**E09/S(HSS)/a**

**2013/14 NHS STANDARD CONTRACT FOR**
**ATAXIA TELANGIECTASIA SERVICE (CHILDREN)**
**PARTICULARS, SCHEDULE 2 – THE SERVICES, A – Service Specification**

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|---|---|
| **Service Specification No.** | **E09/S(HSS)/a** |
| **Service** | Ataxia telangiectasia service (Children) |
| **Commissioner Lead** | |
| **Provider Lead** | |
| **Period** | 12 months |
| **Date of Review** | |

**1. Population needs**

Ataxia-Telangiectasia ('A-T') is an early childhood onset, progressive, multi-organ, neurodegenerative debilitating disorder characterised by cerebellar ataxia, immunodeficiency and greatly increased risk of malignancy. Patients vary at presentation with individual combinations of neurological features. A-T is a very rare condition with an incidence in Caucasians of about 1 per 300,000.[1] There are probably, therefore, about 180 individuals in the UK with A-T.

Target Population: The target population includes children with recurrent sino-pulmonary infections associated with progressive ataxia, any child with progressive cerebellar ataxia or oculomotor apraxia, and children or adults with T-cell leukaemia or lymphoma or unexplained progressive movement disorder.

Reference

1. Woods CG, *et al*. Unusual features in the inheritance of ataxia Telangiectasia. *Hum Genet* 1990;84:555-562.

**1.1 National/local context and evidence base**

Ataxia-Telangiectasia ("A-T") is an early childhood onset, progressive, multi-organ, neurodegenerative debilitating disorder characterised by cerebellar ataxia, immunodeficiency and greatly increased risk of malignancy. Patients vary at presentation with individual combinations of neurological features.

Many patients with A-T are misdiagnosed as having ataxic cerebral palsy or one of several other conditions, resulting in a delay of five or more years before an accurate diagnosis is eventually made. A prompt, accurate diagnosis leads to earlier intervention (e.g. immunoglobulin, vaccinations etc.) and the offer of appropriate genetic advice to parents. It will often also help to avoid expensive and sometimes potentially dangerous procedures; radiotherapy, for example, can be life-threatening to this group of patients but can only be

avoided if the diagnosis is secure.
At present, no treatment is available to ameliorate or to prevent the progressive nature of A-T. However, there have been significant advances in treating the accompanying immune deficiency, resultant sino-pulmonary infections, respiratory insufficiency and movement disorders. In addition, recent studies have raised the hope of a treatment for the marked eye movement disorder in A-T that has a major impact on reading ability.

A single site clinic, having assembled for the specific purpose a team of experts in a rare condition and seeing, on average, at least six patients per session will, simply as a result of economies of scale and acquired expertise, be more cost and medically effective than other geographically dispersed, isolated consultants trying to deliver the same quality of care patient by patient.

The small numbers of patients make statistical comparisons unsound. Certainly, no reliable data are available on A-T patients and their health, as a group, before the establishment of the existing clinic. However, intuitively, the therapy for the immune disorder alone has resulted in fewer admissions to hospital (and consequential disruption to school and home life) and subsequent respiratory dysfunction. Before 1993 when the A-T Society established the current clinic, the available literature suggested a life expectancy into the mid-teens. An American study (Crawford T O et al. *Survival probability in ataxia telangiectasia* Archives of Disease in Childhood, July 2006, pages 610 - 611) puts the prospective median survival at 25 years. Although there is a wide range of life spans of people with A-T, this is a significant improvement over the last 14 years.

**2. Scope**
Only patients who have a confirmed diagnosis of A-T or another rare A-T-like disorder (e.g. ATLD caused by hMRE11 and occasional ataxia oculomotor apraxia type 1) are seen in this clinic.

The service is commissioned to assess and advise on the treatment of paediatric A-T patients (16 years or less).

**2.1 Aims and objectives of service**
**Objectives**

The Clinic has two fundamental priorities:

- provide an accurate diagnosis of the condition
- provide comprehensive advice to responsible local clinicians on the holistic management of the condition
- offer patient centred assessment and advice regarding the many organ specific problems associated with AT, with seamless transition into adult care
- minimise impact on the patient and their family life, education and work practice
- liaise with, and advise, healthcare workers in all relevant disciplines. To enhance quality of life of AT patients
- to increase good quality life expectancy of AT patients.
- to provide an exemplary and comprehensive service for all eligible referred patients with AT
- expert management of patients with AT through the use of the most up-to-date clinical protocols and surgical management

- to operate a rolling programme of clinical audit to test current practice and inform the evolution of care in AT
- to provide care with a patient and family centred focus to maximise the patient experience of care within the nationally designated providers
- to be seen as the leading clinical services and a source of expert advice for the diagnosis and management of AT within the NHS
- to support local healthcare providers to manage patients with AT whenever it is clinically appropriate and safe to do so
- provide high quality information for patients, families and carers in appropriate and accessible formats and mediums
- To develop the experience, knowledge and skills of the multi-disciplinary team to ensure high quality sustainable provision.

Attending the clinic should:

- decrease hospital admissions due to infections and in relation to inappropriate use of ionising radiation
- avoid expensive, unnecessary investigations

**2.2 Service description/care pathway**

**2.1 Service description**

The A-T Society is responsible for allocating patients to the National Ataxia-Telangiectasia Clinics where the patients are assessed by a multidisciplinary team made up of:

- adult and paediatric neurologists
- paediatric immunologist
- paediatric respiratory physician
- neuro-ophthalmologist
- clinical geneticist
- physiotherapist
- occupational therapist
- speech and language therapist
- dietician
- genetics counsellor

Detailed reports with recommendations from each specialist are compiled and returned to the local team caring for the patient.

Only patients who have a confirmed diagnosis of A-T or another rare A-T-like disorder (e.g. ATLD caused by hMRE11 and occasional ataxia oculomotor apraxia type 1) are seen in this clinic.

The service is commissioned to assess and advise on the treatment of paediatric A-T patients (16 years or less). In addition it is the entry point for older patients to ensure that a comprehensive picture of the disease is captured.

The service is administered and coordinated from Nottingham and is supported by a dedicated A-T database derived from the Primary Immunodeficiency patient database at Great Ormond Street Hospital and Newcastle which is already established and funded as part of the NHS Specialised Services Severe combined immunodeficiency and related disorder (SCIDS) service at these hospitals. This database is capable of submitting patient

information across the web to the European Society for Immunodeficiencies' database which already has an A-T sub-registry.

This service provides diagnosis, clinical review and ongoing management advice for patients with Ataxia Telangiectasia (A-T) or other rare A-T like disorders. The service includes:

- diagnostic testing for patients with suspected Ataxia Telangiectasia undertaken with molecular genetic testing
- multi-disciplinary review (typically on a 2-3 yearly basis )
- ongoing advice through comprehensive feedback to local teams on management of each patient's condition.

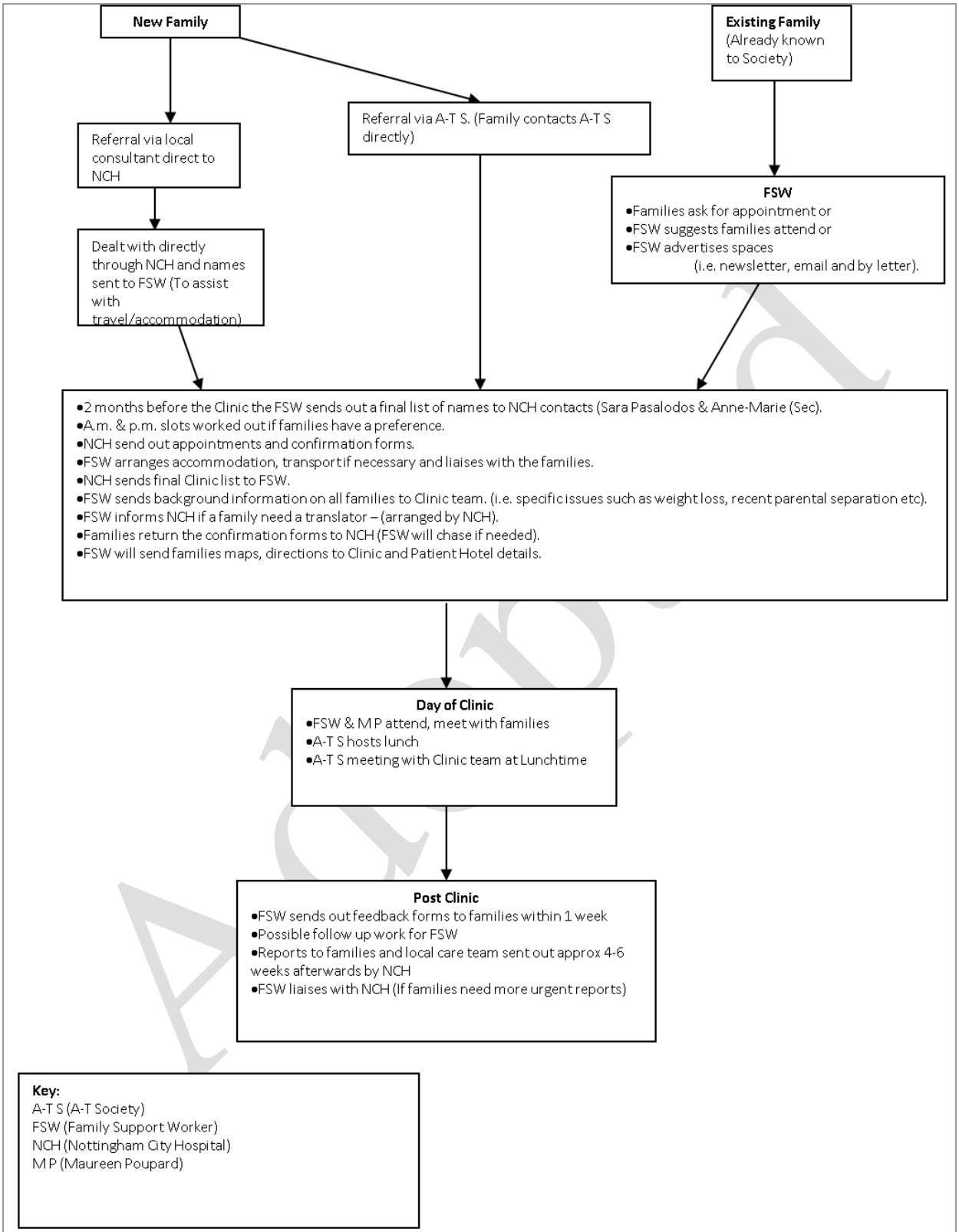
The service will be restricted to those patients where there was a confirmed molecular diagnosis of A-T (or A-T-like disorder) or where there was strong clinical indication of A-T but where diagnostic confirmation was required.

The service is for both paediatric and adult patients. Older patients enter an appropriate care pathway so that a comprehensive understanding of the condition as it affects all UK patients might be achieved.

The service will be provided in association with the Ataxia-Telangiectasia Society who (funded through a separate contractual arrangement with NHS Specialised Services will work jointly with the provider to organise and co-ordinate multi-disciplinary clinics and liaise with patients regarding clinic appointments.

**Communications and patient involvement:**
Nottingham University Hospitals NHS Trust will work with NHS England (NHSE) to ensure that sufficient consideration is given to communications with all stakeholders. The AT society play an active role within this service and are responsible for:

- ensuring clinics are well co-ordinated
- booking patients into clinics
- arranging accommodation for families/carers attending the clinic
- obtaining patient feedback from the clinic
- working with the Trust to act on areas of improvement
- liaising with patients and carers offering advice and support




**Days/hours of operation**

The Nottingham AT Clinic is currently held over 2 days, usually a Thursday and Friday between the hours of 9 am and 5 pm.

Patients and their parents/carers are accommodated either at the Patient Hotel at the City Hospital Campus or at a local hotel.

**Risk management**
Care delivered by the A-T service providers must be of a nature and quality to meet the care standards, specification and Agreement for the service. It is the Trust's responsibility to notify the commissioner on an exceptional basis should there be any breaches of the care standards. Where there are breaches any consequences will be deemed as being the Trust's responsibility.

Patients must be managed in line with the specification and care standards. Any deviation from these which has not been approved by the NHS England is at the Trust's risk both clinically and financially. It is the Trust's responsibility to inform the commissioners of any such non-approved deviations on an exceptional basis.

Where a patient's presentation challenges the assumptions that underpin the specification, service standards and contractual arrangements it is the Trust's responsibility to inform the commissioners on an exceptional basis, prior to any treatment (except for emergency treatment) so that the implications of the patient's requirements can be considered. This does not affect situations where the Individual Funding Application Process applies.

**2.3 Population covered**
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 and the Trust must have separate arrangements in place.

**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 Nottingham AT Clinic are either received through the AT Society or directly to A-T Clinic from Paediatric Neurologists, Community Paediatricians, or Neurologists looking after patients with AT, ATLD or AOA1. In most patients the diagnosis of A-T has been made by chromosome analysis or tests of chromosomal radiosensitivity and further confirmation is obtained by protein and molecular genetics tests performed by Professor Taylor's Laboratory at the University of Birmingham.

Referrals made directly to the Nottingham AT Clinic are forwarded to the AT Society so that they can be put on a single waiting list and allocated to a clinic.

**Referral criteria**

1. Patients with a confirmed diagnosis of A-T (suggestive clinical features with increased chromosomal radiosensitivity and reduced expression of ATM or expression of ATM with no detectable kinase activity in lymphocytes or 2 pathogenic mutations in the *ATM* gene)
2. Patients with probable A-T (suggestive clinical features with raised AFT levels and increased chromosomal radiosensitivity or characteristic chromosomal rearrangements)
3. Patients with a confirmed diagnosis of A-T Like Disorder (ATLD) (suggestive clinical features with reduced expression of hMRE11 in lymphocytes or 2 pathogenic mutations in the *hMRE11* gene)
4. 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 A-T or a confirmed diagnosis of AT, ATLD or AOA1.

**Response time, detail and prioritisation**
Patients that need to be seen urgently can be prioritised to the first available clinic.

**Discharge criteria**
Patients are not usually discharged from the Nottingham AT Clinic. Adult patients (16 and over) are referred to the Papworth multi-disciplinary A-T Clinic Service in partnership with the adult team and the A-T Society.

Children with AT aged 16-18 years would in most instances be referred by their local Paediatric team to their local Transitional / Adult Team, and the Nottingham AT Clinic will then liaise with the relevant local Transitional l/ Adult team for follow-up care of these patients

**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 what services the clinic provides and how it is run by providing feedback from their visit to the clinic to the AT Society
- participate in providing feedback to ensure that the clinic is providing a good quality service and addressing the key concerns of patients and their families.

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

The AT clinic must provide

- a written report including recommendations for optimum management of the patient.
- These must be forwarded to the patient's local community paediatrician or paediatric neurologist (or physician/neurologist for an adult patient), the therapists looking after the patient locally, the GP and the parents or carers of the patient and where appropriate the patient, within 4 weeks from attendance at the clinic

The AT Clinic must actively seek

- additional information when required
- It is the responsibility of the AT clinic to ensure local care providers copy the AT team into all subsequent correspondence regarding the patient.
- The service must ensure there is close contact between the Nottingham and Papworth services to ensure that adult patients with AT receive the best possible care.

External to this the nationally designated AT provider is the leader in the NHS for patient care in this area. They provide a direct source of advice and support when other clinicians refer patients into the nationally designated providers

The nationally designated providers also provide education within the NHS to raise and maintain awareness of AT and its management.

The national providers will form a relationship with local health and social care providers to help optimise any care for AT provided locally for the patient. This may include liaison with consultants, GPs, community nurses or social workers etc.

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

The nationally designated AT provider must be fully integrated into their Trust's corporate and clinical governance arrangements and must fully comply fully with Clinical Negligence Scheme for Trusts (CNST) and Care Quality Commission (CQC) requirements in terms of quality and governance.

The service will ensure that there are:

- regular meetings with patient representatives;
- all practitioners will participate in continuous professional development and networking;
- patient outcome data is recorded and audited across the service.

The commissioners and service will conduct a formal Joint Service Review at least every six months. All centres must participate in the national audit commissioned by NHS Specialised Services - audit meetings should address:

- Clinical performance and outcome

- Process-related indicators, e.g. efficiency of the assessment process, prescribing policy, bed provision and occupancy, outpatient follow up etc.
- Stakeholder satisfaction including feedback from patients, their families, referring surgeon and General Practitioners

Please refer to NHS England Service Standards for the Ataxia-Telangiectasia Service

**4. Key service outcomes**

| ***Quality Performance Indicator*** | ***Threshold*** | ***Method of measurement*** | ***Consequence of breach*** | ***Report Due*** |
|---|---|---|---|---|
| **Infection Control – Methicillin-resistant *Staphylococcus aureus* (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** | | | | |
| **Length of Wait** | | | | |
| **Mortality** | | | | |
| **Unplanned admissions** | | | | |
| **Improving Service Users & Carers Experience** | | | | |

**5. Location of provider premises**

Nottingham University Hospitals NHS Trust
City Hospital campus
Hucknall Road
Nottingham
NG5 1PB

**Subcontractors**

Assessment of chromosomal radiosensitivity, protein-based diagnostic tests and molecular genetic analysis for AT, ATLD and AOA1 to the Nottingham AT Clinic is provided by University of Birmingham