

Management of the Ataxias: Towards Best Clinical Practice

Summary for General Practitioners

Reviewed: September 2010



Management of the Ataxias - Summary for GPs

This is a summary of Guidelines on the diagnosis and management of people with ataxia which were developed through extensive consultation with ataxia specialist neurologists, other healthcare professionals, and the patient support organisation Ataxia UK. The list of contributors is found in the full Guidelines. For a copy of the full Guidelines go to www.ataxia.org.uk or contact Ataxia UK for a print version.

Ataxia means lack of coordination and it is a symptom of many conditions. These Guidelines focus on the progressive ataxias and exclude disorders where ataxia is an epiphenomenon of another neurological condition (such as multiple sclerosis or ataxia due to stroke or alcohol).

The progressive ataxias are uncommon conditions and therefore GPs will not often come across such patients. These Guidelines were produced to increase awareness and understanding of these conditions and to lead to their improved diagnosis and management. In the UK latest estimates suggest there are at least 10,000 adults with progressive ataxia; data for paediatric cases is expected in the next few years.

Conditions covered in the Guidelines

- Hereditary ataxias - including Friedreich's ataxia, spinocerebellar ataxias and episodic ataxias (but excluding ataxia telangiectasia)
- Idiopathic progressive ataxias - forms of cerebellar ataxia with neurodegeneration of unknown aetiology
- Specific neurological disorders where progressive ataxia is the dominant symptom eg cerebellar variant of MSA

Diagnosis

Presentation

Presenting symptoms and signs of ataxia are well known. Patients complain of unsteadiness, slurred speech, and clumsiness.

The clinical signs of cerebellar dysfunction may include the following:

- Speech may be slurred (dysarthric) and have a staccato quality
- Gait ataxia and in extreme cases impaired sitting balance
- Horizontal gaze-evoked nystagmus, hypermetropic/hypometropic saccades and saccadic interposition (jerky pursuit), which may be revealed by extra-ocular movement testing
- Intention tremor
- Dysmetria or “past-pointing”
- Dysdiadochokinesis

It is important to note that some of these signs are found in other disorders and are therefore not specific to progressive ataxias. Also, depending on the underlying cause of the ataxia there can be additional neurological features such as fatigue, muscle spasms, ophthalmoplegia, dysphagia, Parkinsonism, visual disturbance, and cognitive decline.

Referral process

Patients suspected of having ataxia should be referred to secondary care, where they should generally be seen by a neurologist or a paediatrician. Depending on the clinical situation, this referral may need to be undertaken urgently. For example, if a tumour is suspected, referral will have to be within two weeks.

When a diagnosis of progressive ataxia is made referral to a specialist neurologist, or if possible, Ataxia Centre is recommended. For details of specialised Centres see the full Guidelines or contact Ataxia UK. The GP may also refer directly to the specialist neurologist (eg in cases where a diagnosis of a progressive ataxia has already been given).

Investigations

It may be relevant to exclude multiple sclerosis, posterior fossa tumours, alcoholic cerebellar ataxia, or ataxia as a non-metastatic manifestation of malignancy. Ataxia may be caused by

medication, particularly phenytoin. There are some treatable forms of ataxia (eg Vitamin E deficiency) which should always be considered.

Investigations

Family history is crucial in diagnosing patients with progressive ataxia, in view of the frequency with which inherited factors contribute to its causation. Some ataxias are inherited recessively (the most common being Friedreich's ataxia) and others dominantly (eg spinocerebellar ataxias 1, 2, 3 etc, episodic ataxias). Mitochondrial disease may be a cause of inherited ataxia and pre-mutations of the fragile-X gene may be a cause of adult onset ataxia. Genetic tests are available for the specific diagnosis of some inherited ataxias (see full Guidelines for details of mechanisms of inheritance of the different ataxias and implications for families and the genetic tests available).

Primary Care Investigations

Adults

These can exclude common (not necessarily neurological, and possibly treatable) conditions prior to hospital referral. *May include:* U&Es, Creatinine, liver function/enzymes including γ -GT, Ca, Phos, Immunoglobins, Electrophoresis, TFT, Chol, FBC, ESR/CRP, Vitamin B12, Folate, CXR.

Children

Once ataxia has been noticed, an urgent referral to local paediatric services is necessary. The investigations will then depend on the clinical assessment, which includes details such as family history; whether ataxia is acute, episodic, or chronic; precipitants; associated conditions; and examination findings, especially those distinguishing central from sensory ataxia.

The investigation of acquired ataxia in children is generally more urgent because of the necessity of excluding posterior fossa and brainstem tumours and because of the likely chance that the cause will be genetic and the parents may wish further children. For these reasons, identification of recent-onset ataxic signs should lead to urgent referral to local paediatric services.

Other investigations

For details of secondary care investigations and full details of the different types of ataxias identified to date see the full Guidelines. As many progressive ataxias are inherited, genetic tests form an important part of secondary care investigations. Genetic assessment and advice should also be part of this process to explain the implications of the diagnosis to family members. Patients should be advised that (almost inevitably in cases presenting for the first time) they are likely to undergo brain imaging - MRI in the case of adults or CT/MRI in children.

Patient pathways

Following the referral to a neurologist, in many cases it may be relevant for either the GP or neurologist to refer patients to other specialists.

Community paediatric multidisciplinary team

Children should be referred to the Community Paediatric Multidisciplinary team.

Cardiologist

Cardiac abnormalities are common in Friedreich's ataxia, therefore a referral to a cardiologist is required.

Neuro-ophthalmologist and other eye specialist services

Many of the ataxias are associated with eye symptoms such as reduced vision, diplopia or oscillopsia due to nystagmus. A referral to a neuro-ophthalmologist and other specialist services is recommended since in some cases treatment may be available.

Neuropsychologist

Some ataxias may be associated with cognitive problems, therefore in selected cases a referral to cognitive testing may be relevant.

Spinal surgeon / orthopaedic surgeon / orthotist

Patients with Friedreich's ataxia often develop scoliosis. Referral

to spinal surgery and/or orthopaedic surgery may therefore be appropriate in some cases; referral to physiotherapy may well be helpful. Patients with Friedreich's ataxia may develop *pes cavus*, therefore referral to an orthopaedic surgeon with specialty in foot and ankle surgery and to an orthotist may be appropriate.

Neuro-urologist

Bladder problems can be a feature of some of the ataxias. They occur, for example, in multiple system atrophy. Also, in later stages of various spinocerebellar ataxias urinary incontinence is also sometimes experienced.

Neuro-rehabilitation

Patients would benefit from a referral for neurorehabilitation at the early stage of the disease in order to establish strategies to maintain function, eg balance, upper limb coordination, speech and swallowing (see in depth therapy sections in the full Guidelines).

Physiotherapy is often valuable, particularly to preserve mobility, and to avoid other problems, such as ones associated with being in a wheelchair. Regular follow-up is important. Patients will also need advice on walking aids at the different stages of their condition. Referral to a wheelchair clinic for specialist seating advice is important at the appropriate stage of the disease; early referral is advised where possible.

Patients with progressive ataxia often experience dysarthria, which later in the disease may cause communication difficulties. A referral to a ***speech and language therapist*** is therefore important. Dysphagia becomes more common as the disease progresses, therefore this should also be assessed by a speech and language therapist or other appropriately trained professional.

Ataxia patients benefit from regular assessments by an ***occupational therapist***. Occupational therapists have expertise in assessment of daily tasks and providing specific interventions which may include teaching strategies, recommending equipment or adaptations to the home environment.

Counselling

Anecdotal evidence has shown counselling to be beneficial in helping patients diagnosed with a progressive ataxia come to terms with their condition. The impact of such diagnoses may be devastating and counselling may be valuable.

Patient Support groups

Referral to a patient support organisation is recommended for ongoing support and information.

- Ataxia UK provides support to people with all ataxias (registered charity number 1102391) **www.ataxia.org.uk** Helpline 0845 6440606
- Sarah Matheson Trust for multiple systems atrophy (registered charity number 1062308) **www.msaweb.co.uk** Tel: 020 7886 1520
- Ataxia Telangiectasia Society for people with ataxia-telangiectasia (registered charity number 1105528) **www.atsociety.org.uk** Tel: 01582 760733

Reviews and ongoing management

Patients should be offered annual (or 6-monthly) follow-up from a neurologist. This is important for monitoring progression of the condition, identifying new symptoms and providing information on new treatments or diagnostic tests. Regular follow-up with Friedreich's ataxia is necessary specifically to monitor for the development of cardiomyopathy, diabetes, scoliosis and other treatable symptoms. Symptoms of depression should also be identified and treated appropriately.

For the majority of patients, for most of the time, their ongoing management can be provided at the primary care level. In addition to regular input from GPs, other professionals such as community therapists are likely to be involved. For information on the management and treatment of symptoms and treatable forms of ataxia see the full Guidelines.

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