Management of the ataxias: towards best clinical practice

• Ataxia UK •

Conditions covered in these guidelines

- 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
- Hereditary ataxias—including Friedreich's ataxia, spinocerebellar ataxias and episodic ataxias (but excluding ataxia-telangiectasia)
- Idiopathic progressive ataxias—forms of cerebellar ataxia associated with neurodegeneration of unknown aetiology
- Specific neurological disorders in which progressive ataxia is the dominant symptom, e.g. cerebellar variant of multiple system atrophy
- Estimates suggest that there are at least 10,000 adults and 500 children with progressive ataxias in the UK

Presentation

- Patients with ataxia complain of incoordination, resulting in unsteadiness and clumsiness, and slurred speech. Rarely, oscillopsia (due to nystagmus) is reported. The clinical signs seen in patients with ataxia can be summarised as follows:
  - gait ataxia and, in some cases, impaired sitting balance
  - gaze-evoked and/or resting nystagmus, hypermetropic/hypometropic saccades and jerky pursuit
  - speech may be slurred (dysarthric) and have a staccato quality
  - intention tremor
  - dysmetria or 'past-pointing'
  - dysdiadochokinesis
- Depending on the underlying cause of the ataxia, there can be additional neurological features that manifest themselves during the course of the illness. These include cognitive changes and decline, parkinsonism, visual disturbances, ophthalmoplegia, spasticity, peripheral neuropathy, and urinary dysfunction
- Early diagnosis of the underlying cause of the ataxia is important, especially as some causes of ataxia are treatable, e.g. ataxia with vitamin E deficiency

Referral process

- Patients presenting with ataxia symptoms should be referred to secondary care, following primary care investigations if appropriate (see below)
- If the patient is under 16 years old they should be referred to a paediatrician. The recommended pathway then involves being referred to a paediatric neurologist
- If the patient is over 16 years old they should generally be referred to an adult neurologist. Depending on the clinical state of the patient, the referral may need to be urgent, for example in the case of a suspected brain tumour
- Referral to a neurology centre specialising in ataxia is recommended, particularly for patients where a diagnosis of the cause of ataxia has not been achieved
- There are three specialist ataxia centres in the UK accredited by Ataxia UK. Referrals can be made by either primary or secondary care. Please contact Ataxia UK for more information on neurology centres specialising in ataxia management and the three specialist ataxia centres in the UK accredited by Ataxia UK

Investigations

Adults

- Primary care investigations may include:
  - vitamin B12 and folate
  - urea and electrolytes
  - creatinine
  - full blood count
  - liver enzymes
  - gamma-glutamyl transferase
  - thyroid function tests
  - random blood sugar
  - chest x-ray
  - erythrocyte sedimentation rate/C-reactive protein

- The purpose of these tests are to exclude other medical and neurological conditions that may contribute to ataxia

Children

- The investigation of ataxia in children is generally more urgent because of the necessity of excluding posterior fossa and brainstem tumours

- It is also important to consider that there is a need for investigation in children as there is a risk that the cause may be genetic, and their parents may wish to have further children. Referral to a genetic counsellor at this stage is important

- For these reasons, identification of recent-onset ataxic signs should lead to urgent referral to local paediatric services

- For details of secondary care investigations and full details of the different types of ataxias identified to date, please refer to the full guideline

Patient pathways

Referrals

- Following the referral to a neurologist, in many cases it may be relevant for either the GP or the neurologist to refer patients to other specialists. Depending on the symptoms experienced, and the type of ataxia a patient is diagnosed with, a variety of different specialists will be involved in their care

- The following specialist and services may be involved:
  - genetic counseling services
  - community paediatric multidisciplinary team
  - spinal surgeon/orthopaedic surgeon
  - orthotist
  - cardiologist
  - urologist
  - gastroenterologist
  - audiology services
  - neuro-ophthalmologist
  - neuropsychologist/neuropsychiatrist
  - physiotherapy
  - speech and language therapy
  - occupational therapy
  - specialist palliative care team

- GPs are advised to signpost patients to patient support groups such as Ataxia UK

Reviews and follow-up

- Patients should be offered 6–12 monthly reviews from a neurologist or a specialist ataxia neurologist. If it is difficult for patients to travel to the hospital, and their condition is stable, follow-up appointments could be less frequent

- Regular follow-up reviews are important for a number of reasons. Firstly, it enables the neurologist to monitor the progression of the condition and identify any new symptoms that may need treatment. Secondly, patients can benefit from medical advances, such as new diagnostic tests and new treatments

- Regular follow-up of patients with Friedreich's ataxia is necessary, specifically to monitor for the development of cardiomyopathy, diabetes, scoliosis and other treatable symptoms. Annual urine/blood tests for diabetes are also recommended

- For the majority of patients with ataxia, for most of the time, their ongoing management can be provided at the primary care level. In addition to regular input from their GPs, other professionals including community therapists are likely to be involved. Specific community nursing needs may be delivered by district nurses

- The hospital-based neurologist/ataxia specialist will, however, remain involved as a coordinator and instigator of services. Effective communication between primary and secondary care is therefore vital

Common symptoms of ataxia

- Patients with progressive ataxias may experience a variety of symptoms, some of which can be treated medically, these may include:
  - muscle spasticity, spasms and joints contractures
  - tremor
  - dystonia
  - scoliosis
  - pain
  - cardiac problems in Friedreich's ataxia
  - lower urinary tract dysfunction
  - gastroenterological problems (e.g. constipation, faecal urgency, and incontinence)
  - erectile dysfunction
  - swallowing and dysphagia
  - nutritional considerations/deficiencies (e.g. in gluten ataxia, ataxia with vitamin E deficiency)
  - sialorrhea
  - hearing loss
  - eye symptoms (e.g. nystagmus, diplopia, visual impairment)
  - cognition
  - depression and other psychiatric symptoms

- For details of these symptoms and how each may be treated, please refer to the full guideline

Palliative care

- Most palliative care for patients with progressive ataxias, including end of life care, is provided in the community by district nurses, GPs and other members of the primary care team

- For patients with more complex needs help may be required from palliative care specialists

- The progressive ataxias often have a disease course spanning several decades. It can be hard to know when the end of life is approaching, but 'triggers' can be used to identify individuals approaching the end of their lives. One such is the intuitive 'surprise question': 'Would you be surprised if this patient died in the next 12 months?'. There are additional general and specific (for ataxia) prognostic indicators for patients with ataxia (see full guidelines document for more information on indicators specific to ataxias). Answering 'no' to the surprise question and/or the presence of general/specific clinical indicators of decline should act as an alert that the individual with ataxia is approaching the end of life, and the need possibly to initiate multi-professional end of life care.'

Research

- There are many research projects ongoing on the ataxias including clinical trials. For information about these and to find out how patients can participate if they wish, please visit the Ataxia UK website

full guideline available from...
www.ataxia.org.uk
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