Management of the Ataxias: towards best Clinical Practice

November 2009
Development Group:

Dr Rajith de Silva, Neurologist, Queen’s Hospital, Romford
Dr Paola Giunti, Neurologist, National Hospital for Neurology and Neurosurgery, London
Dr Julie Greenfield, Research Projects Manager, Ataxia UK
Professor Barry Hunt, Chair of Scientific Advisory Committee, Trustee of Ataxia UK and parent of a daughter with ataxia (Chair of the Guideline Development Group)

Other major contributors:

Dr Nick Fletcher, Neurologist, The Walton Centre for Neurology and Neurosurgery, Liverpool
Dr Marios Hadjivassiliou, Neurologist, Sheffield Teaching Hospitals NHS Foundation Trust
Dr Elizabeth Harrison, retired General Practitioner, Chair of Ataxia UK and parent of a daughter with ataxia
Dr Andrea Nemeth, Clinical Geneticist, Churchill Hospital, Oxford
Professor Patrick Morrison, Neurogeneticist, Belfast City Hospital, Belfast
Dr Neil Robertson, Neurologist, University Hospital Wales, Cardiff
Professor Tony Schapira, Neurologist, Royal Free Hospital, London
Dr Alison Stevenson, Research Officer, Ataxia UK
Dr Kevin Talbot, Neurologist, Radcliffe Infirmary, Oxford

Specialist section contributors:

Dr Peter Baxter, Paediatric Neurologist, Sheffield Children’s NHS Foundation Trust, Sheffield
Dr Claire Bates, Consultant in Palliative Medicine, Queen’s Hospital, Romford
Elizabeth Cassidy, Lecturer in Physiotherapy, Centre for Research in Rehabilitation, Brunel University, West London
Dr Anna Farrell, Speech and Language Therapist, National Hospital for Neurology and Neurosurgery, London
Ann Holland, Consultant Physiotherapist, National Hospital for Neurology and Neurosurgery, London
Susan Hourihan, Occupational Therapist, National Hospital for Neurology and Neurosurgery, London
Joanne Hurford, Occupational Therapist, National Hospital for Neurology and Neurosurgery, London
Dr Cherry Kilbride, Lecturer in Physiotherapy, Centre for Research in Rehabilitation, Brunel University, West London
Melissa Loucas, Speech and Language Therapist, National Hospital for Neurology and Neurosurgery, London
Dr Anja Lowit, Speech and Language Therapist, Strathclyde University, Glasgow
Dr Antonis Pantazis, Consultant Cardiologist, The Heart Hospital, University College London, London
Lucy Rodriguez, Speech and Language Therapist, National Hospital for Neurology and Neurosurgery, London
Pip Wilford, Occupational Therapist, National Hospital for Neurology and Neurosurgery, London

Expert advice also given by:

College of Occupational Therapy, Specialist Section for Neurological Practice, long-term conditions forum
Mr Fion Bremner, Consultant Neuro-Ophthalmologist, National Hospital for Neurology and Neurosurgery, London
Professor Patrick Chinnery, Neurologist, University of Newcastle upon Tyne, Newcastle upon Tyne
Dr Jennifer Davis, Associate Specialist, Department of Neurology, Queen’s Hospital, Romford
Professor Clare Fowler, Uro-Neurologist, Department of Uro-Neurology, National Hospital for Neurology and Neurosurgery, London
Dr Mark Harrison, Retired Consultant Urologist
Professor Maria Ron, Neuropsychiatrist, Institute of Neurology, University College London, London
Dr Martin Watson, Physiotherapist, University of East Anglia, Norwich

Professor Robert Surtees has made a major contribution to these Guidelines but sadly passed away in 2007. He will be remembered for his vast knowledge and expertise in childhood ataxias and for his kindness. We would like to dedicate this publication to his memory.

Review date: Summer 2011

Disclaimer

Please note that this information is published for information purposes only. No person shall have any claim of any nature whatsoever arising out of or in connection with this publication against the authors, Ataxia UK or any of its officers and employees.

Feedback

We would like to update these guidelines regularly, and would be keen to incorporate readers’ ideas and experiences, especially examples of good practice. Please contact Ataxia UK (email: research@ataxia.org.uk).
Contents

1 Introduction 4

2 Diagnosis 6
  2.1 Presentation 6
  2.2 Referral process 7
  2.3 Investigations 8
  2.4 Genetics 10

3 Patient Pathway 14
  3.1 Referrals 14
  3.2 Reviews and follow-up 16

4 Medical Interventions 18
  4.1 Symptomatic treatments 18
    Cardiac problems 18
    Bladder problems 19
    Sexual dysfunction 20
    Muscle spasms and spasticity 20
    Contractures 20
    Tremors 20
    Dystonia 20
    Depression and psychiatric symptoms 21
    Fatigue 21
    Eye symptoms 21
    Pain 22
    Episodic ataxias 22
  4.2 Disease modifying treatments 22
    Friedreich's ataxia 23
    Gluten ataxia 23
    Treatable progressive ataxias 24
    Treatable causes of childhood ataxias 25

5 Therapies 27
  5.1 Speech and language therapy 27
  5.2 Physiotherapy 32
  5.3 Occupational therapy 38

6 Research 48

7 Palliative Care 49

Appendix 50

References 51
1 Introduction

This document aims to provide recommendations for healthcare professionals on the diagnosis and management of people with ataxia. Ataxia means ‘lack of coordination’ and it is a symptom of many conditions. These guidelines focus on the progressive ataxies, and exclude disorders where ataxia is an epiphenomenon of another neurological condition (see Table 1). Certain aspects of these Guidelines may however be relevant to these other conditions.

They have been developed through extensive consultation with ataxia specialist neurologists and other healthcare professionals in collaboration with the patient support organisation, Ataxia UK. The first edition of these Guidelines was published in March 2007; this is an updated and expanded version.

Table 1: Conditions covered in these Guidelines and other causes of ataxia

<table>
<thead>
<tr>
<th>Conditions covered in these Guidelines</th>
</tr>
</thead>
<tbody>
<tr>
<td>• <em>Hereditary ataxias</em> – including Friedreich’s ataxia, spinocerebellar ataxias and episodic ataxias (but excluding ataxia-telangiectasia*)</td>
</tr>
<tr>
<td>• <em>Idiopathic progressive ataxias</em> – forms of cerebellar ataxia associated with neurodegeneration of unknown etiology</td>
</tr>
<tr>
<td>• <em>Specific neurological disorders in which progressive ataxia is the dominant symptom eg cerebellar variant of MSA</em></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Other causes of ataxia</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Vascular</td>
</tr>
<tr>
<td>• Traumatic</td>
</tr>
<tr>
<td>• Developmental</td>
</tr>
<tr>
<td>• Neoplastic / paraneoplastic</td>
</tr>
<tr>
<td>• Infectious</td>
</tr>
<tr>
<td>• Inflammatory (eg multiple sclerosis)</td>
</tr>
<tr>
<td>• Metabolic</td>
</tr>
<tr>
<td>• Toxic / drug-related (eg alcohol)</td>
</tr>
<tr>
<td>• Epilepsy (in children)</td>
</tr>
</tbody>
</table>

* Information about the extra neurological features of ataxia-telangiectasia is not included in this document. For more in depth knowledge about this condition please refer to the Ataxia-Telangiectasia Society.

The progressive ataxies are generally thought to be rare neurological conditions, and are poorly understood by healthcare professionals. However, recent evidence suggests that the ataxies are more common than previously thought and may be under-diagnosed (see Box 1). This highlights the importance of producing these guidelines: in order to increase awareness and understanding of these conditions, and lead to their improved diagnosis and management.
**Box 1 Epidemiology of the ataxias**

Epidemiological studies of the progressive ataxias in the UK are sparse. Results of further epidemiological studies in the UK are expected in the next few years.

**Recent UK studies:**


- Estimated minimum prevalence: 10.2 in 100,000 people with late onset cerebellar ataxia in South Wales.

**European studies:**

- The most common inherited ataxia in the UK is Friedreich’s ataxia, which is a recessively inherited condition that tends to be of early onset.
  
  Estimated prevalence of Friedreich’s ataxia in studies before the availability of genetic tests:
  
  1 in 50,000. Estimated disease incidence based on carrier frequency of 1 in 85 is 1 in 29,000

These studies suggest that the prevalence of the progressive ataxias is higher than conditions that are generally better known such as Huntington's disease and motor neurone disease.

In the UK, the latest estimates suggest there are at least 10,000 adults with progressive ataxia; data for paediatric cases is expected in the next few years.
2 Diagnosis

2.1 Presentation

The presentation of a patient with ataxia can be considered in many domains. The entity may be transient (eg following a viral infection in a child), episodic (eg in a patient with multiple sclerosis) or progressive (eg in Friedreich’s ataxia, an inherited neurodegenerative disorder). Onset may be acute (eg in a patient with stroke) or slow (eg Vitamin or thyroid deficiencies). Finally the age of onset should be considered; the types of disorders presenting with ataxia in children or young adults (frequently developmental, metabolic or inherited causes) tend to differ from those presenting in older people (vascular, neoplastic or neurodegenerative). The clinician therefore has to synthesise many aspects of the history in coming up with a differential diagnosis for an individual patient.

The family history is crucial in patients with ataxia, in view of the frequency with which genetic/inherited factors contribute to its causation. Almost all forms of genetic transmission are recognised, but generally speaking young-onset ataxias tend to be of autosomal recessive (AR) inheritance (eg Friedreich’s ataxia) whereas the autosomal dominant (AD) ataxias tend to present in young adults and in early middle life. With AR inheritance there is a 1 in 4 risk of further siblings also being affected, but the parents of the patient, whilst carriers of the mutated gene, are themselves clinically unaffected. Parental consanguinity is sometimes identified. With AD transmission, one of the parents is likely to have similar clinical characteristics, but, especially if carrying an unstable triplet repeat containing gene, may have much milder clinical features and also may themselves have presented at a later stage in their lives. In this category, sometimes paternal transmissions particularly tend to lead to dramatically reduced ages of onset and more severe clinical phenotypes in offspring. Mitochondrial disease may be an under-diagnosed cause of ‘inherited’ ataxia, but here the mechanisms of transmission may be complex, including maternal transmission, AR and AD inheritance. Premutations of the fragile-X gene may be a cause of adult-onset ataxia (‘Tremor-ataxia syndrome’) that affects men and women.

Presenting symptoms and signs of ataxia are well known. Patients complain of incoordination and unsteadiness, slurred speech and clumsiness. Rarely oscillopsia (due to nystagmus) is reported. The clinical signs of cerebellar dysfunction can be summarised as follows:

- **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**
- **Speech may be slurred (dysarthric) and have a staccato quality**
- **Intention tremor**
- **Dysmetria or ‘past-pointing’**
- **Dysdiadochokinesis**
It is important to note that some of these signs are found in other disorders too and are therefore not all specific to progressive ataxias.

Midline cerebellar disease may only be detected by testing walking, especially heel-to-toe or tandem gait. It is important to recognise that impairments in motor function and sensation (especially joint position sense) can mimic cerebellar ataxia. Romberg’s sign may detect impaired joint position sense, but in some forms of (spino)cerebellar ataxia, the posterior column sensory modalities are also impaired.

Depending on the underlying cause of the ataxia there can be additional neurological features that manifest themselves during the course of the illness. These can include fatigue, ophthalmoplegia, dysphagia, parkinsonism, visual disturbance and cognitive decline.

**Ataxia may indicate neurological disease and should be referred without delay to secondary care**

### 2.2 Referral process

Patients suspected of having ataxia should be referred for secondary care, where they should generally be seen by a paediatrician or neurologist. Depending on the clinical situation, this referral may need to be undertaken urgently. For example, in a case with a suspected tumour, this will have to be within 2 weeks. In children, after referral to a paediatrician the recommended pathway then involves being referred to a paediatric neurologist.

A referral to a centre specialising in ataxia is then recommended. In selected cases in adults it may be relevant for the general practitioner (GP) to refer directly to the specialist neurologist (eg in cases where a diagnosis of a progressive ataxia has already been given).

Ataxia UK aims to accredit an increasing number of neurological centres across the country as recognised centres for the diagnosis and management of patients with ataxia, and ideally the transfer of patients within secondary care to the nearest recognised centre should take place speedily, when alternative diagnoses such as multiple sclerosis have been excluded. A current list of Accredited Ataxia Centres and other recognised Ataxia Centres in the UK is found in the Appendix.

**When a diagnosis of progressive degenerative ataxia is made referral to a specialist ataxia centre is recommended**
2.3 Investigations

Adults
Details of appropriate investigations for adults, grouped in order of priority are found in Table 2. Also see section 2.4 for genetic tests.

Clinical judgement is paramount in the application of tests. In certain instances the diagnosis may be so likely at the very first clinical encounter that a ‘third line’ genetic test, for example, may be undertaken immediately. Youngish patients with musculoskeletal abnormalities, slowly progressive spinocerebellar ataxia with little or no nystagmus and absent reflexes should have the Friedreich’s ataxia (FRDA) gene expansions excluded early in 2nd line investigations (along with their Vitamin E level).

Table 2: Diagnostic investigations in adults

<table>
<thead>
<tr>
<th>Primary care</th>
<th>Relatively inexpensive, common investigations, not specific for neurology, widely available; to exclude common (not necessarily neurological) conditions, which are sometimes easily treated. Could be ordered by GP prior to hospital referral.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Details of tests:</td>
<td>U &amp; Es, Creatinine, Liver enzymes, γ-GT, Ca, Phos, Igs, Electrophoresis, TFT, Chol, FBC, ESR/CRP, Vitamin B12, Folate, CXR</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Secondary care 1st line</th>
<th>MRI of the brain</th>
</tr>
</thead>
<tbody>
<tr>
<td>Rarer tests selectively ordered by neurologists; some conditions though rare are treatable (eg Wilson’s); includes opinions of specialists eg ophthalmologists; basic genetic ‘screen’ especially if suspicious features eg family history.</td>
<td></td>
</tr>
<tr>
<td>Details of tests:</td>
<td>Lipid-adjusted Vitamin E and lipoproteins, α-FP, Blood film, Lactate, Copper, Caeruloplasmin, 24hr-urinary copper, MRI of cervical spine, Lumbar puncture (cells, protein, glucose*, cytology, oligoclonal bands*, Lactate, Ferritin), Anti-gliadin abs (IgG and IgA), a Syphilis</td>
</tr>
<tr>
<td>*with blood</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>2nd line</th>
<th>Very rare and/or expensive investigations with low yield.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Details of tests:</td>
<td>Phytanic acid, Peripheral nerve conduction studies, Electromyography, Electroencephalography, Neuropsychology, Ophthalmology, Anti-GAD, Anti-VGCC, Anti-Hu/Yo and other paraneoplastic antibodies, CT of chest, abdomen and pelvis, Total body PET, Muscle biopsy, White cell enzymes, Long chain fatty acids, 14-3-3 protein in CSF (prion conditions), b Cholestanol, Coenzyme Q10 (ubiquinone), c Anti-thyroid antibodies</td>
</tr>
<tr>
<td>Genetic tests (see section 2.4)</td>
<td>Remaining genetic tests.</td>
</tr>
</tbody>
</table>
All the diagnostic tests in table 2 should be readily available in primary or secondary care. Tests marked a-c are only available in certain laboratories:

**a** If testing for antigliadin antibodies is not readily available clinicians can contact Dr Marios Hadjivassiliou who runs a specialised gluten ataxia clinic. Contact details: Sheffield Teaching Hospitals NHS Foundation Trust, Sheffield S10 2JF. Tel: 0114 271 2502.

**b** Testing for 14-3-3 protein in the CSF can be carried out at the National CJD Surveillance Unit, Western General Hospital, Edinburgh. Contact Alison Green to discuss individual cases before sending samples for testing. Tel: 0131 537 3075 email: alison.green@ed.ac.uk

**c** Coenzyme Q10 (CoQ10) levels to test for ataxia with CoQ10 deficiency. Testing of CoQ10 deficiency can be performed on skeletal muscle samples or on blood mononuclear cells. Testing is available at the Neurometabolic Unit, National Hospital for Neurology and Neurosurgery, London. Contact Dr Iain Hargreaves or Dr Simon Heales: iain.hargreaves@uclh.nhs.uk or simon.heales@uclh.nhs.uk Tel: 0845 155 5000 ext 723844

**Children**

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

**Table 3: Diagnostic investigations in children**

| 1st line | Neuroimaging is mandatory. A CT or MRI brain scan will exclude a tumour and may indicate a white matter disorder. If the CT is normal and the ataxia does not seem acute, local paediatric services should investigate in the light of clinical judgment and in consultation with the local tertiary paediatric neurology service. |
| 2nd line | Investigations likely to be available to local paediatric teams could include: full blood count with vacuolated lymphocytes; plasma lactate; urate; ammonia; very long chain fatty acids; amino acids; lipid-adjusted Vitamin E and Vitamin B12; biotinidase; thyroid function; serum α-fetoprotein; immunoglobulins; thyroid and antigliadin antibodies; viral serology; karyotype; DNA to keep; urinary vanillylmandelic acid; uric acid; amino-acids and organic acids; urine toxicology; ECG. Urgent referral to tertiary paediatric neurology services is almost always necessary to complete investigations and for advice about management. |
Other investigations could include:
MRI of the brain (if not already done) and spinal cord; electromyography and nerve conduction studies; somatosensory evoked potentials; echocardiography; plasma and urine bile acids; whole blood acylcarnitine analysis; transferrin isoelectric focusing; red cell purine nucleotide species; electron microscopy of lymphocytes; white cell lysosomal enzymes and ubiquinone; lactate, glucose and amino acids in cerebrospinal fluid with simultaneous plasma measurement; genetic investigations such as FISH 22q and Angelman deletions; molecular tests for AT, AOAs and Nijmegan breakage syndrome; chromosomal radiation fragility; DNA for FRDA, common mitochondrial DNA mutations, SCAs, DRPLA and (importantly) to store. It might also be necessary to do more invasive investigations including skin, rectal, bone marrow and muscle biopsies.

For further information on the availability of diagnostic tests see Adults section above and section 2.4.

2.4 Genetics

Referral to Genetics Services
When ordering genetic studies, it is important to consider if referral to the Regional Genetics Service is appropriate. Local practices will vary, but in view of the potential implications for family members of subjects who undergo testing, it is vital that good collaborations exist between neurologists who carry out tests and their Clinical Genetics counterparts (as highlighted in the National Service Framework for long-term conditions; quality requirement 2).^7^ The situation is simplest in the case of symptomatic subjects when the test is being performed primarily for diagnostic purposes. However, it is important that the patient and their family are informed about the potential implications in case the test is positive. This level of information giving can be provided by neurologists, especially those with expertise in this field. Nevertheless, the availability of Clinical Genetics services should be indicated to the patient and their family. The situation is more complicated in the case of ‘at-risk’ subjects, where an individual is at that point clinically unaffected. In addition there may also be approaches for pre-natal testing.

In all of these situations, the individuals should be referred urgently to collaborating Clinical Geneticists, with all of the available data (including the genetic diagnosis of the index case if available and the patient agrees to the release of this information). Good communication between the different specialties and professionals is vital.

When samples for genetic tests are being obtained from patients and their families, informed consent for research studies should also be sought. Routine consent forms can be obtained from Genetic Centres. Almost invariably the Ethics Committee submission in connection with the study will have clarified the appropriate consent to be obtained. Usually participants are given the option of being informed about any results that may emanate from studies, especially were this to be of relevance to them and their family members.
**Genetic tests available**

Table 4 shows the currently available genetic tests, grouped according to modes of inheritance. The ones underlined are available via the UK Genetics Testing Network, and their website ([www.ukgtn.org](http://www.ukgtn.org)) gives details of accredited laboratories across the UK where the tests are undertaken along with turnaround times. The others (ie non-underlined) refer to cloned genes published in the international medical literature, but which are not available routinely as diagnostic tests. For tests other than those routinely available referral to a Clinical Genetics Service is recommended. This document plans to make available the details of laboratories where such tests may be undertaken with regular updates (see below). For a full list of inherited ataxias see references 8-10.

Clinicians who take samples for genetic tests should be made aware of Guidelines on best practice for the ataxias produced by the European Molecular Genetics Quality Network ([see www.emqn.org](http://www.emqn.org)).

**Table 4: Genetic tests (underlined tests available ‘routinely’ via the UK Genetic Testing Network)**

| Genetic tests available | FRDA | AT<sup>a</sup> | AOA1<sup>b</sup> and AOA2<sup>b</sup> | Vitamin E deficiency | POLG 1 | Type I | Type II | Type III | EA type 1<sup>c</sup> | EA type 2 | DRPLA | GSS<sup>d</sup> | POLG 1 | SCA1, SCA2, SCA3 | SCA12, SCA13, SCA17, SCA28 | SCA7 | SCA5, SCA6, SCA10, SCA11, SCA14, SCA15 |
|-------------------------|------|-----------------|--------------------------------------|----------------------|--------|--------|--------|---------|------------------|----------|--------|--------|--------|--------|-----------------|----------------|-------|------------------|
| **Autosomal recessive**  |      |                 |                                      |                      |        |        |        |         |                  |          |        |        |        |        |                 |                |       |                  |
| **Autosomal dominant**   |      |                 |                                      |                      |        |        |        |         |                  |          |        |        |        |        |                 |                |       |                  |
| **Mitochondrial**        |      |                 |                                      |                      |        |        |        |         |                  |          |        |        |        |        |                 |                |       |                  |
| **‘X-linked’**           |      |                 |                                      |                      |        |        |        |         |                  |          |        |        |        |        |                 |                |       |                  |

Genetic tests marked a-e are also available; contact details for arranging these tests are given.

**a** Testing for AT – Nottingham National Ataxia-Telangiectasia clinic. This service has been nationally commissioned by the National Commissioning Group. Referral to this Centre can either be via the Ataxia-Telangiectasia Society (contact Mrs Kay Atkins Tel: 01582 760733 or email atsociety@btconnect.com) or directly to the clinic (contact Dr Mohnish Suri, Director A-T Clinic Service, Dept of Clinical Genetics City Hospital Nottingham Tel: 0115 962 7728).
Testing for AOA1 and AOA2 – Professor Malcolm Taylor CR-UK Institute for Cancer Studies, University of Birmingham, A.M.R.Taylor@bham.ac.uk or Dr Andrea Nemeth, Department of Clinical Genetics, Churchill Hospital, Oxford, OX3 7LJ. andrea.nemeth@imm.ox.ac.uk

Testing for episodic ataxia type 1 - Dr Marios Hadjivassiliou, Sheffield Teaching Hospitals NHS Foundation Trust, Sheffield S10 2JF. Tel: 0114 271 2502 or Dr Mary Davis, National Hospital for Neurology and Neurosurgery, London WC1N 3BG Tel: 0207 837 3611 ext 4250.

Testing for GSS syndrome (Gerstmann-Straussler-Scheinker syndrome) and other prion-related genetic disorders can be carried out at the National Prion Clinic, London (www.nationalprionclinic.org/). It is also provided by the National CJD Surveillance Unit, Edinburgh (www.cjd.ed.ac.uk).

Rare mitochondrial disease service. This service has been nationally commissioned by the National Commissioning Group (www.ncg.nhs.uk). There are three designated sites: London - National Hospital for Neurology and Neurosurgery, Newcastle upon Tyne - Royal Victoria Infirmary and Oxford - John Radcliffe Hospital and Churchill Hospitals.

For further details of other laboratories in European countries providing diagnostic tests go to www.orpha.net (ORPHA97). Tests are available for SCA10, SCA14 and SCA28.

**Guidance for genetic testing**

With the numerous AD spinocerebellar ataxias that have been identified, the Harding classification of considering these as types I, II or III is of some utility, and may inform genetic testing. Type I is so-called ‘complicated’ disease, where in addition to the ataxia, other neurological findings such as dementia, ophthalmoplegia, pyramidal signs and extrapyramidal features may be present. In type II disease there is progressive retinopathy and resulting blindness, and most cases to date have been associated with SCA7. Type III disease is reasonably ‘pure’ spinocerebellar ataxia. In Table 5, the currently available AD ataxia genes are tabulated, and the corresponding Harding group and any especially distinguishing clinical characteristics are indicated.

**Table 5: The autosomal dominant spinocerebellar ataxias (SCAs) – Harding classification and distinguishing clinical features**

<table>
<thead>
<tr>
<th>SCA</th>
<th>Harding type</th>
<th>Clinical/other features</th>
</tr>
</thead>
<tbody>
<tr>
<td>SCA1</td>
<td>I</td>
<td>Pyramidal involvement, ophthalmoplegia</td>
</tr>
<tr>
<td>SCA2</td>
<td>I</td>
<td>Slow saccades, peripheral neuropathy</td>
</tr>
<tr>
<td>SCA3</td>
<td>I</td>
<td>Also known as Machado-Joseph. Pyramidal involvement, ophthalmoplegia, peripheral neuropathy, in a subgroup Parkinsonian phenotype</td>
</tr>
<tr>
<td>SCA6</td>
<td>III</td>
<td>Allelic with EA2 / Familial Hemiplegic Migraine, mild ataxic syndrome</td>
</tr>
<tr>
<td>SCA7</td>
<td>II</td>
<td>Macular degeneration</td>
</tr>
<tr>
<td>SCA8</td>
<td>III</td>
<td>Not specific test*</td>
</tr>
<tr>
<td>SCA10</td>
<td>III</td>
<td>Seizures, Mexican origin</td>
</tr>
<tr>
<td>SCA11</td>
<td>III</td>
<td></td>
</tr>
</tbody>
</table>
There is limited information on the prevalence of inherited ataxias in the UK, however some are believed to be rarer than others and the information below may provide some guidance on diagnostic testing.

- **Friedreich’s ataxia** – the most common inherited ataxia in Caucasian populations.\(^\text{11}\)

- **Spinocerebellar ataxias** – prevalence has regional variability but in general it is thought that SCAs 1, 2, 3 and 6 are the most common worldwide. The most common SCA in the UK is SCA6.\(^\text{12}\) There is high prevalence of SCA3 in Portugal and Brazil, SCA7 in South Africa and SCA2 in Cuba.\(^\text{13, 14}\)

- **Dentatorubral Pallidoluysian Atrophy (DRPLA)** – was thought to be rare in Caucasian populations and most commonly reported in Japan. However a recent study in Wales suggests that DRPLA may not be as geographically restricted as thought and the diagnosis should be considered in UK patients.\(^\text{15}\)

- **SCA13 and SCA14** – in a recent UK study no SCA13 families were found, whereas at least six SCA14 families have been identified.\(^\text{16}\)

---

<table>
<thead>
<tr>
<th>SCA</th>
<th>Syndrome</th>
</tr>
</thead>
<tbody>
<tr>
<td>SCA12</td>
<td>Tremors, common in India</td>
</tr>
<tr>
<td>SCA13</td>
<td>Mental retardation</td>
</tr>
<tr>
<td>SCA15</td>
<td>III</td>
</tr>
<tr>
<td>SCA17</td>
<td>Psychiatric features, dementia, chorea</td>
</tr>
<tr>
<td>SCA28</td>
<td>Slow saccades, ophthalmoplegia</td>
</tr>
</tbody>
</table>

*SCA8 – The clinical validity of genetic testing for SCA8 by CAG repeat sizing, has not yet been established, thus SCA8 testing should not be offered as a routine genetic test if family history is unknown. However, SCA8 testing may be appropriate in large pedigrees where the expansion has been proven to be segregating with the disease. It is still important to note in the report that finding an expansion for SCA8 in a patient does not exclude the presence of another causative mutation.*
3 Patient Pathways

3.1 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.

**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. Other ataxias are not normally associated with cardiological problems. *See section 4.1 for information on treatments.*

**Neuro-ophthalmologist / medical retina specialist / ophthalmologist with specialist expertise in genetics**
Many of the ataxias are associated with eye symptoms such as reduced vision, diplopia or oscilllopsia due to nystagmus. A referral to a neuro-ophthalmologist and other specialist services is recommended since in some cases treatment may be available. *See section 4.1 for information on treatments.*

**Neuropsychologist**
Some ataxias may be associated with cognitive problems, therefore in selected cases a referral to cognitive testing may be relevant. Examples of such ataxias are SCA1, SCA2, SCA3 and DRPLA. Friedreich’s ataxia, the most common inherited ataxia, is not thought to be associated with cognitive problems. Cognitive evaluation may be particularly relevant to determine capacity.

**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 also be helpful. *See section 5.2 for information on physiotherapy.* 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 can sometimes be experienced. *See section 4.1 for information on treatments.*
**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 (e.g., balance, upper-limb coordination, speech and swallowing). 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. See section 5.2 for information on physiotherapy.

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. See section 5.1 for information on speech and language therapy.

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. Occupational therapists work in various health and social care settings. As a general rule, local authority based (social services) occupational therapists have a prime focus on home adaptations and equipment provision. A referral to a community rehabilitation team or neurological outpatient setting should be considered for further assessment of specific areas. Specialist neurological hospitals may offer expert assessment via clinics. For further information on referrals contact the College of Occupational Therapy ([www.cot.co.uk](http://www.cot.co.uk)). See section 5.3 for information on occupational therapy.

**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\(^\text{17}\) and counselling may be valuable.

**Patient Support groups**

Referral to a patient support organisation is recommended. Ataxia UK provides support to people with all ataxias. The Sarah Matheson Trust provides support solely to people with multiple system atrophy, the AT Society supports people with ataxia-telangiectasia and for support on Niemann-Pick Disease there is the Niemann-Pick Disease Group. When progressive ataxia is first diagnosed often patients will not have heard of the condition and will not know anyone else with it. At this stage the support that can be provided by patient support organisations can be crucial. The possibility of meeting others in the same situation, receiving emotional support and information from a Helpline and finding out how others cope with the symptoms, can be of much benefit to people with ataxia. Although each support organisation provides its own services, many will also provide the opportunity for patients to be informed about research developments and take part in research projects.
3.2 Reviews and follow-up

The first follow-up appointment with the neurologist or ataxia specialist after a diagnosis of progressive ataxia is made should be within six weeks, even if the specific cause of the patient’s ataxia has not at this stage been identified. Patients are likely to have many questions to ask their neurologist once they have had a chance to think about the implications of the disorder. Following any definitive diagnoses (for example, genetic ones) further urgent follow up appointments may be warranted, including to discuss the onward referral to other specialists.

Patients should then be offered 6-12 monthly reviews from a neurologist. If it is difficult for patients to travel to the hospital, 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, if patients are discharged and not offered a follow-up appointment they are not likely to benefit from medical advances. For example, new diagnostic tests are regularly becoming available, especially as new genes are identified, thus increasing the possibility of identifying a diagnosis for patients with ‘idiopathic’ cerebellar ataxia. In addition, new treatments may be developed, both ones that may affect disease progression and ones for symptomatic relief.

Clinicians should consider the use of validated ataxia-specific rating scales for measuring progression of the ataxias (see section 6 for details of these scales).

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. It is recommended that patients should be screened once every two years before any cardiac disease is documented, and annually after there is manifestation of asymptomatic cardiac disease (see section 4.1). 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. Travelling long distances to hospitals regularly may be difficult due to practical mobility issues or logistics.
 Individual patients and their GPs need to decide for themselves what level of secondary (versus primary) care is required. 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. Multi-disciplinary, multi-professional working practices that are mutually supportive are important, as recommended by the National Service Framework for long-term conditions. As ataxia is usually chronic and progressive, an even greater reliance on community services with the passage of time is likely. The establishing of durable networks of care at an early stage is therefore crucial. In line with recent recommendations in the National Service Framework for long-term conditions, involvement of symptom and palliative care professionals is recommended, especially as the disorder progresses. The remit at this stage may include providing practical and emotional support for carers, who are often family members (see section 7).
Medical Interventions

4.1 Symptomatic treatments

Patients with progressive ataxia may experience a variety of symptoms, some of which can be treated medically. Specific treatments in these situations should be considered (as described in the sections below). Additional useful sources of information are a recent systematic review of treatments for degenerative ataxias and a review on the diagnosis and treatment of Friedreich’s ataxia.

Cardiac problems

It is essential to involve a Cardiologist in the management of cardiac complications in patients with Friedreich’s ataxia. It appears that as there is phenotypic variation in the neurological presentations of Friedreich’s ataxia, a similarly broad clinical spectrum is seen with cardiac pathology. Generally, however, cardiac involvement is frequent, exceeding 90% of the patients in some studies and reports.

Cardiac abnormalities seen in Friedreich’s ataxia include:

- Evidence of electrocardiographic abnormalities in most, mainly in the form of non-specific repolarisation abnormalities.
- Echocardiographic abnormalities in about 50% usually in the form of concentric hypertrophy of the ventricular myocardium. The hypertrophy is typically mild and is recognised more frequently in younger people. Whether this is suggestive of regression of the hypertrophy during the course of the disease or describes variation in the phenotype is unclear. It is likely though that there is some degree of regression of the hypertrophy in these patients resulting in thinning and dilatation of the left ventricle with time.
- Arrhythmia, predominantly supraventricular.
- Severe or terminal systolic dysfunction with dilation of the ventricle, especially at older ages, but this is less frequent.

Although clinical observations and research have offered new insights into the disorder, the natural course of the cardiomyopathy in Friedreich’s ataxia is largely unknown. Studies have suggested that the electrocardiographic and echocardiographic changes are consistent with the size of the GAA expansion and the neurologic deficit.

Management

At a clinical level, as in all other cardiomyopathies, increased awareness is the key factor for the recognition, diagnosis and management of patients who suffer from these abnormalities. Preventative cardiac screening could possibly have a role, but since experience suggests that the cardiac abnormalities are gradual and progressive, the point at which to start investigating depends on the clinical status. In general, expert opinion
would suggest that it would be advisable for patients to be screened once every two years before any cardiac disease is documented, and annually after manifesting features of asymptomatic cardiac disease. However, if symptoms (such as palpitations, breathlessness or loss of consciousness) arise, patients should see their GP, who should consider an earlier referral to a cardiologist. When required, the cardiac screening should include electrocardiography, echocardiography and (depending on the indication) cardiac MRI and rhythm monitors, such as Holters.

The management of cardiac problems is mainly symptomatic. Therefore, depending on the symptoms, patients would receive anti-arrhythmic and anti-cardiac failure medication. Although not validated specifically in clinical studies with Friedreich’s ataxia patients, it is reasonable to consider beta-blockers, ACE inhibitors/other vasodilators, spironolactone and loop diuretics for the management of patients who present with heart failure symptoms attributed to myocardial dysfunction. Amiodarone and digitalis have roles in the management of atrial fibrillation; rate control can be achieved also with beta-blockers or calcium antagonists, depending on the clinical circumstances. An issue which often needs lengthy multidisciplinary discussion is the type of anticoagulation for atrial arrhythmias in these patients, who are prone to falls and injury. The decision is usually individualised.

Recent advances in research have generated expectations of a novel approach for the treatment of cardiac (and neurological) lesions with the use of antioxidant agents, such as idebenone. The first sets of results are encouraging and are currently undergoing further validation with larger randomised controlled trials (see section 4.2). Furthermore, systematic study and follow-up of patients with Friedreich’s ataxia who exhibit cardiac abnormalities will provide the longitudinal data required to shed light on many aspects of the cardiac manifestation of this disorder, and may help prevent complications and/or delay their progression.

**Bladder problems**

Some people with ataxia experience bladder problems, which whilst not curable can almost always be managed. Whilst the damage to the nervous system is often responsible for these symptoms, as individuals age they are also liable to develop urinary conditions which can affect the population at large, notably prostatic enlargement in older men or stress incontinence in women. If the nerve damage is progressive and affecting the bladder, bladder behaviour may change over time and may require reassessment. Lack of mobility and incoordination add to the problems both of coping with the symptoms and of managing the urinary symptoms.

A thorough continence assessment should be conducted in primary care, with examination for a palpable bladder, digital examination of the prostate through the back passage, genital abnormalities, excoriation secondary to the incontinence etc. Obtaining a frequency volume chart is helpful and assessment of residual urine with a hand-held bladder scanner should be done if residual urine is suspected or initial management is not successful.

Practical advice on the avoidance of smoking, alcohol and caffeine-containing drinks should also be given, and the importance of regular voiding should be explained. A referral to a continence advisor may be appropriate, and occasionally for special assessments/treatments referral to an urologist or a gynaecologist may be needed.

The overactive bladder (presenting with urgency, precipitancy and urge incontinence) is the commonest problem encountered. General advice about fluid intake and regular voiding can be combined with the specific use of anti-cholinergic drugs (oxybutinin, propiverine, solifenacin, trospium, propantheline, tolterodine or darifenacin). In this situation, a post-micturition ultrasound scan of the bladder after the
Initiation of treatment or dose escalations are recommended to exclude retention. If these bladder control treatments are not successful, botulinum toxin injections can be considered.

Failure to empty the bladder leaving residual urine often presents with very similar symptoms to those of the overactive bladder, hence the need for checking residual urines when initial management is not successful. Severe cases of incomplete emptying will have overflow incontinence and need urgent specialist assessment. Problems with emptying the bladder are seen particularly in MSA and SCA3.

Residual urine can be due to loss of the ability of bladder muscle to contract, or to obstruction at the sphincter level either by muscle spasm secondary to the nerve damage or to prostatic enlargement. This distinction can only be made by urodynamic tests measuring bladder pressure during voiding. However both can be managed by catheter drainage, either continuous or intermittent, in the short and long-term. It may be possible to re-establish normal voiding particularly if benign prostatic hypoplasia is present. It is sometimes surprising who can cope with self-catheterisation, and in some cases partners or carers may be willing and able to do this. Specialist assessment will often be necessary.

In the occasional cases of stress incontinence referral for physiotherapy would be appropriate. Whilst pelvic floor exercises will be difficult for many individuals, there are useful biofeedback and stimulation techniques available which may be of value.

**Sexual dysfunction**

Erectile dysfunction may occur in progressive ataxias and is known to commonly be the first manifestation of autonomic dysfunction in multiple system atrophy. Phosphodiesterase-5 inhibitors are the mainstay of treatment, eg sildenafil, tadalafil and vardenafil, but these may exacerbate low blood pressure on standing. Treatment goals should be balanced between the needs of the person and potential side effects of medications, eg hypotension.

**Muscle spasms and spasticity**

Muscle spasms can be experienced by patients with ataxia, and there are effective treatments that can be prescribed. Referral to a physiotherapist is also appropriate *(see section 5.2)*. A comprehensive review of the management of spasticity can be found in the NICE clinical guidelines on the management of multiple sclerosis (February 2004).

**Contractures**

Referral to a physiotherapist and/or orthopaedic surgeon is appropriate *(see section 5.2)*. A review of the management of contractures can be found in the NICE clinical guidelines on the management of multiple sclerosis (February 2004).

**Tremors**

A variety of types of tremors may be present in patients with ataxia. Cerebellar intention tremor is the commonest, and can be difficult to treat. In open-label trials and case reports benefit has been reported for propranolol, primidone, glutethimide, carbamazepine, isoniazid, clonazepam, buspirone, and topiramate. Propranolol, primidone and topiramate are commonly used in other tremor disorders, including essential tremor, but generally speaking good-quality, randomised controlled trial data for the use of any of the agents listed in the management of cerebellar tremor is lacking. The published data up to 2005 have been reviewed by Seeberger. A referral to a physiotherapist may be useful; *(see section 5.2)* for some possible physical interventions for tremors.
There may be a growing role for functional neurosurgery, including deep brain stimulation (DBS), in the management of these symptoms, but here too studies specifically in cerebellar tremor treatment are lacking. In a study of a patient with SCA2, chronic thalamic stimulation was shown to improve severe resting and action tremor. More recently, improvement in unilateral cerebellar tremor in a single subject has been described after Vim (nucleus ventralis intermedius) and latterly PSA (posterior subthalamic area) DBS. In patients where tremor is extremely debilitating and not responsive to medication, a referral to a centre specialising in thalamic stimulation should be considered.

**Dystonia**
A number of treatment options for dystonia, including oral drugs, botulinum toxin injections, surgical techniques and physiotherapy are available. A comprehensive review of the treatment of dystonia has recently been published.

**Depression and other psychiatric symptoms**
Patients with ataxia, as those with other neurological conditions, are susceptible to depression. Practical and effective treatment of symptoms of depression can be carried out in primary care, without the need for specialist psychiatric input. In addition to pharmacological interventions, counselling can offer significant benefits. In special cases, more input from secondary level psychiatric services may be indicated, especially if depression is severe and has not responded to an adequate course of antidepressants or if there is suicidal risk. The less common symptoms of dementia or psychosis may also require input from specialist psychiatric services.

**Fatigue**
Fatigue is the sensation of not having enough energy to carry out both mental and physical activities. Individuals with ataxia due to cerebellar atrophy have reported fatigue with normal everyday activities. Fatigue is reported in many chronic neurological disorders and is thought to be due to several different factors, some of which remain unknown. The extra effort needed to compensate for the loss of coordination may be one such factor. In addition, disturbance of the normal sleep pattern can lead to excessive daytime somnolence (which is not the same as fatigue). Evaluation by a specialist clinic may help to decrease the burden of fatigue after a thorough evaluation and treatment plan.

**Eye symptoms**

**Nystagmus**
Nystagmus often causes decreased visual acuity, as well as sometimes causing oscillopsia (a disabling subjective sensation of movement of the visual world). The eye movements do not require treatments if patients are asymptomatic, however therapy should be considered when visual disability is present (see section 3.1 on recommended referrals). There have been a few randomised controlled trials; these have shown the efficacy of gabapentin in treating symptomatic pendular or gaze-evoked jerk nystagmus, and sometimes downbeat nystagmus. Downbeat nystagmus can also be treated with 3,4-diaminopyridine and baclofen. A number of other studies (non-randomised controlled trials) have shown the efficacy of other medications such as clonazepam and valproate for pendular nystagmus and of 4-aminopyridine for downbeat nystagmus. Botulinum toxin injections have also been reported to be beneficial in some patients, although there are limitations to this approach (discussed in a review in 2002).

**Diplopia**
Diplopia (seeing ‘double’) is not a common symptom associated with the ataxias but does occur. The images are usually separated horizontally (rather than vertically) as a result of a manifest exo- or eso-
deviation of the visual axes, and is concomitant in all directions of gaze. Single vision can be restored using prisms, so eye muscle surgery is rarely necessary.

**Visual impairment**
Damage to the retina or optic nerves is well recognised in association with several of the ataxias. Approximately half of patients with Friedreich’s ataxia have evidence of optic tract disease but severe visual loss is uncommon. Progression deterioration in visual acuity is often seen in patients with SCA7 mutations because of bilateral (and usually symmetric) maculopathy. At this stage there is still no known treatment either to prevent or to treat the retinal or optic nerve manifestations of ataxic disorders, but patients can benefit from a wide range of low vision aids as well as having their visual disability registered.

**Pain**
Neuropathic pain can be a symptom of some ataxias, including Friedreich’s ataxia. There are medications that can be used to treat this symptom. For more information contact the Neuropathy Trust (www.neurocentre.com). Referral to a pain management clinic may be helpful for some patients who experience pain.

**Other Symptoms**
There are no proven treatments for other symptoms so it is not possible to give specific recommendations. There is some evidence, however, for the efficacy of other medications from pilot clinical trials or expert experience. For example, a number of drugs have been used to treat ataxia, but with limited success.

**Episodic ataxias**
Some patients present with an episodic ataxia. Episodic ataxia type 1 and 2 are the most well known but other rarer forms are being identified. In patients with episodic ataxia type 2 (the most common form) ataxic episodes can often be treated with the carbonic anhydrase inhibitor acetazolamide. Long-term use of this drug is associated with the development of kidney stones as a side-effect, hence preventative measures such as drinking citrus juices should be encouraged. As stress often triggers attacks, stress management techniques (eg meditation) can be helpful in controlling symptoms. Alcohol and caffeine should also be avoided, and regular but modest exercise should be encouraged.

Although acetazolamide is the standard treatment for episodic ataxia type 2, some clinicians are also using another carbonic anhydrase inhibitor, dichlorphenamid (Daranide), and the potassium channel blocker 4-aminopyridine has also been shown to prevent attacks in a small trial.

There are also medications that can be used for the treatment of episodic ataxia type 1, although there is some variation on the efficacy of different drugs between individuals. Case studies and other small studies suggest that medications such as acetazolamide, sulthiamine or phenytoin could be effective.

(See www.orphanet.net ORPHA37612 for EA-1 and link to other episodic ataxias.)

**4.2. Disease modifying treatments**

Some forms of progressive ataxia are treatable; hence the importance of ensuring a prompt and accurate diagnosis. For the majority of progressive ataxias there are no proven treatments.
**Friedreich’s ataxia**

In the case of Friedreich’s ataxia, as yet there are no results from large-scale studies justifying the use of any agents to prevent progression, or the development of complications. However, given its prevalence, this is an area of active research interest and a number of small treatment trials have been carried out which are showing some promise, and others are in the pipeline. Friedreich’s ataxia is caused by defects in a gene which results in a decrease in the essential protein frataxin. Cells from people with Friedreich’s ataxia are damaged due to oxidative stress and an accumulation of iron in the mitochondria. Due to these findings there has been much interest in testing the effect of antioxidants (eg idebenone, CoQ10). There are also a number of other studies testing the effect of iron chelators (eg deferiprone) and drugs that have the potential to increase frataxin levels.\(^{42}\)

In particular, there have been many small-scale studies completed thus far that show modest benefits from the antioxidant idebenone on cardiomyopathy, and in some studies also on ataxia rating scales.\(^{43-50}\) Multi-centre larger-scale idebenone trials are underway in Europe (including a study site in the UK) and in the US, to test the effect of idebenone on cardiomyopathy and on neurological outcome measures. Idebenone does not at the moment have approval from the European Medicine Evaluation Agency or the US Food and Drug Agency for the treatment of Friedreich’s ataxia, as results are awaited from the ongoing studies. However, idebenone does have market authorisation with conditions (pending the results of studies to verify its clinical efficacy) in Canada (two doses are approved: a starting dose of 450mg/day for patients below 45kg body weight and 900mg/day for patients above 45kg body weight and physicians have an option to dose up to 1,350mg/day for patients below 45kg body weight and up to 2,250mg/day for patients of more than 45kg body weight if needed).

The effects of Vitamin E and CoQ10 have also been tested in a small open-label trial (n=10) and a larger double-blind controlled study (n=50). In the open-label trial the combination has shown the same benefit on cardiomyopathy as has idebenone and provisional data has shown some stabilisation of neurological function.\(^{51}\) The controlled trial compared a low-dose versus high-dose and no statistical significant difference was shown in ataxia rating scales. (High dose used in the trial was: 2100IU/day Vitamin E and 600mg/day CoQ10 for adults and 30IU/kg/day Vitamin E and 9mg/kg/day CoQ10 for patients under 18 years of age. Low dose used in the trial was: 30mg CoQ10 and a placebo which contained Vitamin E (4IU/day) as a preservative).\(^{52}\)

However, the trial did show that 49% of people improved whether on high or low dose compared with cross-sectional data of patients not on the trial, and that this group had lower baseline levels of CoQ10 than those who did not respond. The authors suggest therefore that levels of CoQ10 in the serum are a predictor to response to CoQ10 and Vitamin E treatment.\(^{52}\)

**Gluten ataxia**

Gluten ataxia has been recently defined as a sporadic otherwise idiopathic cerebellar ataxia associated with the presence of serological evidence of gluten sensitivity (antigliadin (IgA or IgG) or endomysium or transglutaminase antibodies) and has been shown in a one-year controlled trial to be responsive to a gluten-free diet.\(^{53}\)

There is however some controversy reported in the literature as to whether the tests used to diagnose gluten ataxia (particularly antigliadin antibodies) are specific enough to be confident of such a diagnosis.\(^{54}\) A recent study has identified a new antibody against another transglutaminase (TG6) that may prove to be
more sensitive and specific for the neurological manifestations of gluten sensitivity. This test is not yet widely available. Neurologists are often faced with weighing up the options of whether or not to recommend a gluten-free diet. For those patients with gluten ataxia who also have an enteropathy, the recommendation is that they should go on a gluten-free diet without delay. For those patients with ataxia in which an enteropathy cannot be established, but who have serological evidence of gluten sensitivity, it would be reasonable to offer a gluten-free diet, given that the potential harm of such a treatment is low, there are no alternative treatment options and that there is a chance that a patient may benefit. The trial mentioned above did demonstrate that even those patients without enteropathy benefited from a gluten-free diet. Patients should however be made to understand that such benefit can only be seen with strict adherence to the diet with evidence of elimination of the antibodies that need to be tested on a six-monthly basis. Any improvement or stabilisation usually manifests within a year on a strict diet. In some patients where there is evidence of cerebellar atrophy the expected benefit will be in the form of stabilisation rather than improvement.

**Treatable progressive ataxias**

**Ataxia with Vitamin E deficiency**

Patients diagnosed with ataxia due to Vitamin E deficiency should be given Vitamin E supplements (diagnosis of Vitamin E deficiency needs to be made in the context of serum lipid levels, also known as lipid-adjusted Vitamin E, as estimation of free levels of Vitamin E is not reliable and can be misleading). Studies have shown this leads to cessation of progression of neurological symptoms and mild improvement in certain patients, especially in the early stages of the disease. (For further information see [www.ncbi.nlm.nih.gov/omim; OMIM number 277460](http://www.ncbi.nlm.nih.gov/omim; OMIM number 277460)).

**Patients who have ataxia with Vitamin E deficiency often have similar symptoms to Friedreich’s ataxia and this has at times resulted in misdiagnoses**

**Wilson’s disease**

Wilson’s disease is an autosomal recessive inherited disorder of copper metabolism, resulting in pathological accumulation of copper in many organs and tissues. The leading neurologic symptoms in Wilson’s disease are dysarthria, dyspraxia, ataxia, and parkinsonian-like extrapyramidal signs. Symptoms may be fully reversible on treatment with zinc or with copper chelators. (For further information see [www.ncbi.nlm.nih.gov/omim; OMIM number 277900](http://www.ncbi.nlm.nih.gov/omim; OMIM number 277900)).

**Ataxia with CoQ10 (ubiquinone) deficiency**

Primary muscle Coenzyme Q10 (CoQ10) deficiency is an apparently autosomal recessive condition with heterogeneous clinical presentations. Patients with these disorders improve with CoQ10 supplementation, hence the importance of early diagnosis. It has been observed in children as well as in adults with later onset cerebellar ataxia. The prevalence in the UK is currently unknown. Testing of CoQ10 deficiency can be performed on skeletal muscle samples or on blood mononuclear cells. For details on testing see section 2.2. (For further information see [www.ncbi.nlm.nih.gov/omim; OMIM number 60742](http://www.ncbi.nlm.nih.gov/omim; OMIM number 60742)).
Cerebrotendinous xanthomatosis
Cerebrotendinous xanthomatosis is a sterol storage disorder characterized by the accumulation of cholestanol and cholesterol in tendon, the central nervous system and in the bile. Treatment with chenodeoxycholic acid results in at least partial reversal of the neurological symptoms and in cognitive function in some patients. Early diagnosis and initiation of therapy is important. (For further information see www.ncbi.nlm.nih.gov/omim; OMIM number 213700)

Treatable causes of childhood ataxia
Some of the treatable conditions mentioned above can be seen in children, including:
CoQ10 (ubiquinone) deficiency, Episodic ataxia type 2 (intermittent ataxia), Vitamin E deficiency, Cerebrotendinous xanthomatosis and Wilson’s disease.

In addition, the following conditions are also treatable causes of childhood ataxia:

Glucose transporter 1 deficiency (often intermittent) (Glut-1 DS)
Impaired glucose transport across the blood-brain barrier results in Glut-1 deficiency syndrome, characterised by infantile seizures, developmental delay, acquired microcephaly, spasticity, ataxia, and hypoglycorrhachia. A ketogenic diet has been found to be effective treatment. (For further information see www.ncbi.nlm.nih.gov/omim; OMIM number 606777; also contact CLIMB (www.climb.org.uk.)

Hypobetalipoproteinaemia
Hypobetalipoproteinaemia is a rare disorder characterized by low levels of fats, beta-lipoproteins and cholesterol.
(For further information see www.orpha.net ORPHA426; also contact CLIMB www.climb.org.uk)

Hartnup disease
Intermittent ataxia, psychotic behaviour and mental retardation are features of this condition. (For further information see www.ncbi.nlm.nih.gov/omim; OMIM number 34500. Also contact CLIMB www.climb.org.uk.)

Biotinidase deficiency
This is a metabolic disorder characterised primarily by cutaneous and neurologic abnormalities. Can be treated with biotin. (For further information see www.ncbi.nlm.nih.gov/omim; OMIM number 253260. Also contact CLIMB www.climb.org.uk.)

Pyruvate dehydrogenase deficiency
This is a metabolic disorder. (For further information see www.orpha.net ORPHA765 or contact CLIMB www.climb.org.uk).

Structural disorders
These causes are usually surgically treatable. Brain tumours can cause ataxia but usually other symptoms such as headache, vomiting and personality change are present. (For more information see www.directorycancer.com/brain-tumours).

Hydrocephalus can have similar symptoms and can be due to a wide variety of causes.
(For more information contact the Association for Spina Bifida www.asbah.org).
Arnold-Chiari malformation is a congenital malformation at the back of the brain. (For more information contact the Association for Spina Bifida www.asbah.org).

**Acute encephalopathies**
Intoxication, either from recreational or medically prescribed drugs, can cause acute or intermittent ataxia, but is not always immediately recognised. *For more information about metabolic encephalopathies such as branched chain amino-acidurias contact CLIMB* (www.climb.org.uk).

**Non-convulsive status epilepticus / other epilepsies**
Can rarely present as intermittent ataxia (combined with altered consciousness) but seen more commonly in the context of a known epilepsy syndrome. *For more information contact Epilepsy Action* (www.epilepsy.org.uk).

**Sensory ataxias**
Refsum syndrome and chronic inflammatory demyelinating neuropathy (CIDP) are conditions affecting the nerves; Refsum also involves the cerebellum. *For more information see the National Institute of Neurological Disorders and Stroke* (www.ninds.nih.gov).
A full range of therapies should be available for patients with ataxia. These should include: physiotherapy, speech and language therapy, occupational therapy, dietetics, psychological support and support from social services.

5.1 Speech and language therapy

The progressive ataxias may affect communication and/or swallowing function. The most obvious communication difficulty encountered is that of dysarthria which is a motor speech disorder resulting in altered speech clarity, naturalness and intelligibility. Communication may also be affected in cases where there is an associated cognitive impairment impacting on language processing. In addition any difficulties with executive functions may result in changes in communication behaviour.

Difficulty in swallowing is a common symptom of ataxia, particularly as the disease advances. Depending on the pathophysiology of the disease, swallow dysfunction (dysphagia) may occur at the oral, pharyngeal and/or oesophageal stage of swallowing. For example, when there is cerebellar involvement, dysphagia may be characterised by reduced coordination of the oro-pharyngeal muscles involved in swallowing food and drink.

Communication problems

Speech

Dysarthria is a common symptom of the progressive ataxias. The main features vary, and may include any, or a combination of articulatory imprecision, excess and equal stress, harsh and/or tremulous voice quality, and slowed speech rate. Atactic dysarthria appears to be related to a disturbance in the neural mechanisms that underlie the coordination, temporal regulation, and quasi-automatic control of respiratory, phonatory, and articulatory movements for speech. Some researchers view atactic dysarthria as reflecting a global impairment of the respiratory, laryngeal, and articulatory subsystems of speech, although individual variations may be seen in the relative severity of these impairments according to the type of ataxia and disease duration. Deterioration in dysarthria in the spinocerebellar ataxias (SCAs) tends to be slow (eg in one study deterioration was observed over a period of three years). Furthermore, patients with early disease onset (before age 24) have more deterioration in voice quality as compared to patients with late disease onset (after age 43). In addition to atactic dysarthria, patients may also present with features of spastic, bulbar or flaccid dysarthria reflecting a more diffuse pathophysiology. Features of spastic dysarthria have been noted in studies, including perceptions of a strained-strangled voice quality as part of the presentation. It should be noted that the occurrence of symptoms other than those associated with an ataxia is variable, both across individuals as well as disease progression.
Changes in communicative behaviour
There is a growing evidence base in the field of cerebellar disease that some patients may present with impairments in executive function.\textsuperscript{72,73} The presence of moderate to severe executive dysfunction was seen in a small group of subjects with spino-cerebellar ataxia in one study\textsuperscript{74}, whereas another study describes only mild difficulties.\textsuperscript{72} Altered communication associated with executive dysfunction includes difficulty in organising and planning verbal messages resulting in disrupted narrative sequences, also difficulty in generating ideas, judging and weighing options and forming inferences on information heard and read. Rapoport et al emphasised the clinical prudence of being on the alert for cognitive disturbances when assessing, treating, and rehabilitating patients with cerebellar disease.\textsuperscript{75} Speech and language therapists (SLTs) should also be on the alert for any signs of cognitive communication difficulties in patients with progressive ataxia.

Auditory processing disorders
Findings from a small study of ten individuals with Friedreich’s ataxia, suggest that a relatively high proportion of individuals may suffer auditory processing difficulties (particularly with speech understanding in background noise), despite in most cases showing normal sound detection levels on audiogram.\textsuperscript{76}

Impaired communication is perceived by patients with neurological disease as one of the most difficult aspects of their disease.\textsuperscript{77} Reports of patients feeling embarrassed about speaking, having difficulty talking on the telephone and having reduced confidence in communication leading to social isolation are commonly encountered in clinical practice.

Assessment of communication
SLTs undertake a comprehensive assessment of each patient’s communication in the context of their life roles and wishes. Assessment may include perceptual and objective measures of motor speech function and cognitive linguistic functions. The evaluation of the impact of communication difficulties on the individual’s participation in activities of daily living and life roles is essential. Obtaining the views of the family and/or carers where appropriate is also highly recommended. Management may be targeted at the level of impairment, activity limitation or participation restriction, or any combination of these, based on the ICF framework (International Classification of Functioning, Disability and Health, WHO 2001).\textsuperscript{78}

Management of communication
Motor Speech Disorders
Based on the findings of a comprehensive assessment, an individualised treatment programme is devised. A programme may target the presenting speech impairment, as well as issues relating to the patient’s activity and/or participation.\textsuperscript{78} Previous studies of therapy for ataxic dysarthria have shown only modest improvements in speech intelligibility when targeting individual speech parameters, such as respiratory support for speech and articulatory coordination and clarity in isolation.\textsuperscript{66,79} A study reported a short and long-term improvement in phonatory and articulatory functions, speech intelligibility, and overall communication and job related activity following an intensive course of Lee Silverman Voice Treatment (LSVT) in a single case study of cerebellar dysarthria resulting from thiamine deficiency.\textsuperscript{68} The LSVT programme focuses on training patients to maximise their phonatory efficiency and loudness by being loud.\textsuperscript{60}
Management may also involve assisting patients to develop increased self-monitoring of their speech quality and identification of helpful speech strategies. Strategies may include use of over-articulation, shorter phrases and breath top-ups.

When speech intelligibility levels fall below 50% or when reduced intelligibility has a significant impact on functional communication, alternative and augmentative means of communication (AAC) should be considered. The SLT should discuss AAC options with each individual, their communication partners, and the multidisciplinary team. The individual’s language skills, cognitive functioning, motor and perceptual skills, and communication environment are all taken into consideration. AAC can take the form of simple systems such as pen and paper or use of an alphabet chart to supplement speech. Some patients may benefit from a high technology aid such as a Lightwriter with a choice of written and/or voice output. Switch activated communication aid systems should be trialled in cases where there is severe upper-limb and truncal ataxia which cannot be adequately alleviated by appropriate seating and set up. SLTs are frequently involved in sourcing funding for AAC equipment for individual patients.

**Management of cognitive communication difficulties**

Speech and language therapy management of cognitive communication difficulties will encompass education about the underlying impairments impacting on communication. The SLT will identify strategies to manage communication breakdown with the individual and their key communication partners. These strategies are practised within a supportive, therapeutic environment. Strategies include verbal and visual prompts which can be used by the individual or the communication partner in conversations to assist with topic management and turn-taking.

**Management of auditory processing disorders**

Management of auditory processing disorders is challenging. The provision of conventional hearing aids (which are designed to make sounds louder, rather than make signals clearer), tend not to be useful. Provision of communication training including the use of listening strategies and lip-reading may be helpful in optimising understanding as well as increasing awareness of the importance of quiet listening conditions. Radio-frequency listening devices have been reported to improve signal to noise ratio in a noisy environment such as an office. SLTs are best placed to advise patients on communication strategies. Additional support is available from hearing therapists attached to Audiology Departments or Sense Teams in the community.

**Swallowing problems (dysphagia)**

Dysphagia in the progressive ataxias is often gradual and insidious in its onset. Studies indicate the cerebellum’s role in controlling the speed of oral muscle movements, and clinical experience is that the oral phase of the swallow is frequently affected. Abnormal pharyngeal and oesophageal function were identified in a small group of patients with hereditary sensory ataxia.

Symptoms of dysphagia are listed in Table 7 and include difficulties controlling food or drink in the mouth, chewing, dribbling, coughing or choking on food or drink. Swallowing difficulties may be exacerbated when there are co-existing postural or hand-to-mouth coordination difficulties. Patients with dysphagia are at risk of malnutrition, dehydration, and recurrent chest infections. Co-occurring cognitive impairment may place patients at additional risk due to their reduced ability to self-monitor and maintain a safe approach to eating and drinking, including compliance with recommended textures and safe swallowing strategies. Dysphagia can have a significant impact on quality of life, with patients reporting taking extra time for meals, embarrassment associated with eating and drinking leading to avoidance of social gatherings, and fear of choking.
Assessment of swallowing

The SLT will take a case history from the patient and/or family/carers. A comprehensive case history includes identification of signs and symptoms of dysphagia; current eating and drinking behaviour including individual dietary preferences; nutritional status and food supplementation. Given the progressive nature of dysphagia associated with ataxias, there may be under-reporting of swallowing difficulties as patients adopt compensatory approaches to their oral intake, including avoidance of particular food or liquid types. Rigorous questioning and a thorough examination for all signs and symptoms of dysphagia is therefore recommended (see Tables 6 and 7 for signs and symptoms of dysphagia).

SLTs conduct a full clinical examination of swallowing function comprising oral motor and sensory examination and observation of the patient during oral intake. An instrumental examination of swallowing is indicated when information gained from clinical examination is not sufficient to guide management of the presenting dysphagia. Instrumental examinations include videofluoroscopy (radiographic procedure) and/or a fibreoptic endoscopic evaluation of swallowing (FEES).

Table 6: Signs of dysphagia

<table>
<thead>
<tr>
<th>Signs of dysphagia</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Recurrent chest infections</td>
</tr>
<tr>
<td>• Weight loss</td>
</tr>
<tr>
<td>• Dehydration</td>
</tr>
<tr>
<td>• Poor oral hygiene</td>
</tr>
<tr>
<td>• Observed coughing or choking during oral intake</td>
</tr>
</tbody>
</table>

Table 7: Symptoms of dysphagia

<table>
<thead>
<tr>
<th>Symptoms of dysphagia</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Dribbling</td>
</tr>
<tr>
<td>• Difficulty chewing food</td>
</tr>
<tr>
<td>• Food pocketing in the mouth or sticking in the throat</td>
</tr>
<tr>
<td>• Coughing</td>
</tr>
<tr>
<td>• Choking during oral intake</td>
</tr>
<tr>
<td>• Nasal regurgitation</td>
</tr>
<tr>
<td>• Avoiding specific food or liquid consistencies</td>
</tr>
<tr>
<td>• Anxiety associated with oral intake</td>
</tr>
<tr>
<td>• Taking long time to complete meals</td>
</tr>
<tr>
<td>• Avoidance of social eating</td>
</tr>
</tbody>
</table>
Management of dysphagia

Following comprehensive assessment, the SLT advises on management of the dysphagia. Dysphagia management is best conducted within a multidisciplinary team (MDT). The SLT works closely with the dietitian, to ensure optimal nutrition and hydration via the oral and/or alternative route, as well as the physiotherapist / occupational therapist, to ensure optimal feeding/positioning and use of aids or adaptations to deliver food to mouth. There is growing clinical interest in the use of oral muscle strengthening exercises for swallowing in degenerative conditions although the evidence base is not well established. Strengthening exercises should specifically target underlying swallowing pathophysiology eg the use of the Shaker exercise to target reduced anterior hyoid tilt. Examples of management techniques are listed in Table 8.

Table 8: Dysphagia management techniques

<table>
<thead>
<tr>
<th>Dysphagia management techniques</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Modification of consistency of food or drink</td>
</tr>
<tr>
<td>• Introduction of safe swallow strategies including use of a chin tuck position, double swallow, throat clear</td>
</tr>
<tr>
<td>• Advice regarding sitting posture and set up for oral intake</td>
</tr>
<tr>
<td>• Introduction of carer-initiated prompts to maintain safety eg slow rate, small sips, avoiding talking with food or drink in mouth</td>
</tr>
<tr>
<td>• Advice on oral hygiene care</td>
</tr>
</tbody>
</table>

Management of severe dysphagia should include consideration of alternative feeding routes when the oral route is no longer a viable option for maintaining adequate nutrition and hydration. Alternative feeding options for example percutaneous endoscopic gastrostomy tube (PEG) feeding should be discussed with the individual, their family and the MDT.

Speech and language therapy services

It is clinically accepted that patients benefit from being seen at any stage during the disease progression and when they are experiencing specific difficulties with either their communication and/or swallowing. Provision of relevant and timely information is integral to the patient developing an understanding of their disease and supports the ‘expert patient role’, empowering patients to take responsibility for managing their condition effectively. It is recommended that an open referral system should be in place where patients are able to access help from a SLT as and when required. Due to the nature of the progressive ataxias, speech and language therapy input needs to change over time in line with patient need. The newly diagnosed patient and their families may benefit from information regarding help that is available in the future should they require this, and symptoms to be aware of that would warrant assessment and advice from a SLT. When symptoms become more disabling, the SLT will take an active role in providing appropriate exercises or strategies to optimise communication and/or swallow function. Later in the disease, the SLT may be involved in advising on augmentative communication systems and/or alternative routes for nutrition and hydration.
5.2 Physiotherapy

Introduction
This section has systematically examined the published literature to provide considered guidance to physiotherapists working with people with progressive ataxia. There are few randomised controlled trials to draw upon to inform the treatment and management of ataxia, or the role of the physiotherapist. The majority of intervention studies reviewed were small case studies or case series designs. One systematic review of nine studies investigating the effectiveness of physiotherapy for adults with cerebellar dysfunction was identified. Little research has been conducted with people with spinocerebellar or Friedreich’s ataxia. Most research about ataxia rehabilitation has been carried out with people who have multiple sclerosis (MS), or extrapolated from heterogeneous studies of participants with MS, brain injury or children with cerebral palsy. A summary of the findings is seen in this section. For a full copy of the review and details of the methodology and methodological quality scores see the Physiotherapy supplement to this document.

Rehabilitation approaches
Thirteen papers were identified for review. Findings supported those reported in the systematic review by Martin et al (2009). Most studies (seven) were either small case studies or single case experimental designs with only one randomised controlled trial. Participants had wide ranging cerebellar pathology including MS, head injury, cerebellar stroke, brain tumour, cerebellar degeneration and central vestibular dysfunction; one child had Friedreich’s ataxia.

Interventions were individually tailored for all studies except one, varying in type, intensity, duration and frequency. Commonly reported interventions included PNF, Frenkel’s exercises, dynamic training of postural stability with task and activity focus, gait and balance training, along with strengthening and flexibility. Therapeutic equipment was often provided to support function. With the exception of three studies the majority of studies did not describe the intervention in detail and thus would be difficult to replicate in practice. A wide range of outcome measures were used and none of the papers fully reported the validity and reliability for this patient population. The long-term outcome was not consistently reported.

Four studies were considered of sufficient rigor to draw limited conclusions about the efficacy of physiotherapy. Although they were small studies they provide some evidence in support of physiotherapy being able to improve gait, balance and trunk control for people with ataxia and can reduce activity limitations and support increased participation.

Summary
- Dynamic task practice that challenges stability and explores stability limits and aims to reduce upper-limb weight bearing seems an important intervention for people with cerebellar dysfunction to improve gait and balance
- Strength and flexibility training may be indicated in conjunction with the above
- A compensatory approach (which includes orthotics and devices, movement retraining, reducing the degrees of freedom and optimising the environment) seems valuable for teaching people practical, everyday strategies and ways of managing the condition and may be particularly important for those with severe upper limb tremor
Specific interventions for gait

a) Treadmill training
Three studies investigated the effect of treadmill training for individuals at least one year post brain injury and presenting with ataxia. This intervention has not been tested in people with progressive ataxias. All studies were of good methodological quality and included one randomised controlled trial and two case studies. Training varied in duration, frequency, intensity (minimum 20 minutes three times a week for four weeks, to a maximum of daily training for 5 months). One study combined treadmill and over ground training. All studies used a combination of reliable gait parameters and functional outcome measures. Improvements were reported for all studies. The most functionally meaningful improvements were seen in one study which combined over ground training with body weight support treadmill training at an intensity and duration significantly greater than the other studies.

Summary
• Findings from treadmill training studies present encouraging evidence of the efficacy of this intervention for people with ataxia due to brain injury. Intensity and duration of training seem to be significant factors
• Consistent intensive training over many months combined with over ground training may be required

b) Visually guided stepping
Oculomotor and locomotor control systems interact during visually guided stepping ie the locomotor system depends on information from the oculomotor system during functional mobility for accurate foot placement. Crowdy and colleagues demonstrated, in two participants with mild cerebellar degeneration, marked improvements in oculomotor and locomotor performance following eye movement rehearsal. The authors suggest that rehearsal of intended steps through eye movement alone ie looking at foot target placement for each step, before negotiating a cluttered room, might improve performance and safety.

Summary
• This simple strategy, although task specific and short lived in nature, is promising and relatively quick and easy to apply in a functional setting

c) Balance and mobility aids
No studies have specifically evaluated the role of balance and mobility aids for people with ataxia. Clinical experience suggests walking aids should be considered on a case-by-case basis. Jeka reviewed a series of studies on postural control using light touch contact of fingertips or a walking aid as a means of balance. Results showed somatosensory cues from the fingertips provided a powerful reference orientation even when contact force levels were inadequate to provide physical support for the body. Clinical observation suggests that some individuals with ataxia find light touch contact more useful as a strategy than a conventional walking aid. This may explain why some people prefer to use Nordic poles, which help encourage light touch contact, rather than traditional walking sticks that tend towards force contact and a reduction in muscular forces acting through the lower limbs. Decreasing dependency of weight bearing
through the upper limbs in people with ataxia is also supported in another study. Furthermore individuals with cerebellar hemisphere lesions, who are more likely to have dysmetria and tremor, may find balance and mobility aids hard to use because placing and controlling a stick can be as difficult as trying to accurately place legs during swing phase. In a small study of healthy young adults walking aids have been found to compromise the ability to respond to balance disturbances through impeding lateral compensatory stepping and thus can affected safety.

**Summary**

- Light touch as a balance aid may be helpful for postural orientation and stability
- Upper extremity weight bearing during ambulation may perpetuate a deterioration or worsening of gait parameters
- Careful assessment is required for those with dysmetria, dysdiadochokinesia and tremor

**d) Axial weighting**

The effect of weighting the axial skeleton has been studied in five subjects with ataxic gait of unreported aetiology and 19 participants with mixed CNS pathology five of whom had ataxic gait. The latter study reported subjective improvements in gait and posture plus feelings of steadiness but low methodological quality means findings should be viewed with caution. Conversely the former study reported gait characteristics changed unpredictably with axial weights, worsening more often than improving, and concluded that use of axial weights to improve gait for patients with ataxia was not supported.

In addition a case study reported that a 5lb weighted vest, used as part of a general rehabilitation programme for a woman with severe cerebellar ataxia, was a useful adjunct when the patient was carrying out reaching activities in sitting. It is unclear how much the weighted vest contributed to overall improvements, therefore similar use would need to be evaluated on an individual basis.

**Summary**

- A very limited theoretical basis for axial weighting and no evidence to support use of axial weights to improve gait in people with ataxia

**e) Lycra garments**

**Adults**

A pilot proof-of-principle single case experimental design (n=6) was undertaken at the University of East Anglia and led by Dr Martin Watson. A six-week base line period was followed by a six-week intervention phase using custom-made lycra shorts, worn for between eight to ten hours a day. Repeated measures of postural sway, walking effort and speed were taken during all phases including a six week follow up. Results were mixed, some participants benefitted in certain aspects of everyday functional movements. Further studies to identify whether findings can be replicated and if the garments are more useful for some individuals than others are required. Full findings are yet to be published but a summary can be found at www.ataxia.org.uk. For further information contact M.Watson@uea.ac.uk.
Children

Three studies were reviewed that investigated the effect of lycra garments in children.\textsuperscript{112-114} Two studies had good methodological quality\textsuperscript{113,114} but lacked a control group and conclusions should therefore be viewed with caution. Likewise studies were small, different types of lycra garment were compared within and across studies with samples of heterogeneous participants. As such, results of these studies are highly variable across participants and beneficial effects were slight or of limited duration. In addition, two of these studies reported significant adverse effects.\textsuperscript{112,114}

Summary

- Insufficient data is available to support the use of lycra garments for children with ataxia

Specific interventions for upper-limb tremor

Lesions affecting the cerebellar hemispheres give rise to ipsilateral limb symptoms including tremor in addition to dysnergia, disdiadochokinesia and rebound phenomenon. An action tremor occurs during movement, ie it is produced by voluntary contraction of muscle, and includes postural tremor (occurs when voluntarily maintaining a position against gravity, eg holding an arm out straight) and kinetic tremor (occurs during any type of voluntary movement). Kinetic tremor is further subdivided into: simple kinetic tremor, which occurs during voluntary movements that are not target-directed (eg flexion/extension or pronation/supination), and intention tremor, which occurs during target directed, visually guided movements (eg finger-nose test), and worsens at the terminal phase of the movement as the target is approached.\textsuperscript{115}

In addition to affecting activities of daily living the psychosocial consequences of upper-limb tremor can be significant.\textsuperscript{116} The treatment of upper-limb tremor via the action of pharmacological agents and physiotherapy remains wanting. \textit{Also see section 4.1.}

a) Manipulation of visual information

A clinical observation by Pope\textsuperscript{117} that closure of eyes whilst eating may assist in the control of upper-limb ataxia has some support from experimental evidence. These studies with the exception of one\textsuperscript{118} were conducted with people who had intention tremor as a result of MS. Results suggest that kinetic tremor improves if movement is not visually guided\textsuperscript{118-120} and dysmetria improves if visual feedback is manipulated.\textsuperscript{121,122} Saccadic dysmetria was noted to frequently coexist with intention tremor and inaccurate eye movements are likely to impair accurate motor performance of the hand\textsuperscript{123} and individuals with intention tremor or other cerebellar deficits had difficulty using visual information to control arm and hand movements.\textsuperscript{123}

Summary

- Tremor amplitude may be reduced if target directed movements are performed from memory rather than under direct visual guidance\textsuperscript{119} or if the primary saccade and the hand movement to reach the object are performed separately\textsuperscript{121}
b) Cold therapy
Two studies\textsuperscript{120,124} reported functionally significant reductions in upper-limb tremor following cooling of the upper-limb in people with MS. Although both studies report improvements, there were differences in effect which might be related to the duration of cooling; 15 minutes compared to one minute. Several mechanisms have been suggested that may contribute to the reported effects; a temperature-dependent decrease in muscle spindle sensitivity causing a reduction in Ia afferent discharge and thus a reduction in response of the long latency stretch reflex\textsuperscript{120,124} and a decrease in nerve conduction velocity with an increase in stiffness of cooled muscles.\textsuperscript{124}

\begin{center}
Summary
\begin{itemize}
  \item Transient tremor control using cooling could have important functional implications when performing discrete functional activities such as intermittent self-catheterisation, signing documents, working a PC and taking a meal\textsuperscript{124}
  \item Deep cooling may be more effective than moderate cooling in individuals with severe tremor. Upper-limb cooling in general may not be as useful for individuals who also have significant proximal tremor\textsuperscript{124}
  \item Further studies to assess the effects of cooling on functional tasks are warranted
\end{itemize}
\end{center}

c) Wrist weighting
Investigation of wrist weighting as an intervention to reduce upper-limb tremor stretches back several decades.\textsuperscript{116,119,123,125,126,127} There has also been one study that has used a potentially more sophisticated mechanical damping device which as yet lacks clinical utility.\textsuperscript{128} Findings from these studies are inconclusive partly due to methodological issues, inclusion of heterogeneous tremors in the same cohort, the use of differing weights and weighting systems plus various outcome measures without reports of reliability or validity. Beneficial,\textsuperscript{116,126-128} detrimental,\textsuperscript{125} and mixed\textsuperscript{119} effects were reported, along with findings of no difference except slowing of the transport phase of movement.\textsuperscript{123}

\begin{center}
Summary
\begin{itemize}
  \item Evidence in this area is equivocal; it seems weighted wrist cuffs (of different weights) and weighted cutlery may be useful for some individuals under specific circumstances and should be assessed on a case-by-case basis. Patient goals and perspectives should be considered when assessing the value of the intervention
  \item As some individuals show exaggerated tremor for a short time on removal of weights, it is suggested that specific functions such as eating or writing are targeted. The long-term effects are not known; clinical observation suggests some people accommodate to the weight, the risk of fatigue should also be considered
\end{itemize}
\end{center}
Wheelchair seating

Wheelchairs rank among the most important therapeutic devices used in rehabilitation and can make the difference between an active and efficient alignment and a postural catastrophe. Few studies have investigated the physiological and functional impact of postural supports such as specialist wheelchairs, which can present significant methodological challenges. This guidance document reviewed one paper of low methodological quality that included four participants with Friedreich’s ataxia. Findings were equivocal and further research is required.

Despite the lack of research studies, clinical observation suggests that power wheelchair mobility with appropriate postural support is an option to provide people with ataxia with a means of independent mobility. Power chairs may also help conserve energy that can then be used outside the wheelchair for carrying out activities of daily living in antigravity postures. Additionally an appropriate posture in the power chair may facilitate respiration and swallow in those patients who may be compromised in these areas. In the absence of other evidence, clinical experience and patients’ needs should be used to guide clinical reasoning.

Exercise

In general people with ataxia should be encouraged to exercise as part of health promotion and as long as risk factors and health and safety considerations have been assessed. Exercise should be tailored towards what appeals most to participants and may involve exploring several different options as well as building motivation and sustainability into the exercise prescription.

Note of caution: As cardiac abnormalities can occur in people with Friedreich’s ataxia, any exercise programmes should be discussed with the treating doctor. Please refer to the cardiac problems section in the medical guidance of this document (section 4.1).

a) Hydrotherapy and Swimming

No studies directly evaluate the efficacy of hydrotherapy for people with ataxia. However, anecdotal evidence supports the value of hydrotherapy for people with ataxia as a form of exercise. Cook advocates the use of hydrotherapy and swimming for people with ataxia because water activities offer risk and challenge, provide freedom of movement often not available on land and may be beneficial for speech. Hydrotherapy is also considered to offer beneficial effects on health related quality of life. Further studies are required to investigate assumptions concerning physiological and functional benefits.

b) General fitness training

Anecdotal evidence advocates the benefits of general fitness training, yoga and Pilates for people with ataxia to help maintain strength, flexibility and balance. Activities such as horse riding may also confer similar benefits. Psychosocial benefits have also been reported. No studies directly investigating Pilates or yoga or similar forms of exercise were identified in the literature review but further investigation is warranted. In a case study in a patient with Friedreich’s ataxia without cardiomyopathy, aerobic training was shown to have some benefits.
**Specific Impairments**

People with ataxia can experience a number of specific impairments which physiotherapists should be aware of. Clinical experience and feedback from people with ataxia indicates that fatigue can be a common and at times an overwhelming issue. Spasticity, contractures, and dystonia are also symptoms that can occur. Clinicians are referred to section 4.1 of this document and the MS Society Guidance for Physiotherapists (2008) for further direction about managing these symptoms. Bladder and bowel problems (such as frequency, urgency and incontinence) can also be a feature of the ataxias. For specialist advice and assessment referral to a gynaecologist or urologist may be required (see section 4.1). For further advice refer to the Association of Chartered Physiotherapists in Women's Health (ACPWH) who provide assessment and treatment for men and women with bladder and bowel impairment. A referral to a continence nurse may be useful. Finally, neuropathic pain can be a feature of the ataxias (see section 4.1).

*For information on palliative care see section 7.*

### 5.3 Occupational therapy

*This section has been reviewed by the College of Occupational Therapy, Specialist Section of Neurological Practice, long-term conditions forum and has been endorsed by the College of Occupational Therapists.*

**Introduction**

This section aims to provide occupational therapists (OTs) working with people with ataxia with information about how common impairments impact on typical activities and occupations and provide advice on interventions. In the absence of specific research, the philosophical approach, expert opinion and reference to other progressive neurological conditions research will be drawn on.

Occupational therapy (OT) is widely recognised to be a valuable part of the multi-disciplinary care team in ataxia although primary research evidence is limited. In addition OT is an important intervention for patients with progressive neurological conditions in maintaining independence and quality of life and to enable people to participate in self-care, work and leisure activities that they want or need to perform. When it is no longer possible to maintain usual activities, OTs support people in changing and adapting their relationship with their physical and social environment to develop new valued activities and roles.

The focus of OTs on ‘engagement’ in activity, rather than the disorder is important in progressive conditions and OT intervention should focus on goals that support the person and carers thereby adding to quality of life.

OTs should use assessment and outcome tools that measure the person’s satisfaction with the performance of an activity; use of tools that measure impairment would not demonstrate the effectiveness of OT intervention. Appropriate tools include, but are not limited to, AMPS (assessment of motor and process skills), GAS (goal attainment scale), COPM (Canadian Occupational Performance Measure), self-efficacy tools and quality of life measures.
It must be emphasised that the guidance given here is based on practice consensus, not research. Table 9 lists the general considerations for occupational therapy intervention.

**Table 9: General considerations for occupational therapy intervention**

<table>
<thead>
<tr>
<th>General considerations for intervention</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Gather as much background information as possible about the referral</td>
<td></td>
</tr>
<tr>
<td>Complete a full occupational performance history/interview</td>
<td></td>
</tr>
<tr>
<td>Prioritise occupational performance issues/areas of concern and prioritise areas of focus</td>
<td></td>
</tr>
<tr>
<td>Acknowledge and address the carers’ and family’s needs within the assessment process</td>
<td></td>
</tr>
<tr>
<td>Be mindful of the rate of disease progression and how this will impact on your intervention</td>
<td></td>
</tr>
<tr>
<td>Identify the impairments or skills that are of concern and consider how the environment impacts on performance through observing performance within every day tasks</td>
<td></td>
</tr>
<tr>
<td>Identify the individual’s strengths and their resources</td>
<td></td>
</tr>
<tr>
<td>Establish a list of main concerns and prioritise treatment goals</td>
<td></td>
</tr>
<tr>
<td>Decide the approach to your intervention with the person, ie adaptation, rehabilitation, compensation, education, sign-posting or a combination of these</td>
<td></td>
</tr>
<tr>
<td>Implement intervention through performance of activities or environmental adaptations</td>
<td></td>
</tr>
<tr>
<td>Evaluate your outcomes and re-evaluate a need for further input</td>
<td></td>
</tr>
<tr>
<td>Consider the need for future assessment when function changes and how that person can re-access yours or other appropriate services</td>
<td></td>
</tr>
</tbody>
</table>

**Common issues encountered with Activity and Participation**

The effects on a person’s occupational performance are not predictable and will depend on the types of activities that the person needs and wants to participate in. It is important to take a person-centered approach to analysing the problem areas with performance of daily activities and roles.

Evidence suggests that people with ataxia may have a lower quality of life in the early and end stages of the condition. It is therefore important to recognise that even at the early stage of the condition difficulties with roles and activity engagement may benefit from support. As most ataxias are progressive an important consideration is proactive planning for future needs. This can be a difficult situation to deal with in a sensitive manner and OTs must respect the individuals in their own journey of acceptance of this condition. If appropriate, OTs should broach the expectation of decline in the future when considering any major adaptations.

**Common interventions**

Common interventions and practical advice collected from clinical experience are outlined on the following pages.
Self care and toileting

Aims of treatment include minimising the impact of excessive movement and helping the person to optimise their independence where possible. Prioritisation of tasks may mean that the person may be happy to accept assistance with dressing if it allows conservation of energy that could be used for other, higher priority activities such as leisure or work. Don’t assume, ask the person, ensuring together you identify priority areas. Consider if independent dressing is a priority for that person.

Importantly, toileting is often an area that people with ataxia report as difficult and stressful. Rails can help the person to fix their arms and provide greater stability during transfer. Other problems encountered include dressing and undressing to toilet and perineal hygiene. Remember that assessment should be completed and aids trialled as each person is unique.

It is important to anticipate for the future. For example, early referral for level access showers may be appropriate for the person with progressive ataxia.

Practical suggestions

- Encourage sitting to bath or shower and consider providing seating with support for the back and arms
- Use of thermo-regulation devices on taps can be an important safety consideration if the person has difficulty with using taps
- Lever taps may be easier to use than standard taps
- Level access shower can be a useful consideration for a person with ataxia if bath transfers become unsafe or dangerous
- Zippers may be overcome via the use of ‘zip pulls’ and fastenings could be replaced with Velcro instead
- Button hooks can also be helpful
- An add-on bidet or an automatic washing/drying toilet such as a Closo-mat® (www.clos-o-mat.com) or similar device may help
- Toilet rails around the toilet may be of benefit and wherever possible these should be fixed to minimise risk of accidents
- Consider the height of the toilet seat and adapt this where required
- Consider the use of hygiene wipes when away from home
- Alcohol gel can help with hand hygiene when away from home
- Register with RADAR for key access to their public toilets

Eating and drinking

Feeding needs to be considered due to multiple impairments impacting on safe and effective eating and drinking. Before commencing any feeding assessment, standard practice would be to ensure that the need for a speech and language therapy assessment is considered (see section 5.1). Joint working may therefore be appropriate. Feeding solutions may be different depending on contextual factors and solutions for eating at home may be different for social events. Altering positioning and/or seating will maximise posture and support core stability, thus reducing the impact of excessive limb movement.
**Practical suggestions**

- Organise work spaces and utensils to reduce clutter and optimise performance
- Plate guards, and Dycem® non-slip matting can be used as a placemat to limit movement of plate/cup (see www.dycem.com)
- Use lidded/insulated cups or cups with straws for drinking, especially hot liquids such as tea and coffee
- Cups with anti-tremor insert devices can help
- Use of the Neater-eater® device, with its dampening hydraulic mechanism can be very effective in aiding independent spoon or fork feeding (www.neater.co.uk)

**Food preparation**

Preparing food is one of the common concerns in the early stages due to obvious risks. OTs should carry out an activity analysis of food preparation tasks and suggest a variety of methods and aids that may compensate for difficult or unsafe aspects of tasks. This may include completing food preparation tasks in a seated position, having someone else do aspects of the tasks for them, (but not the whole task) such as cutting hard vegetables, or use of devices to aid grip and maximise safety. Again, find out what is important to the person and offer individual assessment of these areas.

**Practical suggestions**

- Kettle tipper devices can help making drinks safer
- Using a travel mug with a lid can sometimes assist with carrying a drink
- Waist height ovens; use of full-length oven gloves; sliding food to a level surface (or level trolley) rather than lifting
- A microwave oven can provide a safer alternative to standard ovens
- Chopping boards with an attached cutting blade can be safer than a separate knife
- A food processor can help with slicing or chopping vegetables

**Household management**

It is important to identify areas of household management important to the person and to recognise the cognitive and physical elements to these tasks. Most people with ataxia will continue to be able to cognitively manage the home but may have difficulty in physically doing heavy housework such as vacuuming or heavy laundry. OTs may wish to discuss the impact of fatigue in order to help balance continued involvement in activities whilst recognising what is a priority to them.

**Practical suggestions**

- Ensure that people have all the benefits that they are entitled to such as DLA (Disability Living Allowance), which can manage the extra costs of living with a progressive ataxia (eg hiring of a home help to assist with heavy household tasks)
**Bed, chair and toilet transfers**

Ensure that the height of the chair is correct for the person to transfer easily on and off. Ensure that the hip and knee angle is at 90 degrees and that the feet are flat on the ground. Armrests greatly enhance the ease of chair transfers and this should be considered when using high-backed dining chairs with arms. Ensure the chair is stable and that armrests are at a suitable height and position to enable the patient to push up.

Educate the patient and carer on sit to stand techniques. Importantly, consider for the carer’s safety.

Some people will need hoist provision for transfers and OTs should ensure that they are adequately trained to perform this and that it is undertaken by a relevant team member. In particular, where full body tremor presents, slings need to provide the most support possible for safety reasons.

<table>
<thead>
<tr>
<th>Practical suggestions</th>
</tr>
</thead>
<tbody>
<tr>
<td>Consider the height of the bed and location within the room</td>
</tr>
<tr>
<td>A bed lever can be beneficial to aid rolling and rising in bed</td>
</tr>
<tr>
<td>Mattress variators, or profiling beds may be of benefit</td>
</tr>
<tr>
<td>Ensure surfaces are the optimal height to ensure the most efficient and safest transfers</td>
</tr>
<tr>
<td>Firmer mattresses will aid bed mobility</td>
</tr>
<tr>
<td>Pressure care needs should be considered if mobility is severely restricted</td>
</tr>
</tbody>
</table>

**Indoor Mobility**

Indoor mobility should be ideally assessed in the environments that the person uses. For example, a person with progressive ataxia may walk well at home using walls and rails but be unable to walk independently in a hospital, work or community setting. Mobility should be assessed jointly with the physiotherapist. OTs should be considering the interaction of the person with their environment and the tasks that the person wants to perform once they have walked/mobilised somewhere. Consider what and how the person plans to carry or transport items while walking. OTs should consider the use of walking aids in the home and other environments. Use of walking frames may need to be reconsidered in very small areas. A combination of devices may need to be considered to move from one area to another, eg a walking frame to the toilet and then the use of hand rails inside the toilet.

When wheelchairs are required, close liaison with the local wheelchair service is recommended (see also section on outdoor mobility). Consider the environment including door widths, issues of access and interaction with furniture/tables to ensure that the person can still access areas they want/need to.

Major home modifications may be required, if not in the first instance, as the condition progresses, and this should be considered earlier, rather than later, with sensitive respect given to the person’s psychological adjustment process. Some people may choose what is considered an unconventional solution which helps them navigate their home such as crawling. A compromise between safety and risk management and patient choice may be required.
Practical suggestions

- Bags worn close to the body may be the most efficient and cause the least impact on balance. Later, it is advisable to avoid carrying items while walking.
- Trolleys may help to transport items, especially food, drinks and heavy items at work or in the home and should be discussed and assessed if thought beneficial.
- One-handed trays such as the Handitray® can help transportation of items.
- Advise removal of items such as scatter rugs and loose electrical cables that may present as risks to mobility in the home environment.
- Good lighting will help optimise performance of tasks and ensure that potential hazards in the home are avoided.

Falls management

Falls may occur in any area that a person mobilises. At home, the person should be taught fall recovery techniques and where appropriate, consider the use of community care alarms such as pendant alarms, and techniques to avoid further injury such as pressure sores while waiting for help to arrive.

OTs should consider joint assessment with or referral to a physiotherapist, and referral to a falls programme/group locally. If there is a family member or carer involved, the occupational therapist and physiotherapist should consider the safety of the carer.

Outdoor and community mobility

Mobilising outdoors can often present particular difficulty for the person with ataxia, as it may be an unfamiliar environment. Educate the carer and the person with ataxia about resting regularly whilst walking outdoors. Consider what and how the person plans to carry while walking (see indoor mobility above). A wheelchair for outdoor use can help to reduce fatigue or maximise safety.

An assessment for optimal seating position can be useful even in the early stages and is essential in the later stages. OTs should consider a referral to local wheelchair services for expert assessment. Consideration should be given to stable cushions and back supports, as canvas backs/seats in standard wheelchairs do not encourage good posture which may impact on function. A compromise between optimising function and providing adequate support is important.

Practical suggestions

- Shop-mobility, taxi card schemes, mobility buses, dial a ride services can be helpful.
- Public transport and rail providers offer subsidised fares and can provide a meet and greet service/access assistance for customers.
- Outdoors motorised scooters or wheelchairs can maximise independence.

Handwriting

This can be an area of particular difficulty for someone with progressive ataxia. If the person is still at school or at university, it is important to work within the provisions of special educational supports such as those provided through support workers. For someone at work, consider a referral to Access to work (AtW) for a full assessment. This may include an Ability-net assessment for suggestions of alternative technological solutions for handwriting problems. Activity analysis may reveal the need for adaptations such as alternative positioning and/or seating, desks and different pens.
**Practical suggestions**

- Ensure the work space and seating are set to maximise support and optimise posture advisable to avoid carrying items while walking
- Dictaphones or voice-activated computer software can be used to compensate for problems with handwriting discussed and assessed if thought beneficial
- Use of weighted pens and thick barrelled pens may help but there is limited supporting evidence
- Consider the type of pen nib and the pressure applied as some people experience fatigue affecting sustained pen grip

**Computer use**

There are many aids to compensate for ataxia when using a computer. Again, this assessment may require joint assessment with a speech and language therapist in considering whether voice-activated software may be appropriate for overcoming problems with using a keyboard to enter information; however, dysarthria may prevent voice-activated software from being useful. Importantly, Information Technology is a constantly changing area with new devices and solutions becoming available all the time. If the person is still at work, funding for this assessment should be gained thorough AtW referral. Alternatively, OTs should investigate charitable organisations that may provide funding to access these services and equipment.

**Practical suggestions**

- A referral to IT solutions experts (such as Ability-net) is strongly advised
- Ability-net website has free advice about IT adaptations for people with ataxia ([www.abilitynet.org.uk](http://www.abilitynet.org.uk))
- Keyboard and mouse modifications can be made to adjust the sensitivity and speed of response
- Alternative mouse such as a Tracker ball can be helpful
- Smaller keyboards or keyguards may help
- Consider the layout and location of equipment for ease of access

**Work**

OTs should consider maintenance of the working role for as long as the person wants it to continue and for as long as that is possible.

It is important to provide education on their rights and responsibilities under the 1995 & 2005 Disability Discrimination Act. Support the individual regarding the disclosure of their diagnosis to others and their employers, if this is a concern.

It should be remembered that the intervention should allow them to develop skills to manage the employer themselves where possible. Where required a work site visit or referral to AtW will ensure a worksite visit is undertaken if appropriate.
OTs may directly intervene by assessing and advising on reasonable adjustments which may include changing work hours, environmental adjustments or assistance with specific tasks or travel which may be supported financially by AtW (for information on AtW see www.jobcentreplus.gov.uk). AtW will help considerably with costs of other aids required such as motorised wheelchairs, ergonomically appropriate seating and desks, and IT devices. When work is no longer possible consider rebalancing the loss of the working role with other activities or help to access relevant benefits.

Driving
If they are a driver, the newly diagnosed person with progressive ataxia is legally obliged to inform the DVLA and their insurance company of their diagnosis as soon as it is confirmed. Reporting the diagnosis may not mean cessation of driving. The DVLA will request information from the person and their medical team and may request attendance at a driving assessment centre before making a decision.

Some people will require driving adaptations to allow safe driving to be completed (see www.direct.gov.uk/en/DisabledPeople/MotoringAndTransport). Specialist centres provide assessment for suitable adaptations as well as driving ability (www.mobility-centres.org.uk). Where appropriate, the Motability scheme can assist people who receive the higher mobility rate of DLA with minor adjustments, lessons or funding a vehicle (www.motability.co.uk).

There are some cases when the condition causes such difficulty with driving that it is unsafe for the person to continue with this role. (www.direct.gov.uk/en/DisabledPeople/MotoringAndTransport). When this is true, OTs should explore alternative community mobility (see section above).

Practical suggestions

- Educate the person and carer on allowing the car door to be opened fully and to consider the height of the transfer being undertaken
- Ensure the person sits their bottom down first before moving their legs into the car
- Try inserting a swivel transfer mat and if the car seat is particularly low a firm foam cushion or blanket in a pillowcase
- Choose a model of car that optimises transfers, door access and storage space

Leisure
OTs should bear in mind that if there is loss of other occupational roles, leisure may be an area that can help to re-address this loss in a different capacity. If the person has lost leisure roles such as participation in sport, consider that they may continue involvement in the activity such as score/record keeping, participation on committees and social participation in their local club. For hobbies such as horticulture, there may be adaptations that can be made to maintain participation, or aspects of tasks can be performed such as visiting gardens locally, planning planting or weeding of raised beds.

Enjoying leisure time with one’s family can be continued albeit in modified ways, such as the use of accessible holiday homes, use of a wheelchair when visiting outdoor areas such as parks and galleries, and ensuring social contact continues in the home or other spaces. Reading can present particular difficulty due to difficulty holding a book or visual problems.
Practical suggestions

- Bookstands can be obtained to hold the book
- Use of elastic bands around the loose pages of books can limit the frustration caused by rustling pages where tremor exists
- Use of a rubber thimble can be useful to help turn pages where fine motor coordination is a problem
- Books may be downloaded online and use of text enlarging buttons can help where vision is a problem
- Talking books are also available if preferred. The RNIB can be a useful support service in this area (see www.rnib.org.uk)
- Electronic page-turners can be purchased but are costly and take up space

Control of the indoor environment

When communication difficulties present, ensure speech and language therapy involvement and assess jointly if available. Telephone use can present particular difficulty in pressing small buttons where tremor and reduced coordination present. Use of electrical equipment may be difficult if handling of small switches is necessary. As the condition progresses OTs should consider referral for regional environmental control unit assessment for optimising control of the indoor environment.

Consider priority activities for the person to participate in; remember that especially in the palliative stage the focus on meaningful activity can provide immense satisfaction and comfort to the person with ataxia and the family.

Practical suggestions

- Consider use of ‘big buttons’ telephones and phones with autodial numbers or voice activation
- Consider use of a telephone with two way record to save conversation for replay later to help keep messages
- Telephone providers such as BT have an inclusion phone services policy including a communications solutions guide obtainable online – see www.btplc.com/inclusion/Needhelp/
- Light switches should be simple and easily reached from a standing or wheelchair position suitable to the person
- Appliance sockets are safest when located off the ground at waist level to avoid complex bending, squatting and reaching
Advice on specialised equipment
Organisations such as the Disabled Living Foundation (www.dlf.org.uk, Tel: 0845 130 9177) or Remap (www.remap.org.uk) provide information and advice on equipment for disabled people and can be a useful resource for OTs.

Conclusion
Despite the limited primary evidence of specific OT intervention, expert opinion highlights involvement of OTs in the multi-disciplinary management of people with progressive ataxias. The above examples provide a guide to suggested OT intervention in this group based on the consensus of OTs working in this area. Future research is recommended into OT intervention, within the context of a multi-disciplinary team, for people with progressive ataxia.
6 Research

There have been a number of developments in the last few years including the identification of new genes causing specific ataxias, pre-clinical studies of potential disease-modifying drugs and clinical trials. Another major thrust of research has been studying the basic biological mechanisms underlying the ataxias.

Research has also focused on the development of tools to measure the severity and progression of ataxia for use in trials such as validated ataxia-specific rating scales (detailed in Table 10, below).

Table 10: Ataxia rating scales

<table>
<thead>
<tr>
<th>Rating Scale</th>
<th>Ataxia Type</th>
</tr>
</thead>
<tbody>
<tr>
<td>International cooperative ataxia rating scale</td>
<td>All ataxias</td>
</tr>
<tr>
<td>(ICARS)(^1)</td>
<td></td>
</tr>
<tr>
<td>Scale for the assessment and rating of ataxia</td>
<td>Spinocerebellar ataxias</td>
</tr>
<tr>
<td>(SARA)(^2)</td>
<td></td>
</tr>
<tr>
<td>Friedreich's ataxia rating scale (FARS)(^3)</td>
<td>Friedreich's ataxia</td>
</tr>
<tr>
<td>Friedreich's ataxia impact scale(^4) (FAIS)</td>
<td>Friedreich's ataxia</td>
</tr>
</tbody>
</table>

It is good clinical practice to offer patients the opportunity to take part in research projects. The charity Ataxia UK provides up-to-date information for patients and healthcare professionals on developments in the ataxia field, including opportunities for patients to take part in research. For example, genetic testing is offered on a research basis for a number of genes causing ataxia eg recessively inherited ataxias at the Oxford Ataxia Centre (contact Ataxia UK for details). Healthcare professionals are encouraged to join Ataxia UK’s Medical Registry and receive regular electronic newsletters with information on any trials recruiting participants. Information on ataxia conferences and research developments is also provided. (Register online at: www.ataxia.org.uk/profreg.php).

Ataxia UK also provides funding for research projects and facilitates research (eg by organising ataxia conferences/meetings, helping to recruit participants in research projects etc) and is willing to hear about any new project ideas (contact research@ataxia.org.uk).

For more information on research developments and taking part in research projects contact Ataxia UK (www.ataxia.org.uk).
Palliative care is an approach that improves the quality of life of patients and their families facing the problems associated with a life-threatening illness such as the progressive ataxias described in this guidance. The aim of palliative care is the prevention and relief of suffering by means of early identification, impeccable assessment and treatment of distressing symptoms and other problems; physical, psychosocial and spiritual* (* World Health Organisation definition 2002). Although the term palliative care is often used to mean ‘end of life care’, palliative care is often very appropriate and applicable earlier in the course of an illness, in conjunction with other therapies that are intended to prolong life. In this context, palliative care will enhance the patient’s quality of life and offer a support system which helps the patient to live as actively as possible until death, thus affirming life while regarding dying as a normal process.

Palliative Care is a generic term which, in describing an approach to care, highlights a responsibility carried by all health care professionals to provide holistic care for their patients with progressive neurological disease. Most palliative care for patients with progressive ataxias will be provided in the community by district nurses, GPs and other members of the primary care team. It is the responsibility of community and hospital multidisciplinary teams caring for patients with progressive ataxia to assess whether the patient has specialist palliative care needs and would benefit from involvement of a specialist team. Specialist needs are usually defined as complex issues arising as a result of the patient’s illness that cannot be managed by the frontline caring team. For example, the patient’s GP may involve the community specialist palliative care team to help manage painful muscle spasms or a neurologist may seek support for patients struggling psychologically with the prospect of progressive neurological disability. Specialists in palliative care work in the community, in hospitals and in independent hospices. Patients with specialist palliative care needs can thus be supported at home, during hospital admissions and within a hospice environment as a day patient or inpatient according to their specific needs.

In recent years increasing emphasis has been placed on helping patients think about and plan for the end of their lives. The Department of Health’s recent launch of the End of Life Care Strategy,\(^\text{154}\) which covers England, aims to improve services to provide adults approaching the end of life with more choice about where they would like to be cared for and die. It encompasses all adults with advanced, progressive illness and care given in all settings. The National Service Framework for long-term conditions also highlights the importance of providing a range of palliative care services specifically to people with long-term neurological conditions such as the progressive ataxias.\(^\text{7}\)

For this approach to be effective, the professionals caring for patients with progressive ataxia need to feel confident to broach the subject of end of life care with patients and their families. This area needs sensitive handling and skilled communication. It also requires anticipation of future potential issues and a willingness to deal with these difficult areas. Primary and Secondary care, Specialist Palliative Care, Neurology and Rehabilitation services need to establish close working relationships to provide coordinated, patient-centred care for patients faced with progressive disability and life shortening illness. In this context, patients and families will receive the support they need as the disorder progresses.
Appendix

A list of neurologists and clinical geneticists at Ataxia UK Accredited Ataxia Centres of Excellence and Other Centres of Expertise*

Adult neurologists (and clinical geneticists where indicated)

**Ataxia Centres**

**Dr Paola Giunti and Professor Nicholas Wood**
Ataxia UK Accredited Ataxia Centre
National Hospital for Neurology & Neurosurgery
London WC1 N3BG

**Dr Marios Hadjivassiliou**
Ataxia UK Accredited Ataxia Centre
Sheffield Teaching Hospital NHS Foundation Trust,
Sheffield S10 2JF

**Other Centres**

**Professor Patrick Chinnery**
Department of Neurology
University of Newcastle
Newcastle upon Tyne NE2 4HH

**Dr Rajith de Silva**
Queen’s Hospital
Romford
Essex RM7 OBE

**Dr Nick Fletcher**
The Walton Centre for Neurology and Neurosurgery
NHS Trust
Liverpool L9 7LJ

**Dr Simon Hammans**
St Richard’s Hospital
Chichester
West Sussex PO19 6SE

**Dr Paul Hart**
St Helier Hospital
Carshalton
Surrey SM5 1AA

**Dr Kevin Talbot/Dr Zameel Cader and Dr Andrea Nemeth (Clinical Geneticist)**
– joint ataxia clinic
Ataxia UK Accredited Ataxia Centre
John Radcliffe Hospital
Oxford OX3 9DU

**Professor Patrick Morrison (Clinical Geneticist)**
Dr Ailsa Fulton / Dr Gavin McDonnell
Belfast City Hospital Trust
Belfast BT9 7AB

**Dr Neil Robertson**
Department of Neurology
University Hospital Wales
Cardiff CF14 4XN

**Professor Anthony Schapira**
Royal Free and University College Medical School
London NW3 2PF

**Dr Alastair Wilkins**
Department of Neurology
Frenchay Hospital
Bristol BS16 1LE

**Dr Paul Worth**
Norfolk and Norwich Hospital
Norwich NR4 7UY

**Paediatric neurologist and paediatric clinical geneticist**

**Dr Peter Baxter**
Sheffield Children's NHS Foundation Trust Sheffield S10 2TH

**Dr Andrea Nemeth (see details above)**

* Please note that this is a list of specialists known to Ataxia UK and to the Guideline Development Group and is not an exhaustive list. We would welcome contact from other neurologists with expertise in ataxia (email Ataxia UK research@ataxia.org.uk)


115. Deuchsl et al. and an Ad Hoc Scientific Committee Consensus statement of the movement disorder society on tremor. Movement Disorders, 13, suppl 3, 2-23.


126. Deuchsl et al. and an Ad Hoc Scientific Committee Consensus statement of the movement disorder society on tremor. Movement Disorders, 13, suppl 3, 2-23.


147. Cano et al. Friedreich's ataxia impact scale: A new measure striving to provide the flexibility required by today's studies. Mov Dis. 2009 Feb 17, [Epub ahead of print]

148. The End of Life Care Strategy can be viewed on the DOH website www.dh.gov.uk