ATAXIA-OCULOMOTOR APRAXIA TYPE 2 (AOA2)

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 2

Ataxia-Oculomotor Apraxia Type 2 (AOA2) is a rare type of inherited progressive cerebellar ataxia. [Orphanet. (2025). *Spinocerebellar ataxia with axonal neuropathy type 2*. [Online]. Available at: https://tinyurl.com/skvaztv8. [Accessed 9 December 2025] It is caused by a defect in a gene. AOA2 results in damage to certain parts of the brain, particularly the co-ordination centre, called the cerebellum.

What are the symptoms?

Individuals with AOA2 experience problems with coordination of limbs, such as when reaching for objects. [AT Society. (2025). *AOA2*. [Online]. Available at: https://atsociety.org.uk/about-a-t/types-of-a-t/aoa2/. Accessed 9 Dec 2025] They can experience involuntary jerking movements (chorea), twisting movements (athetosis), stiffness and twisted posture (dystonia), jerks (myoclonus) and tremor. Over time, these movement disorders tend to settle.

Many people with AOA2 experience slurred speech (dysarthria) which can progress over time. This can also affect the swallowing mechanism.

People with AOA2 tend to have difficulty with moving their eyes (side to side mainly), known as oculomotor apraxia, and generally adapt by moving their head to change focus. This can make reading difficult, but often is not noticed by the individual but by their family.

Some slowing of thought processes can occur in AOA2. Some people will choose to stay in mainstream schools whilst others may prefer a special school.

What causes AOA2?

AOA2 is caused by a change in the SETX gene. The SETX gene contains the instructions for making a protein called senataxin, which is thought to be involved in repair of DNA damage in cells. [*DNA Repair (Amst)*. 2011; 10(2):199-209]

How is AOA2 inherited?

AOA2 is inherited in an autosomal recessive way. This means that in order to have AOA2, a child must inherit the AOA2-causing gene from both parents. If a child has AOA2, 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?

AOA2 generally starts in late adolescence or early teens. [AT Society. (2025). *AOA2*. [Online]. Available at: https://atsociety.org.uk/about-a-t/types-of-a-t/aoa2/. Accessed 9 Dec 2025] Balance declines gradually over time, and individuals may find running easier than walking, falling less. By 16-17 years following onset, individuals may require a wheelchair. As in AOA1, fidgety movements of the arms and face (known as choreoathetoid) may be seen early in the condition, and unlike AOA1 they can persist into childhood. Damage to nerves leading to wasting of the hand and foot muscles and numbness (known as neuropathy) tends to occur later in AOA2 than AOA1.

How is AOA2 diagnosed?

AOA2 is diagnosed through neurological assessments, but there are some useful laboratory tests that can be conducted. The first thing a clinician will do is distinguish AOA2 from other conditions such as AOA1 and a type of ataxia called Ataxia-Telangiectasia (AT).

AOA2 can now be diagnosed using a blood test to look at DNA. Genetic changes in a gene called Senataxin cause the disorder. 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 AOA2?

AOA2 is the most common form of AOA worldwide, with an estimated prevalence of 1 in 900,000 people. [National Ataxia Foundation. (2025). *Ataxia with Oculomotor Apraxia (AOA)*. [Online]. Available at: https://www.ataxia.org/wp-content/uploads/2024/08/AOA.pdf. Accessed 9 Dec 2025]

Management of AOA2

As with other cerebellar ataxias, physiotherapy and speech therapy can be helpful. An orthopaedic assessment may be helpful. There is no single drug to target the underlying cause of AOA2, however certain symptoms can be managed with medication.

People with AOA2 tend to have increased cholesterol levels, meaning they are at a higher risk of heart disease. [National Ataxia Foundation. (2025). *Ataxia with Oculomotor Apraxia (AOA)*. [Online]. Available at: https://www.ataxia.org/wp-content/uploads/2024/08/AOA.pdf. Accessed 9 Dec 2025] To reduce this risk, people with AOA2 are advised to follow a low-cholesterol diet.

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

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