**TABLE e-3. AT PATIENTS WITH MILD/ATYPICAL CLINICAL PRESENTATION**

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| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **CASE**  | **ONSET****(yrs)** | **PRESENTING SYMPTOMS** | **NEUROLOGICAL OUTCOME** | **NON-NEUROLOGICAL SYMPTOMS** | **LAST CHECK****(yrs)** | **ATM** | **AFP** | ***ATM* GENOTYPE** | **CEREBRAL ABNORMALITIES** | **REFERENCE** |
| 1 | 6 | unsteady gait, dysarthria | ataxic gait, nystagmus, dysmetria, oculomotor apraxia, hypomimia, choreiform movements, hyporeflexia | oculartelangiectasias | 16 | 0% | elevated | homozygous 5653delA | moderate atrophy of the vermis and cerebellum | Alterman et al. AJMG, part A 2007, 143A: 1827-1834 |
| 2 | 9,5 | instable sitting position and tandem gait, dysarthria, drooling, dysdiadochokinesis |  | mild oculartelangiectasias | 9,5 | 0% | elevated | homozygous 5653delA | vermian atrophy | Idem |
| 3 | 13 | head/limb tremor,progressive gait imbalance | resting e postural tremor, dysarthria, ataxic gait, axonal sensory-motor polyneuropathy | ocular telangiectasias | 39 | 0% | 23 ng/ml (n. v.<9) | homozygous c.6325T>G | vermian atrophy | Silvestri et al. J Neurol 2010, 257:1738–1740 |
| 4 | 35 | gait difficulties, muscle weakness, cramps | ataxic gait, sensory-motor axonal neuropathy | oculartelangiectasias | 44 | 0% | 82 ng/ml (n. v.<9) | homozygous c.6325T>G | cerebellar atrophy | Idem |
| 5 | 13 | slowly progressive dystonia, irregular tremor | choreiform movements | gastric cancer,prostate cancer | 64 | n.a. | n.a. | homozygous6200 C>A | Purkinje cells loss (post-death examination) | Saunders-Pullman et al. Neurology 2012, 78:649–657 |
| 6 | 16 | slowly progressive dystonia, irregular tremor | choreiform movements | renal cancer | 56 | n.a. | n.a. | homozygous 6200 C>A | n.a. | Idem |
| 7 | 1 | slowly progressive dystonia, irregular tremor | choreiform movements | myeloid leukemia | 43 | n.a. | n.a. | homozygous 6200 C>A | n.a. | Idem |
| 8 | unknown | slowly progressive dystonia, irregular tremor |  | stomach cancer | 53 | n.a. | n.a. | homozygous 6200 C>A | n.a. | idem |
| 9 | unknown | slowly progressive dystonia, irregular tremor | choreiform movements |  | 52 | n.a. | n.a. | homozygous 6200 C>A | n.a. | Idem |
| 10 | 15 | slowly progressive dystonia, irregular tremor | choreiform movements |  | 26 | n.a | n.a | homozygous 6200 C>A | n.a. | Idem |
| 11 | 20 | slowly progressive dystonia, irregular tremor | choreiform movements |  | 40 | n.a. | n.a. | homozygous 6200 C>A | n.a. | Idem |
| 12 | 11 | slowly progressive dystonia, irregular tremor |  |  | 12 | n.a | n.a | homozygous 6200 C>A | n.a | Idem |
| 13 | 1 | slowly progressive dystonia, irregular tremor | choreiform movements,sensorimotor neuropathy, tremor | prostate cancer | 60 | traces | 22.6 µg/L(n.v.< 6.1) | homozygous 6200 C>A | mild vermian atrophy? | Idem |
| 14 | 12 | slowly progressive dystonia, irregular tremor | choreiform movements | stomach cancer | 53 | 0% | 84 µg/L(n.v.<11) | homozygous 6200 C>A | n.a. | Idem |
| 15 | 12 | slowly progressive dystonia, irregular tremor | choreiform movements, sensorimotor neuropathy |  | 51 | n.a. | n.a. | homozygous 6200 C>A | n.a. | Idem |
| 16 | 4 | slowly progressive dystonia, irregular tremor | choreiform movements, sensorimotor neuropathy (after vincristine) | lymphoma | 42 | n.a. | n.a. | homozygous 6200 C>A | n.a. | Idem |
| 17 | 2 | clumsiness, athetosis | choreoathetosis and dystonia,dysarthria, slight drooling | recurrent sinopulmonary infections | 25 | r.k.a. | 77.8 U/ml(n.v.<5) | c.8122G>Ac.8851-1G>T | normal | Claes et al. Neuromol Med 2013, 15:447–457 |
| 18 | 15 | cervical myoclonus | laryngeal dystonia, clawed fingers, postural hand tremor, choreiform movements | ocular telangiectasias | n.a. | n.a. | elevated | c.7886\_7890delc.6154 G>A | n.a. | Charlesworth et al. Neurology 2013, 81:1148–1151 |
| 19 | 13 | cervical dystonia, head tremor | postural hand tremor | ocular telangiectasias | n.a. | n.a. | elevated | c.7886\_7890del c.6154 G>A | n.a. | Idem |
| 20 | 11 | cervical dystoniahead tremor | postural hand tremor | ocular telangiectasias | n.a. | n.a. | elevated | c.7886\_7890del c.6154 G>A | n.a. | Idem |
| 21 | 5 | sluggish movements,difficulty in hand writing,mild dysarthria | spastic paraplegia, hyperreflexia, ataxia, myoclonic jerks, axial dystonic posture, oculomotor disorders, slurred speech |  | 12 | 0% | 468.2 ng/ml (n.v.0-20) | c.G467Ac.1121\_1122delAA | atrophy of thevermis and cerebellum | Nakayama et al. Brain Dev 2015,37:362-365 |
| 22 | 1-2 | uncertainly gait until 4–5 yrs., slurred speech, no progression until 25 yrs | cervical and right upper limb dystonia, head and hand tremor, intermittent involuntary choreatic movements | breast cancer, myoma | 40 | n.a. | elevated | p. V2716Ap. G301VfsX19 | normal | Lohmann et al. J Neurol 2015,262:1724–1727 |
| 23 | 1-2 | unsteady gait,slight slurred speech | dystonic postures, choreoathetosis,dysarthria, slurred speech, oculomotor apraxia |  | 37 | n.a. | 142 IU/mL (n.v.0-5.8) | p. V2716Ap. G301VfsX19 | normal | Idem |
| 24 | early childhood | brief contractionsparticularly in the upper limbs and the neck | intermittent episodes of extension of the fingers of the right hand |  | 34 | n.a. | elevated | pV2716Ap. G301VfsX19 | n.a. | Idem |
| 25 | teen | choreiform movements,dystonia | mild gait ataxia, cerebellar dysarthria and cognitive impairment,hypsometric saccades | ocular telangiectasias | 41 | 0% | 120.2 kU/L (normal value n.a.) | c.1290\_1291delTGc.5177þ5 G>A | midline cerebellar atrophy (42 yrs) | Worth et al. Mov Dis 2013, Vol. 28, No. 4 |
| 26 | early childhood | clumsy gait | generalized dystonia with prominent oromandibular involvement | pharyngeal telangiectasias | 18 | r.k.a. | 406 IU/ml (n.v. ≤ 30) | homozygous 590G>A (197G>Q) | mild cerebellar atrophy | Carrillo et al. Cerebellum 2009, 8:22–27 |
| 27 | 17 | mild ataxia | progressive balance and gait deterioration,supranuclear ophthalmoplegia impaired pursuit, nystagmus | ocular telangiectasias | 29 | r.k.a. | n.a. | homozygous 5762A3> G | moderate atrophy of the cerebellar hemispheres,severe atrophy of vermis (22 yrs) | Sutton et al. Ann Neurol 2004, 55:891–895 |
| 28 | 22 | balance disorders | progressive in coordination and balance, supranuclear ophthalmoplegia impaired pursuit, nystagmus | ocular telangiectasias | 28 | r.k.a. | n.a. | homozygous 5762A3>G | n.a. | Idem |
| 29 | 29 | unsteadiness of gait | prominent distal spinal muscular atrophy, slight cerebellar ataxia,mild dysarthria, severe resting tremor, slight intention tremor | minimal ocular telangiectasias | 38 | slightr.k.a. | elevated | 7622C>T3136C>T | normal | Hiel et al. Neurology, 2006, 67(2):346-349 |
| 30 | 30 | unsteadiness of gait | prominent distal spinal muscular atrophy,slight cerebellar ataxia,mild dysarthria, severe resting tremor, slight intention tremor | minimal ocular telangiectasias | 37 | slight r.k.a. | elevated | 7622C>T3136C>T | slight atrophy of the vermis superior | Idem |
| 31 | 37 | unsteadiness of gait | prominent distal spinal muscular atrophy,slight cerebellar ataxia,mild dysarthria, severe resting tremor, slight intention tremor | minimal ocular telangiectasias | 43 | n.a. | elevated | 7622C>T3136C>T | n.a. | Idem |
| 32 | 32 | unsteadiness of gait | prominent distal spinal muscular atrophy,slight cerebellar ataxia,mild dysarthria, severe resting tremor, slight intention tremor |  | 42 | slightr.k.a. | elevated |  | normal | Idem |
| 33 | 1-3 | mild motor delay,speech difficulties | learning disability,dysarthria | T-cell acute lymphoblastic leukemia | 17 | 0% | 117.9 ng/mL(n.v. 0.6 -3.9) | homozygous c.5585T>A | n.a. | Roohi et al. J Hum Genet, 2017, 62 (5): 581-584 |

Legend: Yrs: years; n.a: not available, n.v.: normal value, VOR vestibular ocular reflex, r.k.a.: reduced kinase activity, ATM: ataxia teleangiectasia mutated, AFP: alpha-fetoprotein