Next-generation sequencing identifies novel gene variants and pathways involved in specific language impairment


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SUPPLEMENTARY FIGURE S1 - Coverage versus read depth of 43 SLI samples
SUPPLEMENTARY FIGURE S2 - Rare and Novel Variants in Candidate Genes

Variants are shown in map order with ‘+’ denoting variant carrier. Variants of putative significance are shown in Figure 1 of the main manuscript. Variants that co-occur with other variants of putative significance are shown in Figure 3 of the main manuscript.

chr2: 125504881, Proband 39 – CNTNAP5, V717A
Variant was not genotyped in family (EVS_EA>1%)

chr3:78766524, Proband 17 – ROBO1, V234A

chr7:69364311, Proband 19 – AUTS2, R117C
Pedigree shown in Figure 3

chr7:146829358, Proband 40 – CNTNAP2, V369L

chr12: 1137072, Proband 23 – ERC1, M1I
Pedigree shown in Figure 1

Both parents self-report a history of speech and language difficulties
SUPPLEMENTARY FIGURE S2 (cont.) - Rare and Novel Variants in Candidate Genes

chr12:13715865, Proband 25 – *GRIN2B*, G1436A

chr15:48063365, Proband 30 – *SEMA6D*, H807D
Pedigree shown in Figure 3

chr16:9916226, Proband 4 – *GRIN2A*, G688A
Pedigree shown in Figure 1

chr16:84438827, Proband 35 – *ATP2C2*, V102M

chr16:84494315, Probands 27, 36 & 39 – *ATP2C2*, R646W
Variant was not genotyped in families (EVS_EA>1%)

chrX:99922289, Proband 41 – *SRPX2*, N327S
Pedigree shown in Figure 1
SUPPLEMENTARY FIGURE S3 - STOP-gain variants identified in SLIC probands

Variants are shown in map order with ‘+’ denoting variant carrier. Stop-gains that cosegregate with disorder are shown in Figure 2 of the main manuscript. Variants that co-occur with other variants of putative significance are shown in Figure 3 of the main manuscript.

chr1:158532597, Proband 34 – OR6P1, Y266X

chr3:63466576, Proband 30 – SYNPR, C31X
Pedigree shown in Figure 3

chr8:39847306, Proband 11 – IDO2, R219X

chr8:107738486, Proband 29 – OXR1, W5X
Pedigree shown in Figure 2
SUPPLEMENTARY FIGURE S3 (cont.) - STOP-gain variants identified in SLIC probands

chr11:1027390, Proband 8 – MUC6, C703X
Pedigree shown in Figure 2

chr11:6190828, Proband 19 – OR52B2, C243X
Pedigree shown in Figure 3

chr16:4745030, Proband 9 – NUDT16L1, Q186X

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No sample available for Mother or Sib
SUPPLEMENTARY FIGURE S4 - Probands with multiple novel or rare variants in one gene
We focus here on potential compound heterozygotes only. Variants are shown in map order with '-' denoting homozygote wildtype and '+' denoting variant carrier. Pedigrees in which the variants were inherited from opposite parents and co-segregated with disorder in the children are shown in Figure 3 of the main manuscript.

chr2:32689842&chr2:32740353, Probands 26&42 – BIRC6, P1736L&A3622V
Both variants inherited from single parent


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Both parents self-report a history of speech and language difficulties

chr3:58104626&chr3:58110119, Proband 40 – FLNB, G925C&G1262A

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Both parents self-report a history of speech and language difficulties

Pedigree shown in Figure 3
SUPPLEMENTARY FIGURE S4 (cont) - Probands with multiple novel or rare variants in one gene

chr11:92086828&chr11:92624235, Proband 7 – FAT3, L517S&L4544F

chr12:49418717&chr12:49432365, Proband 12 – KMT2D, R5266H&A2925V
Pedigree shown in Figure 3


chr14:58924684&chr14:59014632, Proband 19 – KIAA086, L577I&G1625R
Pedigree shown in Figure 3

chr15:42977116& chr15:42981101& chr15:42982237, Proband 12 – STARD9, C1114R&P2442R&V2821L
Pedigree shown in Figure 3

chr15:42977810& chr15:42978141& chr15:42981101& chr15:42982237, Proband 19 – STARD9, I1345S&E1455D&P2442R&V2821L
Pedigree shown in Figure 3
SUPPLEMENTARY FIGURE S4 (cont) - Probands with multiple novel or rare variants in one gene

chr16:23635348&chr16:23641275, Proband 13 – PALB2, L939W&T734S

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chr17:34861135&chr17:34871802, Proband 25 – MYO19, splice donor lost &Y149C
Both variants inherited from single parent