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

|  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **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 | ocular  telangiectasias | 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 ocular  telangiectasias | 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 | ocular  telangiectasias | 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. | homozygous  6200 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>A  c.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\_7890del  c.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 dystonia  head 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.G467A  c.1121\_1122delAA | atrophy of the  vermis 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. V2716A  p. 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. V2716A  p. G301VfsX19 | normal | Idem |
| 24 | early childhood | brief contractions  particularly in the upper limbs and the neck | intermittent episodes of extension of the fingers of the right hand |  | 34 | n.a. | elevated | pV2716A  p. 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\_1291delTG  c.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 | slight  r.k.a. | elevated | 7622C>T  3136C>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>T  3136C>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>T  3136C>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 | slight  r.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