

A12/S(HSS)/b

**2013/14 NHS STANDARD CONTRACT
FOR XERODERMA PIGMENTOSUM SERVICE (ALL AGES)**

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

Service Specification No.	A12/S(HSS)/b
Service	Xeroderma pigmentosum service (All Ages)
Commissioner Lead	
Provider Lead	
Period	12 months
Date of Review	

1. Population Needs

Currently there are 100 Xeroderma Pigmentosum (XP) patients in the UK. Although the disease population in the UK has been roughly static at 100 patients, in the last five years the annual number of new cases has grown from an average of five newly diagnosed patients/year to around eight newly diagnosed patients/year, which is considered to be due to demographic changes related to immigration resulting in an increased frequency of consanguineous marriages. If this continues the service expects the number of patients to increase to around 130 patients over the next 10 years.

The diagnosis of XP can only be made if the enzyme assay result is abnormal ('deoxyribonucleic acid (DNA) repair assay'). This test is available in one lab (University of Sussex).

The aim of the service is to develop a multi-disciplinary service that can serve the XP patient population.

1.1 National/local context and evidence base

The disease:

Xeroderma pigmentosum (XP) is a rare, life-threatening, inherited multi-organ disorder. There are currently 100 patients in the UK with this condition. Inherited defects in the process of repairing ultraviolet-induced DNA damage result in severe sunburn-type reactions to daylight, skin cancers in exposed skin from early childhood, eye disease (keratitis, conjunctivitis, corneal scarring, eye tumours), and progressive neurological degeneration in 20-30% of patients. 45% of patients

develop skin cancers (mean age of onset 8 years), and 40% of patients develop eye problems (11% develop eye cancers: mean age of onset 6 years). Most patients need long term medical care since 70% survive beyond the age of 40 years. The disease is diagnosed with an enzyme assay ('DNA repair assay').

2. Scope

The service consists of:

- one stop multi-disciplinary clinic: Neurology (paediatric and adult), dermatology (including dermatological surgery), ophthalmology, with other specialties involved as required: based mostly in London but 2 clinics per year to be held in North of England;
- laboratory diagnostic service: putting the current diagnostic laboratory service on a stable footing; providing complementation group analysis for the first time (i.e. disease subtyping for prognosis); providing antenatal testing for the first time;
- Managed clinical network: a national managed clinical network for this national service.

2.1 Aims and objectives of service

- Objectives of the service will be to:
- create a one-stop clinic, reducing the cost and inconvenience of multiple attendances;
- achieve uniformly high standards of care wherever patients are treated within the managed clinical network;
- create a group of clinicians in the three key specialties with expertise and experience;
- provide complementation grouping for prognostic information;
- improve neurological management. As ultraviolet (UV) protection and awareness has improved, more patients are avoiding the multiple skin and eye malignancies and surviving longer so that the neurological side is becoming more important. Early detection of sensorineural hearing loss in children enables fitting of hearing aids; diagnosis of mild cognitive impairment enables appropriate arrangements to be made for schooling;
- provide an exemplary and comprehensive service for all eligible referred patients with XP;
- operate a rolling programme of clinical audit to test current practice and inform the evolution of care in XP;
- provide care with a patient and family-centred focus to maximise the patient experience of care within the nationally designated provider;
- be seen as the leading clinical service and a source of expert advice for the diagnosis and management of XP within NHS;

- support local healthcare providers to manage patients with XP 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;
- develop the experience, knowledge and skills of the multi-disciplinary team (MDT) to ensure high quality sustainable provision.

2.2 Service description/care pathway

The service will consist of:

- **A one-stop multi-disciplinary clinic**
 - over a one or two-day period with consultations in neurology (paediatric and adult), dermatology, ophthalmology, psychotherapy, dermatological surgery, and specialised investigations as clinically relevant.
- **A laboratory diagnostic service**
 - The provider will ensure the current diagnostic laboratory service remains on a stable footing by creating arrangements to ensure that, between the University of Sussex and Guys & St. Thomas' NHS Trust, a diagnostic laboratory service is always available whatever changes in service from the University of Sussex may occur over the coming years.
 - A Complementation Group Analysis (CPA) should also be provided for the first time (i.e. disease sub-typing for prognosis).
- **A managed clinical network:**
 - The provider will develop a national managed clinical network for a truly national service. This will ensure patients are well supported in their local acute hospitals.
 - Patients can become known to the service in various ways - via the diagnostic laboratory which receives tissue for testing from dermatologists around the UK; by direct referral from a consultant (dermatology, clinical genetics, neurology, ophthalmology etc.); patients already known to the service, and patients known to the patient support group. In all these situations (apart from the patient support group where the group approaches the patient in the first instance to seek permission to pass on the patient's details to NHS England, the XP Consultant contacts the local consultant and clinical nurse specialist before contacting the patient with a view to the patient having an initial multi-disciplinary clinic 1-2 day 'one stop' assessment arranged.
 - Part of the initial multi-disciplinary assessment at Guys & St Thomas' NHS Trust is to establish the nature of dermatological, eye and neurological involvement and thus arrange a clinically appropriate follow-up plan, with the balance between local and multi-disciplinary service care depending on the requirements for the special expertise of the service, which in turn depends on the type and severity of disease in each organ. Whenever patients are being looked after for any period locally in between the multi-

disciplinary service appointments, the outreach nurses will be in contact with the patients and local doctors, checking that the minimum standards of care specified in the disease management protocols are being maintained.

- **Communications with stakeholders**

- The provider will work with NHS England to ensure sufficient consideration is given to communications between all stakeholders.
- The provider must demonstrate the active involvement and engagement of patients and the patient support group in the shaping and delivery of the services.
- The provider must demonstrate to NHS England that the service is well publicised and marketed to ensure appropriate referrals are received.

- **Governance**

- The nationally designated Xeroderma Pigmentosum Syndrome service must be fully integrated into the trust's corporate and clinical governance arrangements.

- **Risk management –**

- Care delivered by the XP 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 NHS England, are 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.

- **Days/hours of operation**

- At Guys and St Thomas': The multi-disciplinary clinic will run all day on alternate Fridays, the 'severe skin clinic' every Tuesday afternoon; The MDT meeting, the pathology-dermatology MDT meeting, and the outreach nurse-team lead team meetings will all occur on a fortnightly basis at St Thomas'.
- The diagnostic labs will be open three days a week during normal working hours.

- **Discharge planning**

- Where there is shared care between local consultants and the national service, the XP nurses will regularly contact the patient and their family, as well as the local consultant, and provide assessment and information

regarding care needs according to clinical management protocols written by and maintained by the clinical team.

2.3 Population covered

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

2.4 Any acceptance and exclusion criteria

The service is accessible to all patients with diagnosed XP regardless of sex, race, or gender. Providers are required to ensure that staff attend mandatory training on equality and diversity and the facilities provided offer appropriate disabled access for patients, family and carers. When required the providers will use translators and printed information is available in multiple languages.

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

Waiting and referral response times for appointments will be subject to national and local policies.

2.5 Interdependencies with other services

Internally the XP team will link into multiple clinical and administrative teams as a result of the composition of the broad MDT.

The XP service will be responsible for oversight of all aspects of patient care for every XP patient in England.

External to this, the nationally designated XP providers are the leaders in 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 NHS to raise and maintain awareness of XP and its management.

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

3. Applicable Service Standards

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

Governance

The nationally designated XP service must be fully integrated into the trust's corporate and clinical governance arrangements and must comply fully with the clinical negligence scheme for trusts and Care Quality Commission (CQC) requirements in terms of quality and governance.

Each centre will ensure that:

- regular meetings take place with patient representatives;
- all practitioners 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 once every six months. All centres must participate in the national audit commissioned by NHS England. 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

See also NHS England Service Standards for XP

4. Key Service Outcomes

Outcomes expected are:

- earlier diagnosis of tumours;
- better ultraviolet protection to prevent tumours;
- gradual decrease in number of eye and skin tumours;
- detection of neurological defects which can be helped by early intervention;
- identify the number of patients in whom unsuspected hearing or cognitive problems are identified and for whom effective measures are then taken (hearing aids, schooling adjustments);
- reduction in the number of sunburn episodes;
- improvement in quality of life of patients & carers.

Quality Performance Indicator	Threshold	Method of measurement	Consequence of breach	Report Due
Proportion skin & eye tumours excised at an earlier stage	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			
No of skin & eye tumours post procedure				
Earlier diagnosis & treatment				
No. of sunburn episodes				

5. Location of Provider Premises

Guys & St Thomas' NHS Foundation Trust

Sub-contractors

None.