

A17/S(HSS)e

**2013/14 NHS STANDARD CONTRACT
FOR Alström syndrome service (aLL AGES)**

SECTION B PART 1 - SERVICE SPECIFICATIONS

Service Specification No.	A17/S(HSS)e
Service	Alström syndrome service (All Ages)
Commissioner Lead	
Provider Lead	
Period	12 months
Date of Review	

1. Population Needs

Alström is a rare genetic syndrome that usually presents with blindness in childhood. Patients go on to develop obesity due to insulin-resistant diabetes, and a fibrosing cardiomyopathy and renal failure. They may also become deaf. There are thought to be 30-40 families in the UK with the condition.

1.1 National/local context and evidence base

As a result of the clinic it has been possible to define the phenotypic variation and extent of complications. This has included

- Close follow up and treatment of insulin resistance, diabetes, hypertriglyceridaemia and secondary endocrine failure e.g. thyroid, and gonadal
- Demonstration of the prevalence and incidence of cardiomyopathy, renal disease, hepatic fibrosis and urological dysfunction.
- Prevention of pancreatitis.
- Slowing rate of decline in renal function.
- Intensive management during surgery and chest infection to prevent hypoxic death.

Several publications have described these changes and most effective therapy particularly with respect to metabolic problems.

2. Scope

2.1 Aims and objectives of service

The service aims to offer early diagnosis and recognition of complications of Alström syndrome. Prompt intervention with appropriate established therapies. Close liaison with local medical and allied healthcare professional teams. Support and advice to families educational and social service agencies to maximise potential development and social activities.

The service aims to offer early diagnosis and recognition of complications of Alström syndrome. Prompt intervention with appropriate established therapies. Close liaison with local medical and allied healthcare professional teams. Support and advice to families educational and social service agencies to maximise potential development and social activities.

To provide a holistic, patient centred service to patients with Alström Syndrome supported by a multi-disciplinary team:

- to monitor and manage disease progression
- coordinate care and management of patients with Alström
- provide support for local healthcare providers
- provide genetic counselling, a national genetic testing service and prenatal diagnosis opportunities
- establish disease specific centres of excellence and expertise
- provide a national reference network of specialist centres for the clinical management of the syndrome.

The purposes and goals of the service to consolidate diagnosis and care of those with Alström syndrome

The service will:

- improve awareness, diagnosis and management of Alström Syndrome within

the UK by education and provision of highly specific and sensitive diagnostic testing alongside liaison with and education of local healthcare providers.

- offer patient centred assessment and advice regarding the many organ specific problems associated with Alström Syndrome, 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
- enable consistent access to expert care across all relevant disciplines
- provide patient specific assessment and management with close collaboration with local care providers
- reduce the burden of hospital visits and inappropriate management for patients and their families, making the experience enjoyable and educational.

All families with the syndrome are referred to attend the University Hospitals Birmingham (adult) or Birmingham Children's Hospital (children) service.

2.2 Service description/care pathway

Service Structure

Provision of the service will be through ongoing support for patients, families and clinicians as required, which will be coordinated primarily by the clinical nurse specialist and Alstrom Syndrome UK (ASUK). Providers will provide family-centred one-stop clinic for adults & children.

The clinic will be held over two days allowing Providers to perform more extensive assessment of patients.

Assessment and ongoing support provided

Initial Assessment:

Assessments will be tailored to individual needs. During the 2 months leading up to the clinic families liaise with the lead physician directly and through AS UK to plan the necessary focus for their review-see days and hours of operation.

The service should run quarterly two day clinics with overnight stay. These consist of

1. Day 1 morning group discussion, phlebotomy, 24 hr blood pressure monitor, exercise and physiotherapy sessions.
2. Day 2 rotation of audiology, vascular, metabolic, respiratory and cardiac reviews. Special X-rays/magnetic resonance imaging (MRI)/ultrasound or urological review if appropriate.

Delivery of service

Afternoon of second day to sum up clinic and confirm new or changed treatment for each attendee. Planning with families of next visit and referrals to other agencies.

Days and hours of operation

Initial outpatient appointments will take place from Monday to Friday between 0900 and 1630. The annual clinic will be held on a date agreed by the University Hospitals Birmingham or Birmingham Children's team. The lead physician, dietician, exercise physiologist, cardiac technician (for 24 hr blood pressure) phlebotomist attend on the first day. The afternoon is devoted to exercise and lifestyle advice.

The second morning involves a timetabled rotation of visits to audiology, cardiology, respiratory medicine, endocrinology, radiology, rheumatology and vascular medicine. Each patient has their own care pathway according to need. Urology appointments are arranged in advance for the minority with urethral detrusor dys-synergia.

By the second day afternoon the main blood test results and 24 hr blood pressure results are available and discussed with families.

Paediatric service

—

The Alström medical clinic (paediatric)

This will be outpatient or heart investigation unit based; staffed by the designated paediatric consultant and paediatric specialist nurse; dietician; paediatric psychologist; and Alström syndrome UK family liaison officer. The specialist nurse will weigh and measure the child (and parents), and plot on growth charts. The cardiology technician and paediatric cardiologist will undertake an echocardiogram. The designated consultant will complete the structured history sheet; and examine the child for physical signs of obesity complications e.g. acanthosis nigricans, tendon xanthomata. At the end of the clinic the child will be invited to have a snack and have blood pressure checked, together with a dietary assessment. Invasive tests such as blood tests will only be advised if there are inadequate facilities locally or there is an urgency e.g. osmotic symptom of diabetes.

—

Output_

—

Children will leave the Alström clinic by one of the following routes:

- a. Over 18 completed years: refer transitional Alström clinic.
- b. Complications detected e.g. cardiac/renal/respiratory failure: refer to appropriate specialist clinic heart transplant assessment clinic; renal clinic; local specialist service.
- c. Fully coordinated local services, patient stable, national clinic superfluous.
- d. Outreach clinic set up e.g. in Leeds for Yorkshire patients to be referred to
- e. Alternative diagnosis made: refer appropriate specialist clinic if any

Patient and public involvement and communications

Providers will work with the NHS England to ensure that sufficient consideration is given to communications with all stakeholders. The Alström patient group will be closely involved in service design. By continuing this liaison with the patient support group providers will ensure that the families served are active participants in the care-giving role. Clinics will be tailored around identified needs and parents will be given the information they need to better care for their children at home.

Outcomes

- Rapid recognition and treatment of serious complications e.g. cardiomyopathy, uropathy, hepatic fibrosis, hypertriglyceridaemia, restrictive lung function.
- Long term reduction in vascular resistance to alleviate chronic pressure on the heart.
- Reduction in hyperinsulinaemia and hyperglycaemia with individually planned exercise, restricted carbohydrate diet and drug treatment where safe.
- Age at diagnosis of referred patients to fall.
- Availability of genetic test results to improve with turnaround time less than 3 months.
- Number of people developing unexpected heart failure in teenage and young adult age range to fall as annual screening detects cardiomyopathy while still asymptomatic.
- Glycated haemoglobin (measure of glucose control) to fall with more aggressive treatment of insulin resistance
- Serum triglycerides to fall with treatment of dyslipidaemia.
- Stabilisation of BMI-SDS

Risk management/clinical governance

Care delivered by the Alström Service 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.

Accessibility/acceptability

The service is open to all. ASUK leads attend clinic to support families. Information provided in Braille, Moon, large print and voice mail.

The Provider has a duty to co-operate with the commissioner in undertaking Equality Impact Assessments as a requirement of race, gender, sexual orientation, religion and disability equality legislation

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 Alström 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.

2.3 Population covered

As above

2.4 Any acceptance and exclusion criteria

Referrals

Referrals come through ASUK or professional medical referral or patient access via website info.

Criteria for clinic review are:

childhood onset photophobia, nystagmus, and cone rod dystrophy with obesity insulin resistance and /or neuronal deafness, infantile dilated cardiomyopathy gene test preferred with at least one pathological mutation in the AS gene. Children will be accepted into this clinic if they are under 18 completed years and:

- referred from a clinician with a prior diagnosis of Alström syndrome
- referred from a clinician with a diagnosis of a syndrome that overlaps or is confused with Alström e.g. Bardet-Biedl syndrome
- referred from a clinician with a suspected diagnosis of Alström referred from Alström syndrome UK

2.5 Interdependencies with other services

The service can only function with cogent and prompt communication and full cooperation of general practice, social services liaison officers for specific ethnic groups. – Annual or biennial clinic visits can only effect changes for the patient and their family if plans are carried through by local medical educational and social care services.

—

3. Applicable Service Standards

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

Quality Performance Indicator	Threshold	Report due	Consequence of breach
Adults: % patients with an HbA1c <9 % patients with a BMI <35; % patients with triglycerides <10 mean age at death	Significant variation from the national average or, in services with one or two national centres, significant variation from the outcomes achieved in the previous three years	Annual report (September of contract year) with data from previous financial year April to March	Performance notice as set out in Clause 32.4

4. Key Service Outcomes

Outcomes:

- good clinical control with acceptable therapy of disorders of metabolism e.g. control of type 2 diabetes HbA1c <7.5%; hyperlipidaemia serum triglycerides < 5 mmol/l, normalisation of thyroid and gonadal function
- development of detailed management protocols for enhanced local care. Early diagnosis and palliation of progressive major organ fibrosis e.g. renin aldosterone blockade to slow progression of cardiac fibrosis measured by symptoms, ECG, Echocardiogram, and serum BNP levels, and renal fibrosis measured by serial blood pressure, urine albumin excretion, renin levels and serum creatinine
- prevention of hypoxic crises by planned intensive management of surgery and liaison with local teams during acute illness.

—

5. Location of Provider Premises

Birmingham Children's Hospital NHS Foundation Trust

University Hospitals Birmingham NHS Foundation Trust