**Supplementary Table 1: Summary of previous studies of vanishing white matter disease**

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| --- | --- | --- | --- | --- | --- | --- |
| **Author, Year/ Country** | **Number of patients** | **Gender** | **Age at onset** | **Phenotype** | **Mean/median Follow up duration** | **Outcome** |
| **Maletkovic et al., 20081****Multicenter** | 15 | M=9, F=6 | 6 months-38 years | Progressive spastic ataxia (9), paroxysmal deterioration (9), chorea (1), tremor (1), seizures (1), torticollis (1) | NA | NA |
| **Labauge et al., 20092****France** | 16 | M=3, F=13 | 16-62 years | Cerebellar ataxia (9), cognitive decline (8), ovarian failure (8), spastic paraplegia or tetraplegia (7), seizures (7), stress induced worsening (6), psychiatric symptoms (2), subcortical gait (1) | 11.2 years (2-22 years) | Died (2), unable to walk independently (11), walk independently (3) |
| **Robinson et al., 20143****Canada** | 5 | M=2, F=3 | 25 months-20 years | Spasticity (5), ataxia (4), transient focal deficits accompanied by headache (3), dysarthria (2), tremor (1), cognitive decline (1) | 11.7 years | CSD 3 (3), CSD 2 (1), CSD 1 (1) |
| **Turon-Vinas et al., 20144****Spain** | 21 | M=8, F=13 | 1.5-8 years | Spasticity (21), ataxia (16), seizures (9), cognitive impairment (12), hemiparesis (1), dystonia (1), ptosis (1), optic atrophy (4), macrocephaly (3), episodic deterioration (21) | NA | Unable to walk (16), able to walk (5) |
| **Zhang et al., 2015****China5** | 34 | M=19, F=15 | 4 months-9 years 7 months | Progressive motor deterioration (34), episodic aggravation (24), seizures (16) | 4.4 years (3 months-12 years) | Died (10) |
| **Hamilton et al., 20186****Multicenter** | 296 | M=134, F=162 | Antenatal-54 years | Spasticity (175), ataxia (153), hypotonia (69), extrapyramidal signs (29), speech disturbance (125), dysphagia (70), impaired vision (64), impaired hearing (18), impaired cognition (89), seizures (162), exacerbating course (213), psychiatric symptoms (44), headache (26), dizziness (6), peripheral neuropathy (3), scoliosis (7), microcephaly (21), macrocephaly (16), ovarian failure (48), oligozoospermia (1), congenital cataract (4), renal hypodysplasia (2), hepatomegaly/hepatosplenomegaly (2)Asymptomatic (7) | NA | Loss of walking without support was seen in 76.5% of patients with disease onset < 18 years of age and in 60% of patients with disease onset ≥ 18 years of age. Median time to loss of walking without support was 4.8 years in patients with disease onset < 18 years and 7 years in patients with disease onset ≥ 18 years age.  |
| **Güngör et al., 20207****Turkey** | 11 | M=6, F=5 | 35.18±19.13 months | Gait disturbance (5), seizures (2), developmental delay (2), speech disturbance (1), dystonia (1) | 32.73 ± 16.42 months | Died (2), walk independently (3, unable to walk independently (3) |

‘CSD’: clinical score of disability (by Ohlenbusch et al.,2005), ‘F’: female, ‘M’: male, ‘NA’: data not available

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