|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Gene** | **OMIM** | **Function** | **Associated phenotype (OMIM)** | **Inheritance** |
| *FA2H* | \*611026 | structural diversity of sphingolipids | spastic paraplegia 35 (#612319) | AR |
| *GAN* | \*605379 | neurofilament architecture, neuronal maintenance and survival, ubiquitination | giant axonal neuropathy-1 (#256850) | AR |
| *GCSH* | \*238330 | member of the glycine cleavage system | glycine encephalopathy (#605899) | AR |
| *KARS1* | \*601421 | aminoacylation of tRNA-lys | Charcot-Marie-Tooth disease, recessive intermediate, B (#613641)  AR deafness 89 (#613916) | AR  AR |
| *TMEM231* | \*614949 | important component of cilia | Joubert syndrome 20 (#614970)  Meckel syndrome 11 (#615397) | AR  AR |
| *CFDP1* | \*608108 | maintenance of chromatin organization, role in embryogenesis | - | - |
| *CMIP* | \*610112 | plays a role in T-cell signaling pathway | linked to autism spectrum disorder | - |
| *CNTNAP4* | \*610518 | member of the neurexin family, structural maturation of inhibitory interneuron synapses | epilepsy and attention deficit hyperactivity disorder | - |
| *PKD1L2* | \*607894 | member of the polycystin protein family | linked to neuromuscular disease in mice | - |

**Supplementary Table 1. Genes encompassed within 16q22.2q23.3 associated with neurodevelopmental disorders**OMIM: Online Inheritance in Man; AR: autosomal recessive