**Table 1. Summary of clinical manifestations of all published variant AT cases with predominant dystonia**

|  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Families | Present study | Simonin et al.  (2008) | Carrillo et al.  (2009) | Saunders-Pullman et al.  (2012) | | | Meissner et al.  (2013) | Cummins et al.  (2013) | Kuhm C et al.  (2015) | Lohmann et al.  (2015) | Necpál et al. (2017) |
| Origin | Central-southern China | France | Indian | Canadian Mennonite | Canadian Mennonite | Canadian Mennonite | France | United Kingdom | Germany | Turkey | Germany |
| Affected | 2(1M, 1F) | 4(2M, 2F) | 1(F) | 6(4M, 2F) | 2(1M, 1F) | 4(1M, 3F) | 4(2M, 2F) | 1(M) | 1(M) | 3(M) | 1(F) |
| AAO, y | 9,12 | 12-20 | 15 | 1-16 | 11-20 | 1-12 | Adolescence to 18 | 2 | childhood | 1-5 | 12 |
| Dystonic features | Craniocervical | 3/4 | Craniocervical | generalized | Craniocervical | generalized | generalized | generalized | generalized | Neck, upper limb, trunk | Neck, legs, trunk |
| Chorea | 1/2 | 2/4 | - | 5/6 | 1/2 | 4/4 | - | - | - | 1/3 | - |
| Cerebellar ataxia | - | 4/4 | - | - | - | - | - | 1 | - | 1/3 | - |
| Telangiectasia | - | - | 1 | - | - | - | - | 1 | - | - | - |
| Malignancy | - | 2/4 | - | 4/6 | - | 3/4 | 3/4 | - | - | 1 | - |
| Intelligence decline | - | - | NA | NA | NA | NA | NA | - | - | NA | NA |
| Decreased ATM protein | NA | NA | 1 | NA | NA | 2/4 | 2/2 | 1 | NA | NA | NA |
| Elevated AFP level | 2/2 | 2/2 | 1 | NA | NA | 2/4 | 3/4 | 1 | 1 | 1/2 | 1 |
| Decreased IgA, IgG | 2/2 | 1/2 | NA | NA | NA | NA | 1/1 | NA | 1/1 | NA | normal |
| Additional features | - | Axonal neuropathy | - | - | - | Sensorimotor neuropathy | - | - | delayed motor development,  speech and swallowing difficulties | slurred speech, migraine | pneumonia, bronchitis,  pharyngitis, oligoarthritis |
| Brain MRI | Normal | NA | mild cerebellar atrophy | NA | NA | Normal | NA | normal | Normal | normal | normal |
| Family history | Yes | Yes | - | Yes | Yes | Yes | Yes | - | - | Yes | - |
| ATM Mutations | c.T8048C  (p.I2683T) and c.T8578C  (p.S2860P) | c.7271T>G  (p.V2424G)  and  c.193delC（p.Q65Rfs\*11） | c.590G>A  (p.197G>E) | c.6200 C>A (p. A2067D) | c.6200C>A (p.A2067D) | c.6200C>A (p.A2067D) | c.6679C>T  (p.R2227C) and  c.572T>A  (p.I191N) | c.8266A > T (K2756\*)  and  c.743G>T  (p.R248L) | c.8147T>C  (p.V2716A）  and c.8578\_8580delTCT（p.S2860del） | p.G301Vfs\*19  and  c.8147T>C  (p.V2716A) | c.5573G>A (p.W1858\*)  and  c.6154G>A (p.E2052K) |

Abbreviations: AAO, age at onset. y, year. NA, not assessed. AFP, α-fetoprotein.