

# Multidisciplinary Management of Progressive Ataxia

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She is now Professorial Research Associate in the Department of Clinical Movement Neurosciences at UCL Queen Square Institute of Neurology and Honorary Consultant at the National Hospital for Neurology and Neurosurgery.

She established the first Ataxia Centre at the NHNN in 2005, a robust model of translational clinical service. This has been accredited as the first centre of clinical excellence in the UK by Ataxia UK and more recently by the National Ataxia Foundation, charities of ataxia patients in the UK and USA respectively. On this model, two other centres have been established in the UK. In this setting she has established the first Ataxia Multidisciplinary clinic that sees a Neurologist specialised in ataxia together with a speech and language, occupational and physio therapists.

## Abstract

The National Hospital for Neurology and Neurosurgery (NHNN) hosts a specialist centre for people living with Progressive Ataxias. Patients attend from across the UK for differential diagnosis, specialist opinion, expert assessment and exploration of treatment options. This article discusses the treatment and management considerations of the multi-disciplinary team in the context of a condition with a huge variety of symptoms and limited evidence base for clinically effective treatment. It highlights the importance of tailored treatment focused on symptom management and optimising participation. It also discusses service initiatives that enabled patients continued access to treatment during the COVID-19 pandemic. These initiatives were proven to be very successful and therefore continue to run today.

## Key Points

- The management of progressive ataxias requires an MDT approach to capture all facets of this complex disease.
- Best treatment approach requires that therapy is tailored to each individual's needs from each MDT member.
- Allied health professionals play a vital role in empowerment and education of people with progressive ataxias to effectively manage symptoms and optimise function.
- Group therapy is a novel approach to treating people with progressive ataxia that requires further research.



## Introduction

Progressive ataxias are rare and complex disorders affecting an estimated 10,000 adults in the UK [1]. Diagnosing progressive ataxia is challenging due to the high number of genetic causes and is often based on clinical presentation. The Global 100,000 Genome Study has isolated and identified the causal genes, identifying over 40 varieties of spinocerebellar ataxias [11]. This has improved the knowledge of how the differing types might progress. In some ataxias, the neuropathology is purely cerebellar, however, extrapyramidal, spinocerebellar tract, dorsal column, vestibular and peripheral nerve involvement can be present [12,5]. The main clinical features of progressive ataxias are gait and limb ataxia, imbalance, gaze-evoked nystagmus, intention tremor, dysphagia and dysarthria [3]. The progressive nature of ataxia often results in loss of function in daily activities, independence and quality of life [3,4]. Although there is better understanding of the progression of the symptoms of ataxia,

there is little high-quality evidence-based-practice to guide intervention [13]. Most evidence is based on specific genetic phenotypes, highlighting the challenges of generalising treatments to all ataxias [14].

## Treatment and Management

There are research trials investigating pharmacological and gene therapies for ataxia [7], however proven curative treatment or disease-modification is absent. Primary treatment of progressive ataxia is based on symptom management and maintenance of function. The large variation in symptom presentation makes the management of ataxia challenging. Consensus guidelines for the management and treatment of progressive ataxias was developed by an expert group of clinicians in 2016 [1,4].

At NHNN, patients with ataxias are examined in the MDT ataxia clinic comprising of a Consultant Neurologist expert in ataxia, Physiotherapist, Occupational Therapist (OT) and Speech

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and Language Therapist (SLT) to identify their specific and complex ataxia needs and to ensure that a holistic approach is taken. The specificity of this clinic means the history of the patient's progression, in keeping with the stage of their disease, is considered in the context of their treatment and intervention. For example, people who have a diagnosis of Friedrich's Ataxia commonly have reduced muscle mass due to peripheral neuropathy and therefore intervention for foot and hand function is vital. The aim of the clinic is to identify the person's main daily challenges, provide education and advice, discuss medical and pharmacological management and signposting to relevant local services.

#### Allied Health Intervention

Beyond diagnosis, understanding of management options amongst health care professionals is lacking, and as such, patients face enormous challenges in both understanding their illness and obtaining appropriate treatment. Interventions by allied health professionals play a crucial role in the management of people with progressive ataxias [4] due to the complexity and change in symptoms over time. Long-term collaboration, conveyance of hope and supporting day-to-day improves self-identity, self-esteem and sense of control of symptoms [12].

Interventions should be individually tailored to support: specific goals related to activity and participation; integration of restorative and compensatory strategies; targeted impairment based therapy to optimise and preserve function and prevent secondary complications [12]. People with ataxia have overlapping symptoms that affect their functioning. For example, they often experience fatigue which impacts on their balance and speech production. Working as an MDT to optimise symptoms and not in isolation is important for holistic care.

#### Physiotherapy

There is emerging evidence to support positive outcomes for patients with progressive ataxias through physiotherapy intervention [13] including: falls prevention; optimisation of mobility and function; improved quality of life (QOL); reversal of deconditioning [2].

An in-depth assessment covering balance, gait, coordination, core stability, strength, endurance, vestibular function, muscle

length, spasticity and posture is required [16]. Interventions might include:

- Provision of individualised HEPs targeting ataxic impairments alongside cardiovascular fitness and global strength.
- Dynamic task practice targeting core stability with a focus on minimising upper-limb weight bearing to improve engagement within daily tasks, balance and gait.
- Education around optimal activity and exercise tailored to each individual and their lifestyle.
- Exploring meaningful and enjoyable physical activity to maximise functional outcomes and active participation in the longer-term management of their condition.
- Signposting to local services such as: exercise on prescription schemes; Ataxia UK and utilisation of smartphone applications.
- Exploring barriers to physical activity and exercise participation. Often individuals express lack of confidence due to balance or motivation to attend local/virtual activities and/or classes that would be appropriate for someone with ataxia.
- Compensatory approach with consideration of exploring the use of orthotics or devices to maximise independence.

#### Occupational Therapy

Using skills of task analysis, an OT is able to break down individual tasks into its component parts to identify how the person's ataxia is impacting on performance. This informs education, coaching of alternative techniques to change the approach to the activity or selection of adaptive aids. It is vital that OTs focus on activities that are important and meaningful to the person with ataxia. Measurement of satisfaction is used to demonstrate effectiveness of occupational therapy intervention in people with progressive ataxias [6,8]. For example, initial intervention for feeding incorporates coaching of proximal stabilisation techniques and trial of weighted cutlery, although the evidence for weighted tools is low. As the condition progresses, the person might become dissatisfied with their performance when feeding and rely on someone else to feed. The introduction of assistive devices, such as the manual neater eater, can help to maintain independence and reduce reliance on others, with subsequent improvement in satisfaction levels. Psychological adjustment

to a person's increasing disability needs careful consideration throughout the person's treatment. Interventions might include:

- Fatigue management education: balance of rest and activity; addressing poor quality sleep, use of relaxation techniques
- Task adaptation and techniques focusing on optimising postural stability and promoting normal movement to increase the chances of success
- Assessment of hand function and intervention to optimise function and maintain range of movement
- Equipment provision including assistive technologies
- Addressing environmental barriers to participation.

#### Speech and Language Therapy

Like many other disciplines there is currently limited evidence for clinically effective treatments for SLTs in ataxia. Patients living with ataxia may experience changes to their speech and voice [10]. Evidence is growing for exercises targeting control of volume, tremor in the voice, pitch fluctuations and slurring of speech [10]. This impacts a person's ability to participate in meaningful conversations and engage in vocational and leisure activities. Dysphagia (swallowing difficulties) is also a common symptom of ataxia [17]. Difficulties may include difficulties in chewing food and coordinating movements for swallowing which can result in a feeling of food sticking in the throat, as well as coughing and spluttering when eating and drinking. This can lead to embarrassment, discomfort, compromise of chest health (due to food/drink entering the airway) and withdrawal from social situations [15].

The role of the SLT is to educate patients on how ataxia impacts speech, voice and swallowing ability. An in-depth assessment of the speech, voice and swallowing systems is completed. Interventions may include:

- Strategies for safe swallowing such as safe positioning for mealtimes
- Use of adaptive equipment including rate-controlled cups
- Modification of diet or fluids
- Strategies for clear speech
- Exercise-based intervention to improve and maintain speech and voice output.

### Initiatives during COVID-19 Pandemic

During the pandemic there were longer waiting lists and an increase in referrals for all disciplines at NHNN for patients living with ataxia. We hypothesise that this was secondary to national lockdowns affecting the ability of community services to access patients; self-isolating and shielding staff, temporary cessation of 'non-urgent' services and redeployment of staff [9].

Initiatives were undertaken at NHNN to ensure patients were still able to access treatment and intervention.

#### 1. Telehealth clinic

In March 2020 in the first wave of the COVID-19 pandemic, MDT clinics were converted to telehealth appointments. This proved to be a very effective way of managing this patient group, allowing more patients to access the service whilst being able to isolate at home. All patients that attended the telehealth MDT Ataxia Clinic from January-May 2021 were contacted. 16/30 responded and took part in a retrospective telephone survey. 13/16 (81%) patients reported an overall positive experience of which 11 (69%) would recommend to others. Benefits reported included reduced costs and carer burden associated with travel and ease of access at home. Some felt their needs may have been better met in person as they felt physical assessment might have been beneficial. This clinic continues to offer virtual consultations and face-to-face for appropriate patients.

#### 2. Exercise and education group

Reduced exercise tolerance and deconditioning became more commonly reported patient symptoms. Therefore, a virtual education and exercise group was created, comprising of 6-8

patients, which ran weekly for one-hour over a four-week period. Patients were initially assessed for the group in a 1:1 consultation and self-efficacy and activity levels were measured through patient reported outcome measures before and after the course. This data is currently being reviewed, however patients reported enjoyment of exercising in a group format and increased motivation to engage in sustained exercise activity outside of the group. The group continues to run on a quarterly basis.

#### 3. Motor speech group

To capture patients unable to access local communication services, a virtual group was set up to address the main speech and voice challenges faced by people with ataxia. This novel approach targeted the principles of 'smooth and steady' speech and voice production. The group comprised of 3-6 patients, seen for one-hourly, weekly sessions over six weeks. Each patient had a 1:1 initial virtual consultation where communication and QOL were measured. The initial feedback from three pilot groups was very positive, specifically; feeling more confident, enjoying access from their home, particularly as many lived outside of London, and feeling a sense of connectedness with others who have similar communication challenges. Outcome measures also demonstrated improved speech intelligibility as rated by unfamiliar listeners. Due to their success, and lack of availability of this type of service in the community, these groups continue to run at NHNN.

#### Evaluation of the outcomes of practice

● The above practices and the MDT ataxia clinic have seen positive outcomes in patients.

This is based on survey results and anecdotal evidence.

- Improved regular participation in exercises and activity has resulted in reduced re-referral rate to NHNN services.
- Improved understanding of each individual's condition and access to support networks.
- Improved mood.
- Confidence with the utilisation of self-management strategies to optimise and maintain function and increase independence.
- Empowerment of carers/family members to support people to follow tailored programmes provided by Allied Health Professionals.
- Sense of belonging when engaging in a group exercise environment.

#### Conclusion

People living with ataxia experience a plethora of symptoms. Despite the absence of disease-modifying treatments for most ataxias, many aspects of these disorders are treatable, and it is essential that healthcare professionals know how to optimise symptom management. Specialist MDT assessment and management as well as the development of individualised, tailored treatment approaches is therefore vital. Current recommendations on the management of these conditions are largely based on clinical experience and anecdotal evidence, hence the importance of further research. The Covid-19 pandemic has created opportunities of continued patient access to therapy intervention to maintain function, ensure safety and support psychosocial wellbeing. During this time NHNN therapies were able to offer novel approaches to intervention via telehealth which demonstrated positive patient outcomes.

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