Autosomal recessive	605837	https://gene.sfari.org/database/human-gene/HERC2
HNRNPU	1	Developmental and epileptic encephalopathy 54	617391	Autosomal dominant	602869	https://gene.sfari.org/database/human-gene/HNRNPU
HOXA1	7	Athabaskan brainstem dysgenesis syndrome; Bosley-Salih-Alorainy syndrome	601536;	Autosomal recessive	142955	https://gene.sfari.org/database/human-gene/HOXA1
HRAS	11	Costello syndrome	218040	Autosomal dominant	190020	https://gene.sfari.org/database/human-gene/HRAS
HUWE1	X	Intellectual developmental disorder, X-linked syndromic, Turner type	309590	X-linked	300697	https://gene.sfari.org/database/human-gene/HUWE1
IL1RAPL1	X	X-linked intellectual disability with autism	300143	X-linked	300206	https://gene.sfari.org/database/human-gene/IL1RAPL1
IQSEC2	X	Intellectual developmental disorder, X-linked 1	309530	X-linked dominant	300522	https://gene.sfari.org/database/human-gene/IQSEC2
ITGB3	17	Bleeding disorder, platelet-type, 24, autosomal dominant	619271	X-linked	173470	https://gene.sfari.org/database/human-gene/ITGB3
ITPR1	3	Gillespie syndrome; Spinocerebellar ataxia 15; Spinocerebellar ataxia 29, congenital nonprogressive	206700; 606658; 117360	Autosomal recessive; Autosomal dominant	147265	https://gene.sfari.org/database/human-gene/ITPR1
JMJD1C	10	autism spectrum disorders			604503	https://gene.sfari.org/database/human-gene/JMJD1C
KANK1	9	Cerebral palsy, spastic quadriplegic, 2	612900		607704	https://gene.sfari.org/database/human-gene/KANK1
KANSL1	17	Koolen-de Vries syndrome	610443	Autosomal dominant	612452	https://gene.sfari.org/database/human-gene/KANSL1
KATNAL2	18	autism spectrum disorder			614087	https://gene.sfari.org/database/human-gene/KATNAL2
KCND3	1	Spinocerebellar ataxia 19; Brugada syndrome 9	607346; 616399	Autosomal dominant	605407	https://gene.sfari.org/database/human-gene/KCND3
KCNJ10	1	SeSAME (EAST) syndrome	612780	Autosomal recessive	602208	https://gene.sfari.org/database/human-gene/KCNJ10
KCNMA1	10	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy; Liang-Wang syndrome	609446; 618729	Autosomal dominant	600150	https://gene.sfari.org/database/human-gene/KCNMA1
KCNQ2	20	Developmental and epileptic encephalopathy 7	613720	Autosomal dominant	602235	https://gene.sfari.org/database/human-gene/KCNQ2
KCNQ3	8	Developmental and epileptic encephalopathy 2	300672	X-linked	602232	https://gene.sfari.org/database/human-gene/KCNQ3
KDM5A	12	El Hayek-Chahrour neurodevelopmental syndrome	620820	Autosomal recessive	180202	https://gene.sfari.org/database/human-gene/KDM5A
KDM5C	X	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type	300534	X-linked Recessive	314690	https://gene.sfari.org/database/human-gene/KDM5C
KDM6A	X	Kabuki syndrome 2	300867	X-linked	300128	https://gene.sfari.org/database/human-gene/KDM6A
KDM6B	17	Stolerman neurodevelopmental syndrome	618505	Autosomal dominant	608137	https://gene.sfari.org/database/human-gene/KDM6B
KIAA0232	4	autism spectrum disorders			619237	https://gene.sfari.org/database/human-gene/KIAA0232
KIF1A	2	Spastic paraplegia 30, autosomal dominant; NESCAV syndrom; Neuropathy, hereditary sensory, type IIC	620607; 610357; 614213; 614255	Autosomal recessive; Autosomal dominant	601255	https://gene.sfari.org/database/human-gene/KIF1A
KIRREL3	11	autism spectrum disorders			607761	https://gene.sfari.org/database/human-gene/KIRREL3
KMT2C	7	Kleefstra syndrome 2	617768	Autosomal dominant	606833	https://gene.sfari.org/database/human-gene/KMT2C
KMT2E	7	O'Donnell-Luria-Rodan syndrome	618512	Autosomal dominant	611360	https://gene.sfari.org/database/human-gene/KMT2E
LAMA1	18	Poretti–Boltshauser syndrome	615960	Autosomal recessive	150320	https://gene.sfari.org/database/human-gene/LAMA1
LAMB1	7	Lissencephaly 5	615191	Autosomal recessive	150240	https://gene.sfari.org/database/human-gene/LAMB1
LRP1	12	Developmental dysplasia of the hip 3	620690	Autosomal dominant	107770	https://gene.sfari.org/database/human-gene/LRP1
LRP2	2	Donnai-Barrow syndrome	222448	Autosomal recessive	600073	https://gene.sfari.org/database/human-gene/LRP2
MAGEL2	15	Schaaf-Yang syndrome	615547	Autosomal dominant	605283	https://gene.sfari.org/database/human-gene/MAGEL2
MAOA	X	Brunner syndrome	300615	X-linked Recessive	309850	https://gene.sfari.org/database/human-gene/MAOA
MBD5	2	Intellectual developmental disorder, autosomal dominant 1	156200	Autosomal dominant	611472	https://gene.sfari.org/database/human-gene/MBD5
MCPH1	8	Microcephaly 1, primary, autosomal recessive	251200	Autosomal recessive	607117	https://gene.sfari.org/database/human-gene/MCPH1
MECP2	X	Rett syndrome; PPM-X syndrome	312750	X-linked;	300005	https://gene.sfari.org/database/human-gene/MECP2
MED13	17	Intellectual developmental disorder, autosomal dominant 61	618009	Autosomal dominant	603808	https://gene.sfari.org/database/human-gene/MED13
MED13L	12	Impaired intellectual development and distinctive facial features with or without cardiac defects	616789	Autosomal dominant	608771	https://gene.sfari.org/database/human-gene/MED13L
MEF2C	5	Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language	613443	Autosomal dominant	600662	https://gene.sfari.org/database/human-gene/MEF2C
MEIS2	15	Cleft palate, cardiac defects, and impaired intellectual development	600987	Autosomal dominant	601740	https://gene.sfari.org/database/human-gene/MEIS2
MET	7	{Osteofibrous dysplasia, susceptibility to}; ?Deafness, autosomal recessive 97	607278; 620019	Autosomal dominant	164860	https://gene.sfari.org/database/human-gene/MET
MTHFR	1	Homocystinuria due to MTHFR deficiency	236250	Autosomal recessive	607093	https://gene.sfari.org/database/human-gene/MTHFR
MYT1L	2	Intellectual developmental disorder, autosomal dominant 39	616521	Autosomal dominant	613084	https://gene.sfari.org/database/human-gene/MYT1L
NAV2	11	autism spectrum disorders			607026	https://gene.sfari.org/database/human-gene/NAV2
NBEA	13	Neurodevelopmental disorder with or without early-onset generalized epilepsy	619157	Autosomal dominant	604889	https://gene.sfari.org/database/human-gene/NBEA
NCKAP1	2	autism spectrum disorders			602223	https://gene.sfari.org/database/human-gene/NCKAP1
NCOA1	2	autism spectrum disorders			601626	https://gene.sfari.org/database/human-gene/NCOA1
NF1	17	Neurofibromatosis type 1; Neurofibromatosis-Noonan syndrome; Watson syndrome	162200; 601321; 193520	Autosomal dominant	613113	https://gene.sfari.org/database/human-gene/NF1
NFIA	1	Brain malformations with or without urinary tract defects	613735	Autosomal dominant	600727	https://gene.sfari.org/database/human-gene/NFIA
NFIX	19	Malan syndrome; Marshall-Smith syndrome	614753; 602535	Autosomal dominant	164005	https://gene.sfari.org/database/human-gene/NFIX
NIPBL	5	Cornelia de Lange syndrome 1	122470	Autosomal dominant	608667	https://gene.sfari.org/database/human-gene/NIPBL
NLGN1	3	{Autism, susceptibility to, 20}	618830	Autosomal dominant	600568	https://gene.sfari.org/database/human-gene/NLGN1
NLGN2	17	autism spectrum disorders			600567	https://gene.sfari.org/database/human-gene/NLGN2
NLGN3	X	{Autism susceptibility, X-linked 1}	300425	X-linked	300336	https://gene.sfari.org/database/human-gene/NLGN3

NLGN4X	X	{Autism susceptibility, X-linked 2}; Intellectual developmental disorder, X-linked	300495	X-linked	300427	https://gene.sfari.org/database/human-gene/NLGN4X
NOTCH1	9	Adams-Oliver syndrome 5; Aortic valve disease 1	616028; 109730	Autosomal dominant	190198	https://gene.sfari.org/database/human-gene/NOTCH1
NR2F1	5	Bosch–Boonstra–Schaaf optic atrophy syndrome	615722	Autosomal dominant	107773	https://gene.sfari.org/database/human-gene/NR2F1
NR3C2	4	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy; Pseudohypoaldosteronism type I, autosomal dominant	605115; 177735	Autosomal dominant	600983	https://gene.sfari.org/database/human-gene/NR3C2
NR4A2	2	Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism	619911	Autosomal dominant	601828	https://gene.sfari.org/database/human-gene/NR4A2
NRP2	2	autism spectrum disorders			601872	https://gene.sfari.org/database/human-gene/NRP2
NRXN1	2	Pitt-Hopkins-like syndrome 2; {Schizophrenia, susceptibility to, 17}	614325; 614332	Autosomal recessive	600565	https://gene.sfari.org/database/human-gene/NRXN1
NRXN2	11	Neurodevelopmental disorder with autistic features			600566	https://gene.sfari.org/database/human-gene/NRXN2
NRXN3	14	Neurodevelopmental disorder with autistic features			600567	https://gene.sfari.org/database/human-gene/NRXN3
NSD1	5	Sotos syndrome 1	117550	Autosomal dominant	606681	https://gene.sfari.org/database/human-gene/NSD1
NTNG1	1	Autism Spectrum Disorder			608818	https://gene.sfari.org/database/human-gene/NTNG1
NTRK2	9	Developmental and epileptic encephalopathy 58; Obesity, hyperphagia, and developmental delay	617830; 613886	Autosomal dominant	600456	https://gene.sfari.org/database/human-gene/NTRK2
NTRK3	15	Susceptibility to Anxiety Disorders; Susceptibility to Ventricular Septal Defects			604738	https://gene.sfari.org/database/human-gene/NTRK3
NUP155	5	Atrial fibrillation, familial 10	611495	Autosomal recessive	607614	https://gene.sfari.org/database/human-gene/NUP155
OCRL	X	Lowe syndrome; Dent disease 2	300555	X-linked	300535	https://gene.sfari.org/database/human-gene/OCRL
OPHN1	X	Intellectual developmental disorder, X-linked syndromic, Billuart type	300486	X-linked recessive	300127	https://gene.sfari.org/database/human-gene/OPHN1
OXTR	3	deficits in sociobehavioral domains			167055	https://gene.sfari.org/database/human-gene/OXTR
PAH	12	Phenylketonuria	261600	Autosomal recessive	612349	https://gene.sfari.org/database/human-gene/PAH
PAX5	9	autism spectrum disorders			167414	https://gene.sfari.org/database/human-gene/PAX5
PAX6	11	Aniridia; WAGR syndrome	106210	Autosomal dominant	607108	https://gene.sfari.org/database/human-gene/PAX6
PCCA	13	Propionicacidemia	606054	Autosomal recessive	232000	https://gene.sfari.org/database/human-gene/PCCA
PCCB	3	Propionicacidemia	606054	Autosomal recessive	232050	https://gene.sfari.org/database/human-gene/PCCB
PCDH15	10	Usher syndrome, type 1D/F digenic	601067	Autosomal recessive	605514	https://gene.sfari.org/database/human-gene/PCDH15
PCDH19	X	Developmental and epileptic encephalopathy 9	300088	X-linked	300460	https://gene.sfari.org/database/human-gene/PCDH19
PCDH9	13	autism spectrum disorders			612284	https://gene.sfari.org/database/human-gene/PCDH9
PHF2	9	autism spectrum disorders			604966	https://gene.sfari.org/database/human-gene/PHF2
PHF3	6	autism spectrum disorders			607789	https://gene.sfari.org/database/human-gene/PHF3
PHF8	X	Intellectual developmental disorder, X-linked syndromic, Siderius type	300263	X-linked recessive	300560	https://gene.sfari.org/database/human-gene/PHF8
PHIP	6	Chung-Jansen syndrome	617991	Autosomal dominant	612870	https://gene.sfari.org/database/human-gene/PHIP
PIK3R2	19	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1	603387	Autosomal dominant	603157	https://gene.sfari.org/database/human-gene/PIK3R2
POGZ	1	White-Sutton syndrome	616364	Autosomal dominant	614787	https://gene.sfari.org/database/human-gene/POGZ
POMGNT1	1	Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3	613151; 253280	Autosomal recessive	606822	https://gene.sfari.org/database/human-gene/POMGNT1
PPM1D	17	Jansen-de Vries syndrome	617450	Autosomal dominant	605100	https://gene.sfari.org/database/human-gene/PPM1D
PRICKLE2	3	EPILEPSY, PROGRESSIVE MYOCLONIC, 5			609629	https://gene.sfari.org/database/human-gene/PRICKLE2
PRODH	22	Hyperprolinemia, type I; {Schizophrenia, susceptibility to, 4}	239500; 600850	Autosomal recessive; Autosomal dominant	606810	https://gene.sfari.org/database/human-gene/PRODH
PTCHD1	X	{Autism, susceptibility to, X-linked 4}	300830	X-linked recessive	300828	https://gene.sfari.org/database/human-gene/PTCHD1
PTEN	10	Cowden syndrome 1; Lhermitte-Duclos disease	158350	Autosomal dominant	601728	https://gene.sfari.org/database/human-gene/PTEN
PTK7	6	autism spectrum disorders			604985	https://gene.sfari.org/database/human-gene/PTK7
PTPN11	12	Noonan syndrome 1; LEOPARD syndrome 1; Metachondromatosis	163950; 151100; 156250	Autosomal dominant	176876	https://gene.sfari.org/database/human-gene/PTPN11
PTPRC	1	Immunodeficiency 105, severe combined	619924	Autosomal recessive	151460	https://gene.sfari.org/database/human-gene/PTPRC
PTPRD	9	autism spectrum disorders			601598	https://gene.sfari.org/database/human-gene/PTPRD
PTPRT	20	severe intellectual disability			608026	https://gene.sfari.org/database/human-gene/PTPRT
RAB39B	X	Waisman syndrome; Intellectual developmental disorder, X-linked 72	311510; 300271	X-linked	300774	https://gene.sfari.org/database/human-gene/RAB39B
RAC1	7	Intellectual developmental disorder, autosomal dominant 48	617751	Autosomal dominant	602048	https://gene.sfari.org/database/human-gene/RAC1
RAD21	8	Cornelia de Lange syndrome 4;	614701	Autosomal dominant	606462	https://gene.sfari.org/database/human-gene/RAD21
RAI1	17	Smith-Magenis syndrome	182290	Autosomal dominant	607642	https://gene.sfari.org/database/human-gene/RAI1
RBFOX1	16	autism spectrum disorders			605104	https://gene.sfari.org/database/human-gene/RBFOX1
RELN	7	Lissencephaly 2 (Norman-Roberts type); {Epilepsy, familial temporal lobe, 7}	257320; 616436	Autosomal recessive; Autosomal dominant	600514	https://gene.sfari.org/database/human-gene/RELN
RIMS1	6	Cone-rod dystrophy 7	613829	Autosomal dominant	606629	https://gene.sfari.org/database/human-gene/RIMS1
RNF135	17	macrocephaly, macrosomia, and facial dysmorphism syndrome	613675	Autosomal dominant	611358	https://gene.sfari.org/database/human-gene/RNF135
ROBO2	3	Vesicoureteral reflux 2	610878	Autosomal dominant	602431	https://gene.sfari.org/database/human-gene/ROBO2
RORA	15	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia	618060	Autosomal dominant	600825	https://gene.sfari.org/database/human-gene/RORA
RPL10	X	Intellectual developmental disorder, X-linked syndromic 35; {Autism, susceptibility to, X-linked 5}	300998; 300847	X-linked recessive	312173	https://gene.sfari.org/database/human-gene/RPL10
RPS6KA3	X	Coffin-Lowry syndrome; Intellectual developmental disorder, X-linked 19	303600; 300844	X-linked dominant	300075	https://gene.sfari.org/database/human-gene/RPS6KA3
RSRC1	3	Intellectual developmental disorder, autosomal recessive 70	618402	Autosomal recessive	611638	https://gene.sfari.org/database/human-gene/RSRC1
SATB2	2	Glass syndrome	612313	Autosomal dominant	608148	https://gene.sfari.org/database/human-gene/SATB2

SCN1A	2	Dravet syndrome; GEFS+ type 2; Developmental and epileptic encephalopathy-6	607208; 604233;	Autosomal dominant; Autosomal dominant;	182389	https://gene.sfari.org/database/human-gene/SCN1A
SCN2A	2	Developmental and epileptic encephalopathy 11; Episodic ataxia, type 9; Seizures, benign familial infantile, 3	613721; 618924; 607745	Autosomal dominant	182390	https://gene.sfari.org/database/human-gene/SCN2A
SCN8A	12	Developmental and epileptic encephalopathy 13	614558	Autosomal dominant	600702	https://gene.sfari.org/database/human-gene/SCN8A
SCN9A	2	Small fiber neuropathy; Paroxysmal extreme pain disorder; Neuropathy, hereditary sensory and autonomic, type IID; Insensitivity to pain, congenital; Erythermalgia, primary	133020; 167400; 243000; ;	Autosomal recessive; Autosomal dominant	603415	https://gene.sfari.org/database/human-gene/SCN9A
SETBP1	18	Intellectual developmental disorder, autosomal dominant 29; Schinzel-Giedion midface retraction syndrome	616078; 269150	Autosomal dominant	611060	https://gene.sfari.org/database/human-gene/SETBP1
SETD2	3	Intellectual developmental disorder, autosomal dominant 70; Luscan-Lumish syndrome; Rabin-Pappas syndrome	620157; 616831; 620155	Autosomal dominant	612778	https://gene.sfari.org/database/human-gene/SETD2
SGSH	17	Mucopolysaccharidosis type IIIA (Sanfilippo A)	252900	Autosomal recessive	605270	https://gene.sfari.org/database/human-gene/SGSH
SHANK2	11	{Autism susceptibility 17}	613436		603290	https://gene.sfari.org/database/human-gene/SHANK2
SHANK3	22	Phelan-McDermid syndrome	606232	Autosomal dominant	606230	https://gene.sfari.org/database/human-gene/SHANK3
SKI	1	Shprintzen-Goldberg syndrome	182212	Autosomal dominant	164780	https://gene.sfari.org/database/human-gene/SKI
SLC1A1	9	Dicarboxylic aminoaciduria; {?Schizophrenia susceptibility 18}	222730; 615232	Autosomal recessive	133550	https://gene.sfari.org/database/human-gene/SLC1A1
SLC25A12	2	Developmental and epileptic encephalopathy 39	612949	Autosomal recessive	603667	https://gene.sfari.org/database/human-gene/SLC25A12
SLC6A1	3	Myoclonic–atonic epilepsy; Neurodevelopmental disorder with intellectual disability	616421	Autosomal dominant	611873	https://gene.sfari.org/database/human-gene/SLC6A1
SLC6A3	5	Parkinsonism-dystonia, infantile, 1	613135	Autosomal recessive	126455	https://gene.sfari.org/database/human-gene/SLC6A3
SLC6A4	17	{Anxiety-related personality traits}; {Obsessive-compulsive disorder}	607834; 164230	Autosomal dominant	182138	https://gene.sfari.org/database/human-gene/SLC6A4
SLC6A8	X	Cerebral creatine deficiency syndrome 1	300352	X-linked	300036	https://gene.sfari.org/database/human-gene/SLC6A8
SLC9A6	X	Christianson syndrome	300243	X-linked	300231	https://gene.sfari.org/database/human-gene/SLC9A6
SLC9A9	3	{?Autism susceptibility 16}	613410		608396	https://gene.sfari.org/database/human-gene/SLC9A9
SMAD4	18	Myhre syndrome	139210	Autosomal dominant	600993	https://gene.sfari.org/database/human-gene/SMAD4
SMARCA2	9	Nicolaides-Baraitser syndrome	601358	Autosomal dominant	600014	https://gene.sfari.org/database/human-gene/SMARCA2
SMARCA4	19	Coffin-Siris syndrome 4	614609	Autosomal dominant	603254	https://gene.sfari.org/database/human-gene/SMARCA4
SMC1A	X	Cornelia de Lange syndrome 2	300590	X-linked	300040	https://gene.sfari.org/database/human-gene/SMC1A
SMC3	10	Cornelia de Lange syndrome 3	610759	Autosomal dominant	606062	https://gene.sfari.org/database/human-gene/SMC3
SOX5	12	Lamb-Shaffer syndrome	616803	Autosomal dominant	604975	https://gene.sfari.org/database/human-gene/SOX5
SOX6	11	Tolchin-Le Caignec syndrome	618971	Autosomal dominant	607257	https://gene.sfari.org/database/human-gene/SOX6
SPAST	2	Spastic paraplegia 4, autosomal dominant	182601	Autosomal dominant	604277	https://gene.sfari.org/database/human-gene/SPAST
SRCAP	16	Floating-Harbor syndrome	136140	Autosomal dominant	611421	https://gene.sfari.org/database/human-gene/SRCAP
ST8SIA2	15	autism spectrum disorders			602437	https://gene.sfari.org/database/human-gene/ST8SIA2
STXBP1	9	Developmental and epileptic encephalopathy 4	612164	Autosomal dominant	602926	https://gene.sfari.org/database/human-gene/STXBP1
SUPT16H	14	Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum	619480	Autosomal dominant	603235	https://gene.sfari.org/database/human-gene/SUPT16H
SYN1	X	Intellectual developmental disorder, X-linked 50; Epilepsy, X-linked 1, with variable learning disabilities and behavior disorders	300115; 300491	X-linked	313440	https://gene.sfari.org/database/human-gene/SYN1
SYNE1	6	Spinocerebellar ataxia, recessive 8; Arthrogryposis multiplex congenita 3	610743; 618484	Autosomal recessive	608441	https://gene.sfari.org/database/human-gene/SYNE1
SYNGAP1	6	Intellectual developmental disorder, autosomal dominant 5	612621	Autosomal dominant	603384	https://gene.sfari.org/database/human-gene/SYNGAP1
TAF1	X	X-linked syndromic intellectual disability-33		X-linked	313650	https://gene.sfari.org/database/human-gene/TAF1
TBC1D23	3	Pontocerebellar hypoplasia, type 11	617695	Autosomal recessive	617687	https://gene.sfari.org/database/human-gene/TBC1D23
TBL1XR1	3	Intellectual developmental disorder, autosomal dominant 41; Pierpont syndrome	616944; 602342	Autosomal dominant	608628	https://gene.sfari.org/database/human-gene/TBL1XR1
TBX1	22	22q11.2 deletion/DiGeorge spectrum (TBX1-related)	188400	Autosomal dominant	602054	https://gene.sfari.org/database/human-gene/TBX1
TCF4	18	Pitt-Hopkins syndrome	610954	Autosomal dominant	602272	https://gene.sfari.org/database/human-gene/TCF4
TCF7L2	10	{Diabetes mellitus, type 2, susceptibility to}	125853	Autosomal dominant	602228	https://gene.sfari.org/database/human-gene/TCF7L2
TEK	9	Venous malformation, multiple cutaneous and mucosal	617272; 600195	Autosomal dominant	600221	https://gene.sfari.org/database/human-gene/TEK
TMLHE	X	{Autism, susceptibility to, X-linked 6}	300872	X-linked	300777	https://gene.sfari.org/database/human-gene/TMLHE
TRAPPC9	8	Intellectual developmental disorder, autosomal recessive 13	613192	Autosomal recessive	611966	https://gene.sfari.org/database/human-gene/TRAPPC9
TRIO	5	Neurodevelopmental disorder with dysmorphic facies and variable intellectual disability			601893	https://gene.sfari.org/database/human-gene/TRIO
TRIP12	2	Intellectual developmental disorder, autosomal dominant 49	617752	Autosomal dominant	604506	https://gene.sfari.org/database/human-gene/TRIP12
TRPM3	9	Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures	620224	Autosomal dominant	608961	https://gene.sfari.org/database/human-gene/TRPM3
TRRAP	7	Developmental delay with or without dysmorphic facies and autism	618454	Autosomal dominant	603015	https://gene.sfari.org/database/human-gene/TRRAP
TSC1	9	Tuberous sclerosis complex 1	191100	Autosomal dominant	605284	https://gene.sfari.org/database/human-gene/TSC1
TSC2	16	Tuberous sclerosis complex 2	613254	Autosomal dominant	191092	https://gene.sfari.org/database/human-gene/TSC2
TSHZ1	18	Aural atresia, congenital	607842	Autosomal dominant	614427	https://gene.sfari.org/database/human-gene/TSHZ1
TTI2	8	Intellectual developmental disorder, autosomal recessive 39	615541	Autosomal recessive	613425	https://gene.sfari.org/database/human-gene/TTI2
TTN	2	Dilated cardiomyopathy 1G; Myofibrillar myopathy 9	604145; 603689	Autosomal dominant	188840	https://gene.sfari.org/database/human-gene/TTN
UBE3A	15	Angelman syndrome	105830	Autosomal dominant	601623	https://gene.sfari.org/database/human-gene/UBE3A
UBN2	7	autism spectrum disorders			613841	https://gene.sfari.org/database/human-gene/UBN2
UBR1	15	Johanson-Blizzard syndrome	243800	Autosomal recessive	605981	https://gene.sfari.org/database/human-gene/UBR1
UNC80	2	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2	616801	Autosomal recessive	612636	https://gene.sfari.org/database/human-gene/UNC80
UPF3B	X	X-linked intellectual disability 63	300676	X-linked	300298	https://gene.sfari.org/database/human-gene/UPF3B

PrenatalAutism - list of genes screened and genetic diseases associated to ASD

USH2A	1	Usher syndrome, type IIA	276901	Autosomal recessive	608400	https://gene.sfari.org/database/human-gene/USH2A
USP7	16	Hao-Fountain syndrome	616863	Autosomal dominant	602519	https://gene.sfari.org/database/human-gene/USP7
USP9X	X	Intellectual developmental disorder, X-linked 99	300919; 300968	X-linked dominant	300072	https://gene.sfari.org/database/human-gene/USP9X
VPS13B	8	Cohen syndrome	216550	Autosomal recessive	608879	https://gene.sfari.org/database/human-gene/VPS13B
WDFY3	4	Microcephaly 18, primary, autosomal dominant	617520	Autosomal dominant	612242	https://gene.sfari.org/database/human-gene/WDFY3
WDFY4	10	autism spectrum disorders			613316	https://gene.sfari.org/database/human-gene/WDFY4
WWOX	16	Developmental and epileptic encephalopathy 28; Spinocerebellar ataxia, autosomal recessive 12	616211; 614322	Autosomal recessive	605131	https://gene.sfari.org/database/human-gene/WWOX
XPC	3	Xeroderma pigmentosum, complementation group C	278720	Autosomal recessive	613208	https://gene.sfari.org/database/human-gene/XPC
YY1	14	Gabriele-de Vries syndrome	617557	Autosomal dominant	600013	https://gene.sfari.org/database/human-gene/YY1
ZBTB18	1	Intellectual developmental disorder, autosomal dominant 22	612337	Autosomal dominant	612337	https://gene.sfari.org/database/human-gene/ZBTB18
ZMYND11	10	Intellectual developmental disorder, autosomal dominant 30	616083	Autosomal dominant	608668	https://gene.sfari.org/database/human-gene/ZMYND11
ZNF804A	2	autism spectrum disorders			612282	https://gene.sfari.org/database/human-gene/ZNF804A