**e-Table 2**

***ACO2* homozygous missense mutation associated**

**with complicated hereditary spastic paraplegia**

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| **Table 2 Genotypes and phenotypes of *ACO2* mutations** | | |  |  |
| **Mutation** | **Zygosity** | **Clinical Phenotype** | **Families described** | **Reference** |
| Leu74Val | Heterozygous (only observed in combination with Gly661Arg) | Isolated optic neuropathy | 1 | 29 |
| Ser112Arg | Homozygous | Developmental delay, cerebellar ataxia & cerebellar atrophy | 2 | 28 |
| Gly259Asp | Homozygous | Episodes of central apnea, optic neuropathy & cerebellar atrophy | 1 | 29 |
| Val364Ala | Heterozygous (only observed in combination with Leu776Asnfs\*49) | Developmental delay, optic neuropathy & cerebellar ataxia | 1 | 31 |
| Phe414Val | Homozygous | Spastic paraplegia, complicated by microcephaly and intellectual disability; cerebellar atrophy without clinical cerebellar signs (in one sibling) | 1 | This paper |
| Arg607Cys | Heterozygous (only observed in combination with Pro712Leu) | Developmental delay, auditory neuropathy, cerebellar ataxia without optic neuropathy | 1 | 30 |
| Gly661Arg | Heterozygous (only observed in combination with Leu74Val) | Isolated optic neuropathy | 1 | 29 |
| Pro712Leu | Heterozygous (only observed in combination with Arg607Cys) | Developmental delay, auditory neuropathy, cerebellar ataxia without optic neuropathy | 1 | 30 |
| Leu736Asn | Heterozygous (only observed in combination with Leu776Asnfs\*49) | Developmental delay, optic neuropathy & cerebellar atrophy | 1 | 29 |
| Leu776Asnfs\*49 | Heterozygous (only observed in combination with Leu736Asn & Val364Ala) | Developmental delay, optic neuropathy & cerebellar atrophy | 2 | 29 & 31 |