AOA2

Clinical features of AOA2

Introduction

AOA2 tends to start in late adolescence or early teens. Balance gradually declines and it may be noticed that running is easier than walking i.e. the child falls less. By 16-17 years after onset of the condition a wheelchair may be required.

As in some patients with A-T and AOA1, choreoathetoid (fidgety) movements of arms and face may occur early on and unlike AOA1 they tend to persist into adulthood. Peripheral nerve damage (neuropathy) giving rise to wasting of the hand and foot muscles and some numbness tends to occur in the later stages of the condition.

Co-ordination of Limbs

This becomes abnormal and patients may have trouble reaching for objects. Involuntary movements include those described above in A-T. These include chorea (small jerking movements), athetosis (twisting movements), dystonia (stiff, twisted postures), myoclonus (jerks) and tremor. Fortunately, the movement disorders tend to settle with age.

Slurred Speech (Dysarthria)

This is very common and may progress with time. Most patients, however, can make themselves understood. The lack of coordination of speech can also occasionally involve the swallowing mechanism.

Eye Movements (Oculomotor Apraxia)

Patients have difficulty in moving their eyes (side to side mainly) and may adapt by moving their head to change focus. This can interfere with reading but often may not be noticed by the patient and is picked up by other family members.

Intellect

Marked mental retardation is not seen in AOA2 but there may be a mild slowing in thought processes. Some may continue in mainstream schools but others may prefer the setting of a special school.

Muscle Wasting and Numbness (Peripheral Neuropathy)

Later in the condition sensation in the feet and hands may be reduced and wasting of the foot and hand muscles can be seen. Attention to foot care and foot wear is required in a similar way to the follow up of patients with diabetic nerve damage.

Drugs

No single drug can help all people with AOA2. Certain symptoms can be reduced with medication.

Laboratory findings and diagnosis AOA2

AOA2 is a clinical diagnosis but there are some useful laboratory tests that can be carried out. The main point of testing is to exclude A-T and to try to distinguish from AOA1.

Patients with AOA2 do not have the DNA repair problems or sensitivity to irradiation seen in A-T but their alpha-fetoprotein is nearly always increased. The diagnosis can now be confirmed by a blood test looking at DNA. Genetic abnormalities (mutations) in a gene called senataxin cause the disorder. This gene test is only available via Professor Taylor's laboratory in Birmingham.

Genetics and Incidence

AOA2 is a genetic disorder which does tend to run in families. It is an autosomal recessive condition which means brothers and sisters may have it but the parents are often unaffected. If a child in a family has the condition there is a 1 in 4 chance that other offspring will be affected. Prenatal diagnosis is possible but is not an NHS service at present.

The estimated incidence of AOA2 is probably greater than A-T which is known to be 3 per million

Treatment

As with A-T and AOA1, speech therapy, physiotherapy and possibly orthopaedic assessment should also be considered in dealing with the different aspects of AOA2.

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