E06/S(HSS)/a

2013/14 NHS STANDARD CONTRACT
FOR ALKAPTONURIA SERVICE (ADULTS)

PARTICULARS, SCHEDULE 2 – THE SERVICES, A- SERVICE SPECIFICATIONS

Service Specification No.	E06/S(HSS)/a
Service	Alkaptonuria service (Adults)
Commissioner Lead	
Provider Lead	
Period	12 months
Date of Review	

1. Population Needs

1.1 National/local context and evidence base

Alkaptonuria (AKU) is a rare autosomal recessive condition affecting 1 in every 250,000 to 1 in 1000,000 people. It is a metabolic disorder caused by the lack of the enzyme homogentisate 1, 2-dioxygenase (HGD).

AKU has three main features:

- the first and earliest of which occurs in the urine, which darkens on standing owing to oxidation of the HGA
- premature severe arthritis affecting the spine and large weight-bearing joints follows pigmentation (also called ochronosis), leading to multiple joint replacements and a w aortic valve stenosis
- wheelchair-bound existence.

Evidence

Clinical: There are some good studies that have described the clinical features and the morbidity of AKU (Phornphutkul et al, 2002, O'Brien et al 1963, Cox & Ranganath 2011). There are good publications describing the spinal features of the disease (Akeda et al 2008, Kusakabe et al 1995, Al-Mahfoudh et al 2008). There are many descriptions of arthritis of AKU (Toth et al 2003, Perry et al, Al-Mahfoudh et al 2008). Cardiac valve disease has been highlighted by several authors (Pettit et al 2011, Phormphutkul et al 2002). Renal stone disease has been the focus of study by many groups (Zibolen et al 2000, Krizek et al 1971). Tendon and muscle ruptures

are prevalent (Manoj Kumar & Rajasekaran 2003

2. Scope

2.1 Aims and objectives of service

The aims and objectives described earlier relate to the desire to establish an inpatient-based service that will provide health care and treatment for patients with Alkaptonuria (AKU).

Outcomes:

- patient identification and contact
- comprehensive assessment of disease burden
- formulation of Management plans and shared care arrangements
- identification of new disease burden
- commencement and utility of new treatments
- develop audit tools
- education
- training
- monitoring for safety and toxicity of novel treatments.

The clinical aspects of the NAS will be assessed as follows:

- decrease in joint replacement
- improvement in Heath questionnaires of daily living and pain
- decrease in Alkaptonuria severity score index (AKUSSI) in treated cohort.

2.2 Service description/care pathway

Identified patients in England will be invited to attend the National Alkaptonuria Service (NAS) for a 3-day assessment. The NAS will:

- deliver annual multi-professional assessment to maximise diagnosis and treatment opportunities
- deliver patient-centred approach to ensure that individual patients receive the best available management
- ensure continuity of care at local level when patients are not at the National Centre
- deliver high quality assessment
- deliver accurate identification of morbidity in patients
- enable appropriate treatment
- co-ordinated care
- deliver effective communication and education to ensure continuity of care
- have an ability to deal with crises

- deliver quality auditable care
- deliver opportunities to enhance research

- deliver efficiency through appropriate and co-ordinated care

Significant morbidity of patients with AKU will be detected by Multidisciplinary team through a comprehensive set of investigations .
For patients aged between 16 and 34, the standard assessment will be adapted to respond to their degree of impairment and disability.

Care pathway

The care pathway (Appendix 2) outlines the patient journey in the NAS. This includes 3 days of inpatient assessment, education and treatment. Upon completion of this inpatient programme, patients are discharged back to local health care until the next annual visit. On an exceptional basis patients may require assessment in between their annual visits which will be accommodated by the NAS. Any assessment or treatment interventions that are beyond the normal scope of the NAS (appendix 2) are not included in the nationally commissioned service and agreements with the patient's local commissioner may be required.

In addition to the discharge summary that will accompany patients at discharge, a comprehensive report will be produced quickly to enable continuity of care with the local health care providers no later than 5 working days post discharge.

Where necessary, further communication will take place between the NAS and Local providers to ensure effective service delivery for patients.

The daily programme of assessments and treatments is outlined in Fig 3. While this time-table is a guide, there will be sufficient flexibility to ensure this programme is completed over three days.

A nurse will be dedicated to each patient to ensure that patient's journey is comfortable over the 3 day programme. Such an approach will ensure that all parts of the multidisciplinary clinical assessments and treatment are carried out efficiently and in a timely manner.

2.3 Population covered

NHS England commissions the service for the population of England

At present the NHS England contract includes provision for the service to treat eligible overseas patients under S2 [Under EU regulations, patients can be referred for state funded treatment to another European Economic Area (EEA) member state or Switzerland, under the form S2 (for EU member states) or the form E112 (for Iceland, Norway, Liechtenstein and Switzerland)] referral arrangements. Providers

are reimbursed for appropriately referred and recorded activity as part of the NHS England contract.

Trusts performing procedures on EU-based patients outside of S2 arrangements will need to continue to make the financial arrangements directly with the governments involved, separately from their contract with the NHS England.

With regard to S2, the mechanism for recovery of costs has been via the Department for Work and Pensions Overseas Healthcare Team. They are responsible for agreeing reconciliation and recovery of costs with European administrations. These arrangements were implemented in October 2009, though a similar process existed previously. The financial flows are therefore back into the Treasury rather than back to Trusts.

2.4 Any acceptance and exclusion criteria

Exclusion criteria

Any AKU patient who cannot travel will not be able to avail himself / herself of the NAS.

For assessment, patients under age 16 years will be excluded from the NAS.

For treatment with nitisinone, pregnant patients and those under 16 years will be excluded.

All patients will be expected to have a definite diagnosis of AKU, made on the basis of characteristic clinical features and homogentisic acid assay. The referral letter will be used to screen patients to ensure they fulfil the criteria for inclusion. All patients will have an initial screen for routine bloods immediately prior to attendance that could be made available with the referral letter or brought by patients when they arrive in Liverpool.

2.5 Interdependencies with other services

The NAS will work closely with the patient group AKU Society in bringing identified patients to Liverpool. The NAS will work closely with the patient body to ensure that the service remains responsive and relevant to patient needs.

Full feedback of all activities on AKU patients will be fed back to GP's responsible for the overall care of the patient. Any concerns and issues will be dealt with by full and effective communication. The AKU society will assist in communicating with GP's where appropriate.

GP's will be able to approach the NAS regarding patients under their care for advice on management. GP's can ask the NAS to provide a service not available locally although this care would be outside the scope of the nationally commissioned service.

The national service will provide specialist care based on specialised knowledge and expertise. This service will exclude care and procedures that can be provided effectively by local healthcare. Patients visit the National Service annually and therefore local mechanisms will be cultivated to ensure appropriate follow-up care.

3. Applicable Service Standards

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

Governance

The providers of the NAS service must ensure they are fully integrated into their Trust's corporate and clinical governance arrangements and must comply fully with the Clinical Negligence Scheme for Trusts (CNST) and Care Quality Commission (CQC) requirements in terms of quality and governance.

Each centre 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.

4. Key Service Outcomes

Quality Performance Indicator	*Threshold*	*Method of measurement*	*Consequence of breach*	*Report Due*
Decrease in joint replacement				
Improvement in Heath questionnaires of daily living and pain				
Decrease in AKUSSI in Treated cohort				
Decrease in ear ochronosis by biopsy over 5 years		Ear cartilage biopsy at base line and		

		5 years		

5. Location of Provider Premises

The Royal Liverpool and Broadgreen Univeristy Hospitals NHS Trust

APPENDIX 2 AKU Severity Score Index (AKUSSI):

Treatments delivered by the NAS and those for which advice will be given to local providers within the comprehensive management plan

Feature	Score	Feature	Score
Eye Pigment R Eye Nasal	4	L Eye Nasal	4
R Eye Temporal	4	L Eye Temporal	4
Ear Pigment R Ear None	0	L Ear None	0
R Ear Present	2	L Ear Present	2
R Ear Marked	4	L Ear Marked	4
Prostate Stones	4	**Kidney Stones**	4
Strokes (CVA)	8	**Parkinson's disease**	8
Osteopenia	4	**Fracture**	8
Teeth pigment	4	**Skin** pigment	4
Salivary stone	4	**Middle ear pigment**	6
Hearing impairment	4	**Pigmented larynx**	8
Atrial fibrillation	4	**Any other arrhythmia**	2
Aortic sclerosis	6	**Aortic stenosis**	8
Congestive heart failure	8	**Tendon** rupture	8
Ligament rupture	8	**Muscle** rupture	8
ALL CLINICAL FEATURES ONLY			136
Clinical joint pain score (1 for each large joint area; 14 large joint areas)			14
Scintigraphic scan joint score (2 for each large joint; 14 large joints areas)			28
Number of arthroscopies (2 for each)			6
Number of joint replacements Each joint 4 (Max 10 large joints)			40
NON SPINE JOINT AGGREGATE SCORES			88
Clinical Spinal pain score (2 each for cervical, thoracic, lumbar, sacroiliac)			8
Pain or Isotope Bone Scan score (Pubic symphysis)			4
Kyphosis	4	Scoliosis	4
Scintigraphic scan spine score (5 areas; 4 point for each area; costochondral, Lumbar, Thoracic, Cervical, Sacroiliac)			20
SPINE AGGREGATE SCORES			40
AKUSSI (Clinical + Non-spine + Spine) Scores			266