

**Supplementary Table 1.** Clinical characteristics and treatment response in patients with AVED and dystonia

Study	Age at onset (yr)	Initial symptoms	Age at dystonia onset (yr)	Dystonia distribution	Vitamin E treatment (target dose/day; total duration)	Clinical course
Krendel et al., <sup>1</sup> 1987	7	Gait difficulty, clumsiness, dysarthria	19	GD, Neck, UE, LE	3600 IU; 5 months	Stable
Stumpf et al., <sup>2</sup> 1987	4	Unsteady gait	30	Face	800 mg; 6 months	Stable
Amiel et al., <sup>3</sup> 1995	7	Ataxia	16	Face, UE	1000 mg; 5 years	Improved: social contact, speech
Jackson et al., <sup>4</sup> 1996	7	Head and hand tremor	22	GD, Face neck, Trunk, LE	1200 IU; 3 years	Stable
Yokota et al., <sup>5</sup> 1997	30	ND	ND	ND	ND	Stable
Cavalier et al., <sup>6</sup> 1998	3	ND	ND	ND	ND	ND
Schuelke et al., <sup>7</sup> 1999	9	Clumsiness	13	ND	40 mg/kg; 18 months	Improved: psychiatric, cognitive, ataxia, dysarthria, sensation, deep tendon reflexes
Angelini et al., <sup>8</sup> 2002	8	Cervical myoclonus-dystonia	At onset	Neck, Trunk	2000 mg; 1.75 years	Improved: ataxia, dystonia, myoclonus
Roubertie et al., <sup>9</sup> 2003	8.6	Gait instability, slurred/scanning speech	11.75	GD, Neck, Trunk, UE, LE	2000 mg; 5 years	Improved: ataxia, dystonia, myoclonus
Mariotti et al., <sup>10</sup> 2004	8	ND	ND	ND	2400 mg; 5 years	Improved: ataxia Onset: progressive dystonia
Becker et al., <sup>11</sup> 2016	11	Dystonic head tremor	At onset	Neck, UE	2400 IU; 10 months	Onset: ataxia
	11	Dystonic head tremor	At onset	GD, Trunk, UE, LE	2400 IU; 10 months	Improved: dysarthria, dysdiadochokinesia
Pradeep et al., <sup>12</sup> 2019	17	Head tremor	17	Neck	1400 IU; 2 years	No response
	18	Head tremor	18	Neck, UE	1400 IU; 2 years	No response
Zea Vera et al., <sup>13</sup> 2021	16	Gait difficulty	16	UE, LE	2000 IU; 2 years	Stable
Zhang et al., <sup>14</sup> 2022	14	Unsteady gait	18	Neck	1200 IU; 2 years	Stable
Current report	22	Dystonic head and left UE tremor	22	Neck, UE, trunk, LE	1200 IU; 3 months	Stable

AVED, ataxia with vitamin-E deficiency; GD, generalized dystonia; IU, international units; LE, lower extremity; mg, milligram; ND, not described; UE, upper extremity.

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