SFARI GENE Q3/2025 REPORT

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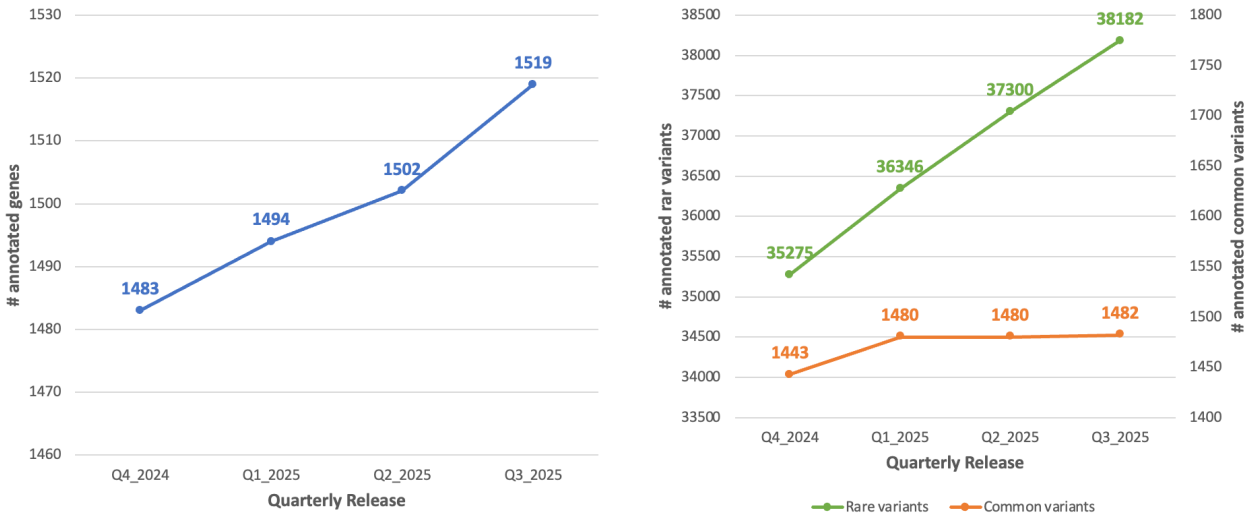
1. Human Gene Module
Quarterly Report, Q3/2025

1.1 Updated Human Gene Dataset

A total of ***seventeen*** new genes were added to the Human Gene Module for the Q3/2025 release, bringing the overall number of ASD candidate genes in the module to **1,519** (HG_Figure 1, panel A). In-depth annotation of **882** rare variants and two common variants were completed in this quarter leading to a total of **38,182** rare and **1,482** common variants, respectively (HG_Figure 1, panel B). Annotation of **114** new references was accomplished for the Human Gene module in Q3/2025, bringing the total number of references to **6,567**.



HG_Figure 1. Number of ASD-linked genes and variants in the Human Gene Module over the last four quarters. (A) The number of genes has grown from 1483 to 1519 (B) The number of rare variants has increased from 35,275 to 38,182; number of common variants has increased from 1,443 to 1,482.

1.2 Highlights of Q3/2025 Human Gene Dataset

In **Q3/2025**, we have curated new ASD-associated genes and variants from recently published studies, spanning genome-wide investigations to smaller focused cohort analyses. New gene selection was strictly guided by multiple lines of evidence relevant for ASD, and several existing ASD genes were updated with newly reported variants. Importantly, a number of reports described novel rare variants in ethnically and geographically diverse populations, expanding representation and providing a broader view of the global genetic architecture of ASD.

The Q3/2025 dataset is described in the following sections:

- **New genes added in this quarter (HG_Table 1)**
- **Summary evidence for new genes (Section 1.4)**
- **New reports added to existing genes (HG_Table 2)**
- **New articles added to the SLOE dataset (HG_Table 3)**

HG_Table 1. New genes added in Q3/2025

Gene Symbol	Rare Single Gene Variant	Syndromic	Functional
DOT1L			*1, 2*
GABRA2			*1*
CAPZA2			*1, 2*
MAPK8IP3			*1*
TBCB			*1*
CXXC5			*1*
NPAS3			*1*
ARHGAP30			*1*
ZNF644			*1*
DENND2B			*1*
PTPRS			*1*
DNAJC5			*1*
CTPS1			
NMT1			
CBX4			
HNRNPL			
KDM1A			

Rare single gene evidence: Defined as a gene with at least two rare variants in the protein-coding region identified in individuals diagnosed with ASD.

Syndromic evidence: Presentation of syndromic features in addition to a diagnosis of ASD

Functional evidence details:

(1) In vitro / in vivo functional analysis of patient-derived variants
(2) Mouse model recapitulating core ASD-associated phenotypes

1.3 Description of Q3/2025 Human Gene Dataset

The **Q3/2025 release** expands the curated landscape of ASD genetics with the addition of newly identified genes and variants from a broad range of recent studies. In curating these entries, we emphasized the selection of robust genetic and functional evidence, prioritizing findings supported by multiple lines of evidence. This approach ensured that only well-validated candidates were incorporated, strengthening the overall quality of the resource. The specific evidence used to guide the selection and annotation of these new genes is summarized below.

- Kim et al., 2025, analyzed 78,685 individuals from 21,735 ASD families across the Korean Autism cohort, Simons Simplex Collection (SSC), and SPARK. They introduced a within-family standardized deviation (WFSD) framework that measures proband phenotypes relative to unaffected family members, thereby correcting for shared genetic and environmental background. Genetic analyses focused on disruptive de novo variants (dnDIS), including protein-truncating variants (LOEUF < 0.37) and deleterious missense variants (MPC ≥ 2). This innovation provided more precise effect estimates than raw phenotype scores and enhanced gene discovery. Using the WFSD approach, the authors identified 38 genes enriched in probands with severe phenotypes, of which 20 overlapped with exiting SFARI Gene(s) and 18 represented novel ASD-associated genes. Importantly, identification of established high-confidence ASD genes such as SCN2A, SHANK3, PTEN, CHD2, MED13L, and WDFY3 reaffirmed the stronger phenotypic associations, underscoring the validity of the WFSD framework. Out of the 18 newly identified genes discovered using the WFSD framework, we further refined the list by applying stringent genetic filters. Specifically, we prioritized genes with pLI scores > 0.9 that also had at least one de novo loss-of-function variant previously reported in an ASD proband. This resulted in the addition of five genes: CTPS1, NMT1, CBX4, HNRNPL, and KDM1A. This additional filter increased confidence that these candidates likely represent true ASD risk genes rather than background signals, thereby providing stronger biological plausibility for their involvement in ASD.

- Chen et al., 2025 systematically evaluated 227,878 non-coding de novo mutations (ncDNMs) identified in ASD probands from the Simons Simplex Collection (SSC) and the MSSNG cohort to assess their functional impact. Using integrative analyses that combined regulatory annotations, gene expression assays, and variant burden tests, they demonstrated that a subset of ncDNMs can significantly downregulate the expression of both previously established ASD risk genes and novel candidate genes. The findings highlight that non-coding variation, while less well studied than protein-coding mutations, exerts measurable effects on gene regulation and contributes to ASD pathogenesis. Moreover, by expanding the mutational spectrum beyond coding regions, this work provides new insights into the role of regulatory disruption in neurodevelopmental disorders and underscores the importance of incorporating ncDNMs into future ASD gene discovery efforts. The criteria for extraction of new Q3_2025 genes from this report required at least one de novo LoF variant in an ASD proband from previous studies, in addition to at least one ncDNM identified in Chen et al., 2025 2025.This filtration step resulted in the addition of seven genes: CXXC5, NPAS3, ARHGAP30, ZNF644, DENND2B, PTPRS, and DNAJC5.

1.4 Summary Evidence of New Genes

DOT1L

Maroni et al., 2025 identified a cohort of 16 individuals through collaborating clinicians and GeneMatcher with monoallelic DOT1L variants presenting with a variable neurodevelopmental disorder characterized by language delay (13/16), motor delay (9/16), intellectual disability (4/16), a diagnosis of ASD (4/16), seizures/epilepsy (3/16), and craniofacial anomalies (14/16). Additional functional studies in this report identified patient-specific missense variants with either decreased (the newly identified p.Asp157Asn variant) or increased (the previously reported p.Glu123Lys variant) methyltransferase activity. In mouse model, heterozygous forebrain-specific Dot1l conditional knockout mice demonstrated altered early vocalization development in both male and female pups, while a sex-specific sociability deficit in the three-chamber test in female Dot1l cKO mice. A previous study (Nil et al., 2023) had reported nine unrelated individuals with seven different de novo heterozygous missense variants in DOT1L through the Undiagnosed Disease Network (UDN), the SickKids Complex Care genomics project, and GeneMatcher presenting with some degree of global developmental delay/intellectual disability and at least one major congenital anomaly in most individuals. Subsequently, functional assessment of DOT1L missense variants in Drosophila and human cells in this report demonstrated gain-of-function effects in flies and increased H3K79 methylation levels in flies and human cells. A number of de novo DOT1L variants, including two de novo loss-of-function variants in probands from the Simons Simplex Collection, have been reported in ASD probands (Iossifov et al., 2014; Satterstrom et al., 2020; Zhou et al., 2022; Fu et al., 2022).

GABRA2

Heterozygous variants in GABRA2 are responsible for developmental and epileptic encephalopathy 78 (DDE78; OMIM 618557). Adamo-Croux et al., 2025 reported six new patients with GABRA2 variants identified through a French national collaboration; all six individuals presented with epilepsy and developmental delay, and four were reported to present with autism spectrum disorder. Maljevic et al., 2019 had previously identified a DDE78 patient with ASD and a de novo GABRA2 missense variant, as well as two siblings with an attenuated form of DEE78 caused by a mosaic paternally-inherited missense variant (the proband was diagnosed with ASD, while his sister did not have autistic features); functional analysis in Xenopus oocytes demonstrated that both of these ASD-associated missense variants resulted in a loss-of-function effect. Several de novo missense variants in this gene have been identified in ASD probands, including a missense variant that was absent in ExAC and gnomAD and predicted to be damaging by CADD, REVEL, and MPC in a SPARK proband (Takata et al., 2018; Zhou et al., 2022; Fu et al., 2022). Gabra2 interacts with collybistin, the protein encoded by the ARHGEF9 gene (Hines et al., 2018). Hines et al., 2022 found that mutating the collybistin-binding motif within the large intracellular loop of Gabra2 and replacing it with the binding motif for gephyrin from Gabra1 (Gabra2-1) resulted in strongly downregulated collybistin expression in addition to deficits in working and recognition memory, hyperactivity, anxiety, and reduced social preference, recapitulating the frequently reported features of patients with ARHGEF9 mutations.

CAPZA2

Hellman et al., 2024 reported a p.Arg295Leu missense variant in the CAPZA2 gene in a proband diagnosed with autism and presenting with developmental delay and intellectual disability; this missense variant had been previously reported in a proband presenting with developmental delay and a history of seizures in Huang et al., 2020. Guo et al., 2025 subsequently found that CAPZA2 heterozygous knockout mice demonstrated reduced expression in the hippocampus and prefrontal cortex, as well as exhibited motor dysfunction and anxiety-like behaviors, impairments in spatial and non-spatial memory, and deficits in social interactions; these phenotypes were mirrored in mice heterozygous for the human-specific Arg259Leu missense variant previously reported in Huang et al., 2020 and Hellman et al., 2024. Huang et al., 2020 also reported a proband with a p.Lys256Glu missense variant who was diagnosed with autism and presented with developmental delay and intellectual disability; functional studies of this variant in Drosophila demonstrated a reduced ability to rescue the lethality phenotype in flies that were null for cpa (the CAPZA orthologue in Drosophila), consistent with a partial loss-of-function effect.

MAPK8IP3

In a report describing 32 individuals with pathogenic or likely pathogenic MAPK8IP3 variants recruited through the Cure MAPK8IP3 Foundation, Sudnawa et al., 2025 found that, in addition to phenotypes frequently associated with neurodevelopmental disorder with or without variable brain abnormalities (NEDBA; OMIM 618443), autism was observed in 31.3% of individuals in this cohort. Previous reports describing individuals with NEDBA found a diagnosis of autism spectrum disorder in 2/13 patients in Platzer et al., 2019 and autistic behavior in 2/5 patients in Iwasawa et al., 2019; both reports also demonstrated functional effects of patient-associated MAPK8IP3 variants in C. elegans and zebrafish. A number of de novo missense variants in the MAPK8IP3 gene have been reported in ASD probands, including a p.Tyr94Cys variant originally identified in an SSC proband that was experimentally shown to result in an adverse locomotion phenotype in C. elegans in Platzer et al., 2019 (Iossifov et al., 2014; Yuen et al., 2017; Zhou et al., 2022; Trost et al., 2022), while a de novo MAPK8IP3 nonsense variant was identified in a Chinese ASD proband in Wang et al., 2023.

TBCB

Morag et al., 2025 described a cohort of ten individuals from eight families of Ashkenazi descent, all with the same homozygous missense variant in the TBCB gene (p.Tyr197Asn) and presenting with a neurodevelopmental disorder characterized by global developmental delay, autism spectrum disorder, and late childhood-onset spastic paraparesis; patient-derived fibroblasts displayed reduced TBCB expression. Additional functional assessment of the p.Tyr197Asn variant using the S. cerevisiae orthologue ALF1 found that mutant ALF1 resulted in increased benomyl sensitivity in yeast, resembling a loss-of-function phenotype, while the homologous mutant in Drosophila led to reduced survival and impaired climbing ability. De novo missense variants in TBCB have also been identified in ASD probands from the Simons Simplex Collection and the SPARK cohort (Iossifov et al., 2014; Zhou et al., 2022).

CXXC5

Chen et al., 2025 integrated cortex cell-specific cis-regulatory element annotations, a deep learning-based variant prediction model, and massively parallel reporter assays to systematically evaluate the functional impact of 227,878 non-coding de novo mutations (ncDNMs) in ASD probands from Simons Simplex Collection (SSC) and Autism Speaks MSSNG resource (MSSNG) cohorts and identified ncDNMs that down-regulated expression of the CXCC5 gene in two SSC probands. Additional de novo variants in this gene, including a loss-of-function variant and two missense variants, have been identified in ASD probands (De Rubeis et al., 2014; Zhou et al., 2022; Fu et al., 2022).

CBX4

To evaluate the effects of ASD-associated de novo variants in a family relative context, Kim et al., 2025 defined within-family standardized deviations (WFSD) by subtracting phenotype scores of unaffected family members and standardizing the result in 21,735 families from three ASD cohorts (the Korean Autism cohort, the Simons Simplex Collection, and SPARK); their analysis found that more genes enriched in de novo damaging protein-truncating variants (LOEUF < 0.37) and missense variants (MPC > 2) were identified using WFSD compared to raw phenotype scores, with 38 genes uniquely identified in the WFSD group, including the CBX4 gene. A de novo loss-of-function variant in CBX4 was identified in an SSC proband in Iossifov et al., 2014, while a de novo missense variant with a MPC > 2 was identified in a MSSNG proband in Zhou et al., 2022.

NPAS3

Chen et al., 2025 integrated cortex cell-specific cis-regulatory element annotations, a deep learning-based variant prediction model, and massively parallel reporter assays to systematically evaluate the functional impact of 227,878 non-coding de novo mutations (ncDNMs) in ASD probands from Simons Simplex Collection (SSC) and Autism Speaks MSSNG resource (MSSNG) cohorts and identified a ncDNM that down-regulated expression of the NPAS3 gene in a SSC proband. Additional de novo variants, including a loss-of-function variant and two missense variants, have been identified in the NPAS3 gene in ASD probands (De Rubeis et al., 2014; Satterstrom et al., 2020; Fu et al., 2022; Trost et al., 2022). Li et al., 2022 found that Npas3 deficiency in mice resulted in impaired cortical astrogenesis, which correlated with abnormal brain development and autistic-like behaviors. Michaelson et al., 2017 found that Fmr1 and Ube3a were transcriptionally regulated by NPAS3, as was the neurogenesis regulator Notch. Rare coding variants in NPAS3, including a frameshift variant that was experimentally shown to result in loss of transcriptional activity, had been previously reported in individuals with NDDs from the Baylor Genetics clinical exome sequencing database in Rossi et al., 2021.

ARHGAP30

Chen et al., 2025 integrated cortex cell-specific cis-regulatory element annotations, a deep learning-based variant prediction model, and massively parallel reporter assays to systematically evaluate the functional impact of 227,878 non-coding de novo mutations (ncDNMs) in ASD probands from Simons Simplex Collection (SSC) and Autism Speaks MSSNG resource (MSSNG) cohorts and identified a ncDNM that down-regulated expression of the ARHGAP30 gene in a SSC proband. A de novo loss-of-function variant, a de novo missense variant, and a de novo in-frame deletion variant have also been identified in the ARHGAP30 gene in ASD probands (Iossifov et al., 2014; Zhou et al., 2022).

ZNF644

Chen et al., 2025 integrated cortex cell-specific cis-regulatory element annotations, a deep learning-based variant prediction model, and massively parallel reporter assays to systematically evaluate the functional impact of 227,878 non-coding de novo mutations (ncDNMs) in ASD probands from Simons Simplex Collection (SSC) and Autism Speaks MSSNG resource (MSSNG) cohorts and identified a ncDNM that down-regulated expression of the ZNF644 gene in a MSSNG proband. A de novo loss-of-function variant and a de novo missense variant have also been identified in the ZNF644 gene in ASD probands (Iossifov et al., 2014; Tan et al., 2024).

DENND2B

Chen et al., 2025 integrated cortex cell-specific cis-regulatory element annotations, a deep learning-based variant prediction model, and massively parallel reporter assays to systematically evaluate the functional impact of 227,878 non-coding de novo mutations (ncDNMs) in ASD probands from Simons Simplex Collection (SSC) and Autism Speaks MSSNG resource (MSSNG) cohorts and identified a ncDNM that down-regulated expression of the DENND2B gene in a MSSNG proband. Additional de novo variants, including a loss-of-function variant and several

missense variants, have been identified in this gene in ASD probands (Satterstrom et al., 2020; Zhou et al., 2022; Tan et al., 2024). Murthy et al., 2025 described 11 individuals with monoallelic variants in DENND2B with a shared constellation of features (developmental delay, intellectual disability and psychiatric/behavioral concerns, and episodes of psychosis and/or catatonia); 3/8 were reported to have an ASD diagnosis, and nine of the ten observed patient variants were confirmed to result in loss of DENND2B function in zebrafish.

PTPRS

Chen et al., 2025 integrated cortex cell-specific cis-regulatory element annotations, a deep learning-based variant prediction model, and massively parallel reporter assays to systematically evaluate the functional impact of 227,878 non-coding de novo mutations (ncDNMs) in ASD probands from Simons Simplex Collection (SSC) and Autism Speaks MSSNG resource (MSSNG) cohorts and identified a ncDNM that down-regulated expression of the PTPRS gene in a MSSNG proband. Additional de novo variants in the PTPRS gene, including a loss-of-function variant and four missense variants, have been identified in ASD probands (Iossifov et al., 2014; Krumm et al., 2015; Satterstrom et al., 2020; Zhou et al., 2022; Fu et al., 2022; Trost et al., 2022).

DNAJCS

Chen et al., 2025 integrated cortex cell-specific cis-regulatory element annotations, a deep learning-based variant prediction model, and massively parallel reporter assays to systematically evaluate the functional impact of 227,878 non-coding de novo mutations (ncDNMs) in ASD probands from Simons Simplex Collection (SSC) and Autism Speaks MSSNG resource (MSSNG) cohorts and identified a ncDNM that down-regulated expression of the PTPRS gene in a MSSNG proband. Additional de novo variants in the PTPRS gene, including a loss-of-function variant and four missense variants, have been identified in ASD probands (Iossifov et al., 2014; Krumm et al., 2015; Satterstrom et al., 2020; Zhou et al., 2022; Fu et al., 2022; Trost et al., 2022).

CTPS1

To evaluate the effects of ASD-associated de novo variants in a family relative context, Kim et al., 2025 defined within-family standardized deviations (WFSD) by subtracting phenotype scores of unaffected family members and standardizing the result in 21,735 families from three ASD cohorts (the Korean Autism cohort, the Simons Simplex Collection, and SPARK); their analysis found that more genes enriched in de novo damaging protein-truncating variants (LOEUF < 0.37) and missense variants (MPC > 2) were identified using WFSD compared to raw phenotype scores, with 38 genes uniquely identified in the WFSD group, including the CTPS1 gene. A de novo loss-of-function variant in the CTPS1 gene was reported in a MSSNG proband in Zhou et al., 2022, while a de novo missense variant with a MPC > 2 was identified in a Korean ASD proband in Kim et al., 2024.

NMT1

To evaluate the effects of ASD-associated de novo variants in a family relative context, Kim et al., 2025 defined within-family standardized deviations (WFSD) by subtracting phenotype scores of unaffected family members and standardizing the result in 21,735 families from three ASD cohorts (the Korean Autism cohort, the Simons Simplex Collection, and SPARK); their analysis found that more genes enriched in de novo damaging protein-truncating variants (LOEUF < 0.37) and missense variants (MPC > 2) were identified using WFSD compared to raw phenotype scores, with 38 genes uniquely identified in the WFSD group, including the NMT1 gene. De novo loss-of-function variants in NMT1 have been reported in ASD probands from the Simons Simplex Collection and the MSSNG cohort, and a de novo missense variant in this gene was reported in an ASD proband from the Autism Sequencing Consortium (Iossifov et al., 2014; Satterstrom et al., 2020; Trost et al., 2022).

HNRNPL

To evaluate the effects of ASD-associated de novo variants in a family relative context, Kim et al., 2025 defined within-family standardized deviations (WFSD) by subtracting phenotype scores of unaffected family members and standardizing the result in 21,735 families from three ASD cohorts (the Korean Autism cohort, the Simons Simplex Collection, and SPARK); their analysis found that more genes enriched in de novo damaging protein-truncating variants (LOEUF < 0.37) and missense variants (MPC > 2) were identified using WFSD compared to raw phenotype scores, with 38 genes uniquely identified in the WFSD group, including the HNRNPL gene. Zhou et al., 2022 reported a de novo loss-of-function variant in the HNRNPL gene in an SSC proband and a likely deleterious de novo missense variant in this gene in a MSSNG proband (Zhou et al., 2022).

KDM1A

To evaluate the effects of ASD-associated de novo variants in a family relative context, Kim et al., 2025 defined within-family standardized deviations (WFSD) by subtracting phenotype scores of unaffected family members and standardizing the result in 21,735 families from three ASD cohorts (the Korean Autism cohort, the Simons Simplex Collection, and SPARK); their analysis found that more genes enriched in de novo damaging protein-truncating variants (LOEUF < 0.37) and missense variants (MPC > 2) were identified using WFSD compared to raw phenotype scores, with 38 genes uniquely identified in the WFSD group, including the KDM1A gene. A de novo loss-of-function variant in the KDM1A gene was reported in a SPARK proband in Zhou et al., 2022, and de novo missense variants with MPC >2 in this gene were reported in an SSC proband and a proband from the Korean Autism cohort in Iossifov et al., 2014 and Kim et al., 2024, respectively.

HG_Table 2. Selected examples of new studies added to existing genes in Q3/2025

Gene	Title	First Author	Journal/Book	Publication Year
BCL11A	Bcl11a deficiency in cerebellar Purkinje cells causes ataxia and autistic-like behavior by altering Vav3	Zhang J	Mol Psychiatry	2025
BCKDK	BCKDK gene mutations as a rare condition responsible for comorbid neurodevelopmental delay, autism, and epilepsy: a case series of four patients	Karimzadeh P	Ann Med Surg (Lond)	2025
SHANK3	Shank3 oligomerization governs material properties of the postsynaptic density condensate and synaptic plasticity	Jia B	Cell	2025
CHD3	Neurobehavioral profile of individuals with pathogenic variants in CHD3	Ionescu A	Eur J Hum Genet	2025
FOXP1, FOXP2	FOXP genes regulate Purkinje cell diversity and cerebellar morphogenesis	Khouri-Farah N	Nat Neurosci	2025
CACNA1I	Two pairs of CACNA1I (CaV3.3) variants with opposite effects on channel function cause neurodevelopmental disorders of varying severity	El Ghaleb Y	PLoS Genet	2025
CUL3	Short Stature and Response to Growth Hormone Treatment in CUL3-Related Neurodevelopmental Disorder	Loid P	Horm Res Paediatr	2025
CLCN4	CLCN4-Related Neurodevelopmental Condition: Characterization of Speech and Language Abilities	Garrett A	Am J Med Genet A	2025
BMAL1	Rare variants in BMAL1 are associated with a neurodevelopmental syndrome	Cuddapah VA	Proc Natl Acad Sci U S A	2025
RNU4-2	Identification of a pathogenic RNU4-2 variant in patients with mitochondrial disease: Broadening the spectrum of non-coding RNA gene variants in mitochondrial dysfunction	Nakamura K	J Hum Genet	2025
SCN2A	Development and Adaptive Function in Individuals With SCN2A-Related Disorders	Goad BS	Neurology	2025
CTNNB1	Genotypic, functional, and phenotypic characterization in CTNNB1 neurodevelopmental syndrome	Žakelj N	HGG Adv	2025

1.5 Human Gene Standardization

We continue to standardize the allele change and residue change data fields to the terminology developed by the Human Genome Variation Society (HGVS) for the Human Gene dataset. All new variant annotations were performed with standardized terminology and include genomic coordinates in GRCh38 genome build for allele change, residue change and correct genome build.

1.5 Development of a Candidate Gene Pool

The Single Line of Evidence (SLOE) dataset comprises genes and variants reported in the scientific literature that lack sufficient evidence to meet the established inclusion criteria for the database. This dataset serves as a repository for genetic findings that may have potential relevance but have not been fully validated or sufficiently supported by robust data. Regular queries of the SLOE dataset play a crucial role in gene selection process by facilitating cross-referencing and validating newly identified ASD candidate genes. By systematically examining the SLOE dataset, we compare recently discovered variants against existing evidence, allowing for a thorough assessment of their relevance and significance.

In Q3/2025, we updated SLOE dataset by adding eight new articles, detailed in HG_Table 3.

HG_Table 3: New articles added to the SLOE dataset.

PMID	Title	Author, Year
40592404	Diagnostic yield of clinical exome sequencing in 868 children with neurodevelopmental disorders	Neuens , 2025
40558542	Exome Study of Single Nucleotide Variations in Patients with Syndromic and Non-Syndromic Autism Reveals Potential Candidate Genes for Diagnostics and Novel Single Nucleotide Variants	Belenska-Todorova, 2025
40642607	Unveiling Hidden Genetic Architectures: Molecular Diagnostic Yield of Whole Exome Sequencing in 50 Children With Autism Spectrum Disorder Negative for Copy Number Variations	Wang, 2025
40731902	Clinical Application of a Customized Gene Panel for Identifying Autism Spectrum Disorder-Associated Variants	Greco, 2025
40738258	Massively parallel characterization of non-coding de novo mutations in autism spectrum disorder	Chen, 2025
40756852	Genetic diagnostic outcomes from a 10-year research programme in autism in Aotearoa New Zealand	Musgrave, 2024
40819013	Genetic Heterogeneity of Autism Spectrum Disorder: Identification of Five Novel Mutations (RIMS2, FOXG1, AUTS2, ZCCHC17, and SPTBN5) in Iranian Families via Whole-Exome and Whole-Genome Sequencing	Mirahmadi, 2025
40841582	Prospective study to analyze the yield and clinical impact of trio exome sequencing in 137 Indian children with autism spectrum disorder	Bajaj, 2025