

A14/S(HSS)/a

**2013/14 NHS STANDARD CONTRACT
FOR ATAXIA TELANGIECTASIA SERVICE (ADULT)**

SERVICE SPECIFICATIONS

Service Specification No.	A14/S(HSS)/a
Service	Ataxia telangiectasia service (Adult)
Commissioner Lead	
Provider Lead	
Period	12 months
Date of Review	

1. Population Needs

Ataxia-telangiectasia (AT) is a heterogeneous disorder with around 200 people in the UK affected. Patients with 'classical' AT present in childhood with a progressive, multi-organ, neurodegenerative disorder characterised by cerebellar ataxia, immunodeficiency and greatly increased risk of malignancy. Patients may present in adulthood with "variant" AT with milder, but similar, symptoms and a less aggressive disease course. All patients vary at presentation with individual combinations of neurological features.

1.1 National/local context and evidence base

AT is a rare disease with currently no treatment to cure or prevent the progressive nature of AT. However, through a holistic approach that is patient centred there is evidence for clinical effectiveness which includes increased probability of survival, better quality of life, improved body weight, identification of silent aspiration and improvement in lung function. Due to the rarity and complexity of AT, it is important for patients to be assessed by Papworth Adult AT Service (PAATS) to allow staff to further develop their expertise in this condition and to facilitate data collection to develop the evidence base.

2. Scope

Ataxia-Telangiectasia is a heterogeneous disorder. Patients with "classical" AT present in childhood with a progressive, multi-organ, neurodegenerative disorder characterised by cerebellar ataxia, immunodeficiency and greatly increased risk of malignancy.

Patients may present in adulthood with “variant” AT with milder, but similar, symptoms and a less aggressive disease course. All patients vary at presentation with individual combinations of neurological features.

2.1 Aims and objectives of service

Patients with Ataxia Telangiectasia (AT) have a mean life expectancy of twenty five years reflecting increased susceptibility to leukaemia, lymphoma, pulmonary infection, chronic lung disease and neurological decline including significant problems with swallowing. They experience severe ataxia limiting mobility and the ability to self-care. All of these elements of AT are potentially amenable to intervention to improve outcomes and quality of life.

The Papworth Adult AT Service (PAATS) will address the needs of two major groups of patients. Firstly patients who already have a diagnosis of AT and have been under the care of the Nottingham paediatric service. They require follow up, monitoring and intervention as new problems arise. The second group are those with a suspected diagnosis who require full diagnostic testing, assessment and management. AT is rare disease with around 200 people in the UK affected therefore the centralisation of expertise is essential in order for patients to receive care by staff who fully understand the disorder and who can support them through their life. Gathering a cohort of patients with a rare disorder together also affords an excellent opportunity to describe this rare condition in more detail and assess interventions in an evidenced based fashion.

Patients with AT experience respiratory, neurological and immunological difficulties as well as difficulties managing activities of daily. It is essential that their assessments cover all of these aspects of AT in order to provide optimal care.

The aims of this service are:

To provide a national service for the treatment of adults with AT.

- to work with Nottingham University Hospital NHS FT to ensure the smooth transition of patients from paediatric service
- to establish standards of care for adults with AT and to provide education nationally on the diagnosis and management of AT
- to work with stakeholders to undertake research to improve the understanding and treatment of this rare disease
- to work collaboratively both nationally and internationally with other providers of care for patients with AT to improve outcomes.

Objectives:

- offer a multi-disciplinary review for patients with AT on an annual basis which is individualised to their specific requirements
- diagnose, assess and treat patients diagnosed with AT in adulthood
- identify newly developing symptoms and disabilities, manage them appropriately and document outcomes

- develop detailed management protocols for enhanced local care
- liaise closely with primary and secondary care to optimise individual patient care
- improve the evidence base for the treatment of AT

Outcomes:

- maximise life expectancy and quality of life
- minimise hospital admissions and the requirement for unplanned medical intervention
- increase understanding of the disease through data collection, research and shared learning

2.2 Service description/care pathway

Patients are referred through the transition clinics held at Nottingham Hospital. The timing of these is dictated by both hospitals following discussion with the patients. Occasionally patients may come through another route such as through secondary care or through the AT Society.

Following the referral the patient is assessed by a multidisciplinary team made up of:

- respiratory physician
- immunologist
- neurologist
- physiotherapist
- occupational Therapist
- speech and language therapist
- dietician
- social worker.

Following their assessment and diagnostic tests, an MDT discussion occurs to ensure that all aspects of care are addressed to provide the best management plan. Where imaging needs to be discussed this will be through one of the existing thoracic multi-disciplinary meetings where the immunologist is present.

The patient, their GP and other secondary care providers are written to with the results from their assessment along with a management plan. This can then be followed up through a telephone consultation should the patient require further information.

This service provides clinical review and ongoing management advice for patients with AT or other rare AT like disorders with the occasional requirement for a new diagnosis. The service includes:

- multi-disciplinary annual review tailored to the needs of the patient
- ongoing advice through comprehensive feedback to local teams on management of each patient's condition.

Patients are predominately but not exclusively referred into the pathway through the

paediatric centre at Nottingham. Transition clinics will be held on an annual basis during which pre-selected patients in the age range 16 to 18 will be seen by both Nottingham and Papworth teams. During the assessment, a time of transition will be agreed with the patient as to when their care will be transferred from Nottingham to Papworth.

Patients will attend Papworth on an annual basis unless their local hospital or GP need input outside this visit.

Patients will be asked to complete a questionnaire prior to their attendance in order for their assessment to be tailored to their requirements.

As part of their multi-disciplinary assessment patients will also have a range of tests which may include the following:

New patients	Elective follow up patients
Clerking, nursing and medical assessment. Blood tests to assess for immunodeficiency	Clerking, nursing and medical assessment Blood tests to assess for immunodeficiency
Blood gases	Blood gases
Sleep study	Sleep study
Pulmonary function tests	Pulmonary function tests
Assessment by speech and language therapist, physiotherapist, occupational therapist, dietician, social worker	Assessment by speech and language therapist, physiotherapist, occupational therapist, dietician, social worker
Assessment by Consultant Respiratory Physician, Consultant Neurologist and Consultant Immunologist	Assessment by Consultant Respiratory Physician, Consultant Neurologist and Consultant Immunologist
Baseline CT	Not routinely indicated
Video Fluoroscopy	Video Fluoroscopy only repeated if the baseline test showed evidence of silent aspiration (around 40% of patients) or if develops symptoms
Magnetic Resonance Imaging (MRI) (brain imaging)	Not routinely indicated
Neurophysiology	Not routinely indicated

It is unlikely that Papworth will accommodate non elective referrals as these would be

dealt with by the patient's local provider, however in the event of this type of admission the follow up would be individualised as to the reason for admission.

Patients will generally stay under the care of Papworth for their whole lifetime. Referrals will be made to local services in addition to their care under Papworth as appropriate.

Days/Hours of operation

Patients and their carer are accommodated on the Respiratory Support and Sleep Centre Ward at Papworth Hospital, which is open 24/7.

Add extra text as appropriate here e.g. General paediatric care (+ Annex1), Pregnancy or Operational Delivery Networks

2.3 Population covered

Geographic coverage/boundaries

The AT service covers patients registered with an English or Scottish General Practitioner, resident in the European Union and eligible for treatment in the NHS under reciprocal arrangements. Patients from Wales and Northern Ireland are not part of this commissioned service the Trust must have separate funding arrangements.

2.4 Any acceptance and exclusion criteria

The service is commissioned by NHS England for all eligible patients from England and Scotland. The clinic can be accessed by any eligible patient who has been confirmed to have AT irrespective of gender, age, sex, disability, religious belief. Interpreters or use of a language line will be provided for families for whom English is not their first language.

The service is expected to demonstrate equitable geographical access across the country and take actions to address gaps in access.

The provider will provide information to patients on public transport access and accommodation for patients and relatives as needed.

Referral criteria, sources and routes

Referrals to the Papworth service are predominantly made through Nottingham's Paediatric AT Clinic or from other hospitals or the patient's GP.

In most patients the diagnosis of AT has been made by chromosome analysis or tests of chromosomal radio sensitivity performed by Professor Taylor's Laboratory at the University of Birmingham.

Referral criteria

- Patients with a confirmed diagnosis of AT (suggestive clinical features with increased chromosomal radio sensitivity and reduced expression of ATM or expression of ATM with no detectable kinase activity in lymphocytes or 2 pathogenic mutations in the *ATM* gene)
- Patients with probable AT (suggestive clinical features with raised AFP levels and increased chromosomal radio sensitivity or characteristic chromosomal rearrangements)
- Patients with a confirmed diagnosis of AT Like Disorder (ATLD) (suggestive clinical features with reduced expression of hMRE11 in lymphocytes or 2 pathogenic mutations in the *hMRE11* gene)
- Patients with a confirmed diagnosis of ataxia with oculomotor apraxia type 1 (AOA1) (suggestive clinical features with reduced expression of aprataxin in lymphocytes or 2 pathogenic mutations in the *APTX* gene)

Exclusion criteria

The service is restricted to those patients where there is a probable diagnosis of AT or a confirmed diagnosis of AT, ATLD or AOA1.

Response time, detail and prioritisation

Patients will be seen on an annual basis following their transition from Nottingham. Patients that need to be seen urgently can be alerted to the Papworth team by the patient's GP, local hospital or by Nottingham. They will then be prioritised as clinically appropriate.

2.5 Interdependencies with other services

The key stakeholders include:

- Nottingham University Hospitals NHS Trust
- University of Birmingham (molecular and protein-based diagnostic service for AT, AT like disorder (ATLD), and ataxia with oculomotor apraxia type 1 (AOA1))
- AT Society
- patients and their families
- Primary Care Trusts in whose areas the affected patients and their families reside
- local clinicians responsible for the patients.

Patients and their families should be able to:

- influence the range of assessment undertaken during their annual visit through the completion of a pre-admission questionnaire which will highlight the specific concerns and needs to the patient
- receive a written report with the results of their assessments including recommendations for their optimum management, a copy of which must be

forwarded to the patient's local therapist, the GP, and any other appropriate provider of care such as those in secondary

- contact the hospital to discuss any aspect of their assessment or management plan
- be involved with the production of information / literature/ website information
- participate in providing feedback to ensure that Papworth is providing a good quality service and addressing the key concerns of patients and their families
- influence what services the hospital provides and how it is run by providing feedback from their visit to the clinic to the AT Society.

The feedback questionnaires should form a part of the review meeting to demonstrate how and what improvements have been made.

Relevant networks and screening programmes

There are no national/clinical networks/expert patient programmes and screening programmes applicable to the service.

3. Applicable Service Standards

3.1 Applicable national standards e.g. NICE, Royal College

In addition the core members for the AT service will meet NHS England on a bi-annual basis to review the performance standards.

4. Key Service Outcomes

Quality and performance standards

Quality Performance Indicator	Threshold	Method of measurement	Consequence of breach	Report Due
Infection Control – MRSA	As agreed between the Provider and the Co-ordinating Commissioner			
Infection Control – C Diff	As agreed between the Provider and the Co-ordinating Commissioner			
Numbers waiting	NA			
Length of Wait	NA			
Mortality	NA			
Unplanned admissions	NA			
Improving Service Users & Carers Experience	Annual patient satisfaction questionnaire through the AT society. Papworth’s internal inpatient questionnaire.			
Outcomes	Maximise life expectancy and quality of life. Minimise hospital admissions and the requirement for unplanned medical intervention. Increase understanding of the disease through data collection, research and shared learning.			

5. Location of Provider Premises

Papworth Hospital NHS Foundation Trust
 Papworth Everard
 Cambridgeshire
 ENGLAND 23 3RE

Subcontractors

The University of Birmingham to cover the molecular diagnostic testing.
 Cambridge University Hospitals NHS Foundation Trust for the provision of neurological diagnostic tests.

King’s Lynn Hospital for the session commitment for the service’s Consultant Neurologist.