ATAXIA-OCULOMOTOR APRAXIA TYPE 1 (AOA1)

This leaflet has been adapted from patient information pages provided by the AT Society. AOA1 and other types of AOA are not the same as Ataxia-Telangiectasia but share some symptoms.

Ataxia-Oculomotor Apraxia Type 1

Ataxia-Oculomotor Apraxia Type 1 (AOA1) is a rare type of inherited progressive cerebellar ataxia. [Orphanet. (2025). *Ataxia-oculomotor apraxia type 1*. [Online]. Available at: https://www.orpha.net/en/disease/detail/1168. Accessed 8 Dec 2025]. It is caused by a defect in a gene. AOA1 results in damage to certain parts of the brain, particularly the co-ordination centre, called the cerebellum. [Albaradie, R. et al. Ataxia with oculomotor apraxia type 1 associated with mutation in the APTX gene: A case study and literature review. *Exp Ther Med*. 2022; 24(6):709.]

What are the symptoms?

People with AOA1 have difficulty moving their eyes (side to side mainly), known as oculomotor apraxia, and may adapt by moving their head to change focus. [AT Society. (2025). *AOA1*. [Online]. Available at: https://atsociety.org.uk/about-a-t/types-of-a-t/aoa1/. Accessed 8 Dec 2025] This can interfere with reading, but often may not be noticed by the individual, but instead by other family members.

Fidgety movements of the arms and face (chorea) may appear early in AOA1, but may disappear as the condition progresses. Peripheral nerve damage (neuropathy) causing wasting of the hand and foot muscles and numbness tends to occur in the later stages of the condition. Co-ordination of limbs becomes abnormal and individuals might have trouble reaching for objects. Involuntary movements include athetosis (twisting movements), dystonia (stiff, twisted postures), myoclonus (jerks) and tremor. Fortunately, these movement disorders tend to settle with age.

Slurred speech (dysarthria) is very common and may worsen over time. The lack of co-ordination of speech can also occasionally include problems with swallowing.

AOA1 can involve some mild slowing in thought processes. Some people with AOA1 may continue in mainstream schools, whilst others may prefer the setting of a special school.

What causes AOA1?

AOA1 is caused by a defect in the APTX gene, which leads to the lack of production of a protein called Aprataxin.

How is AOA1 inherited?

AOA1 is inherited in an autosomal recessive way. This means that in order to have AOA1, a child must inherit the AOA1-causing gene from both parents. If a child has AOA1, there is a 1 in 4 chance that their siblings will have the condition. Prenatal diagnosis is possible, but is not an NHS service at present. Carrier testing may be done for at-risk family members. For more information on inheritance see Ataxia UK's *'Ataxia: what's that'?'* leaflet.

When do symptoms start?

AOA1 may develop at the toddler stage or a bit later. The child may appear clumsy and may start to walk later than their siblings. Balance gradually declines and it may be noticed that running is easier than walking i.e. the child falls less. 10-11 years after onset, a wheelchair may be needed.

How is AOA1 diagnosed?

AOA1 is diagnosed through a neurological examination, but there are some useful laboratory tests that can be carried out. The main aim of testing is to exclude a type of ataxia known as Ataxia-Telangiectasia (which also occurs in children) and to try to distinguish from another type of Ataxia-Oculomotor Apraxia, AOA2. The diagnosis can now be confirmed by a blood test looking at DNA for changes in the APTX gene. AOA1 and AOA2 are tested for with most available gene panels. Patients who test positive can be referred to the national ataxia telangiectasia centres in Cambridge and Nottingham for expert clinical management.

How common is AOA1?

There is currently little information on the prevalence of AOA1 in the UK. The estimated incidence of AOA1 is 3 people per million.

Management of AOA1

As with other cerebellar ataxias, physiotherapy and speech therapy can be helpful. An orthopaedic assessment may be considered. A visit by an occupational therapist may be useful in order to assess the need for items such as walking aids, or for adaptations to the home.

Certain symptoms can be reduced with medication. Individuals may be considered for a trial of a vitamin called co-enzyme $Q_{10,}$ but no proper studies have confirmed a positive response seen in one AOA1 family.

Due to having higher cholesterol levels, people with AOA have an increased risk of developing heart disease. Following a low-cholesterol diet is recommended for most people with AOA. [National Ataxia Foundation. (2024). *Ataxia with Oculomotor Apraxia (AOA).* [Online]. Available at: https://www.ataxia.org/wp-content/uploads/2024/08/AOA.pdf. Accessed 8 Dec 2025]

It is important to see a neurologist, who will monitor the condition, on a regular basis.

This information leaflet was written by Ataxia UK in collaboration with Dr Santosh Mordekar, Consultant Paediatric Neurologist, Sheffield Children’s Hospital.

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For more support or information please contact:

Ataxia UK, 12 Broadbent Close, Highgate, London N5 5JW Tel: +44 (0)20 7582 1444

www.ataxia.org.uk

Ataxia UK, 12 Broadbent Close, Highgate, London N6 5JW www.ataxia.org.uk
helpline@ataxia.org.uk Tel: 020 7582 1444 Helpline: 0800 995 6037