**S2 Table.** Gene function and clinical phenotypes associated with the 18 protein-coding genes with more than one read > 5 RPKM but with overall average gene expression <5. Developmental time period when RPKM >5 also shown. (1: 16 pcw – 17 pcw, 2: 19 pcw – 24 pcw, 3: 4 mos – 1 yr, 4: 2 yrs – 4 yrs, 5:8 yrs – 13 yrs, 6: 15 yrs – 21 yrs, 7: 23 yrs – 40 yrs). (pcw = post-conception weeks, mos = months, yrs = years).

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **gene symbol** | **OMIM number [1]** | **Developmental time period when RPKM >5** | **Function of protein product** | **associated disease** | **Associated neurologic phenotype** | **Other associated phenotype** | **Ref** |
| PNPLA3 | 609567 | 1 | Unknown | Variants associated with non-alcoholic fatty liver disease and alcoholic cirrhosis | speech abnormalities, absence of ASD | Hair pulling, male genital anomalies | [2-4] |
| PRR5 | 609406 | 2,3 | Component of the mammalian target of rapamycin 2 (mTOR2 complex) | Unknown | Unknown | Unknown | [5] |
| PHF21B | 616727 | 1,2 | Nuclear protein that functions to reduce cell migration and colony formation when overexpressed | Implicated as tumor suppressor gene in head and neck cancers | speech abnormalities, absence of ASD, facial asymmetry, abnormal reflexes | Hair pulling, male genital anomalies, dysplastic toenails, large hands | [2, 6] |
| NUP50 | 604646 | 1-3, 5-6 | Nucleoprotein that is part of the nuclear pore complex that allows bidirectional flow of macromolecules through nuclear envelope | Unknown | speech abnormalities, absence of ASD, facial asymmetry, abnormal reflexes, neonatal hypotonia | Hair pulling, male genital anomalies, dysplastic toenails, large hands, tall stature | [2, 7] |
| FAM118A | n/a | 2, 4 | Unknown | Unknown | Neonatal hypotonia, speech abnormalities, absence of ASD, facial asymmetry, abnormal reflexes | Dysplastic toenails, large hands, tall stature, hair pulling, male genital anomalies | [2] |
| PPARA | 170998 | 2,4 | Nuclear transcription factor that regulates expression of fatty acid oxidation enzymes | Increased susceptibility to hyperapobetalipoproteinemia | Neonatal hypotonia, speech abnormalities, absence of ASD, macrocephaly | Dysplastic toenails, large hands, late to walk, hair pulling, male genital anomalies | [2, 8] |
| TTC38 | n/a | 2,4 | Unknown | Unknown | Neonatal hypotonia, speech abnormalities, absence of ASD, macrocephaly | Dysplastic toenails, large hands, late to walk, hair pulling, male genital anomalies | [2] |
| GTSE1 | 607477 | 1 | Protein localized to microtubules and functions in cell cycle | Unknown | Neonatal hypotonia, speech abnormalities, absence of ASD, macrocephaly | Dysplastic toenails, large hands, late to walk, hair pulling, male genital anomalies | [2, 9] |
| CELSR1 | 604523 | 1 | Helps establish polarity of hair follicles in anterio-posterior axis | Increased susceptibility to neural tube defects | Neonatal hypotonia, speech abnormalities, absence of ASD, macrocephaly | Dysplastic toenails, large hands, late to walk, hair pulling, male genital anomalies | [2, 10-12] |
| TBC1D22A | 616879 | 2, 5-7 | Golgi protein predicted to function as GTPase- activating protein for RAB33 | Unknown | Neonatal hypotonia, absence of ASD, macrocephaly | Dysplastic toenails, large hands, late to walk, hair pulling | [2, 13] |
| ALG12 | 607144 | 1-6 | Catalyzes the addition of the 8th mannose residue onto the lipid-linked oligosaccharide precursor during the synthesis of complex oligosaccharide-linked glycoproteins within the Golgi apparatus and endoplasmic reticulum | Congenital disorder of glycosylation, type Ig | Neonatal hypotonia, microcephaly | Late to walk, anorexia, psychomotor retardation, facial dysmorphism, failure to thrive, blindness, deafness, male genital hypoplasia, cardiac abnormalities, generalized edema | [2, 14, 15] |
| CRELD2 | 607171 | 1-4 | Transmembrane protein with epidermal-like growth factor repeats that mediate protein-protein interactions | Unknown | Neonatal hypotonia | Late to walk | [2, 16] |
| TRABD | n/a | 1-4, 6-7 | Unknown | Unknown | Neonatal hypotonia | Unknown | [2] |
| HDAC10 | 608544 | 1-4, 7 | Histone deacetylase | Unknown | Neonatal hypotonia | Unknown | [2, 17] |
| MAPK12 | 602399 | 1-7 | Part of mitogen-activated protein kinase family of proteins that mediate extracellular signaling | Unknown | Neonatal hypotonia | Unknown | [2, 18] |
| TYMP | 131222 | 2 | Catalyzes the phosphorylation of thymidine to thymine, promotes neuronal survival, chemotactic for endothelial cells acting as an angiogenic factor | Mitochondrial DNA depletion syndrome 1 | Polyneuropathy, leukoencephalopathy | Muscle atrophy, gastrointestinal dysmotility, external opthalmoplegia, ptosis | [19, 20] |
| CPT1B | 601987 | 1-7 | Mitochondrial carnitine palmitoyltransferase which allows the transport of long-chain fatty acyl-CoA’s from cytoplasm to the mitochonidrial membrane for beta-oxidation | Unknown | Unknown | Cold intolerance | [21] |
| RABL2B | 605413 | 1-2 | Member of RAB family of proteins, which are a group a GTP-binding proteins involved in exocytic and endocytic pathways. Also has role in intraflagellar transport and ciliogenesis | Unknown | Unknown | Infertility, polydactyly, retinal degeneration | [22, 23] |

(ref = references; ASD= autism spectrum disorder; ID= intellectual disability)

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