Highly specialised services 2019

NHS England and NHS Improvement

Highly Specialised Services 2019

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Contents

1. Introduction

Purpose of this document

The primary purpose of this document is to provide key information about highly specialised services during 2018/19. In summary, the information comprises:

- a description of each service
- a list of the expert centres that deliver the service
- NHS England's total expenditure for each service
- a measure of the activity that each service undertakes (patient numbers fewer than 30 are not included because of the risk of identifying individual patients)
- clinical outcomes from the service
- information about geographical equity in access to the service
- new highly specialised services

Appendix A details the membership of highly specialised services in the European Reference Networks.

Appendix B summarises NHS England's commissioning arrangements for highly specialised services across the devolved nations.

Equality statement

Promoting equality and addressing health inequalities are at the heart of NHS England's and NHS Improvement's values. Throughout the development of the policies and processes cited in this document, NHS England has:

- given due regard to the need to eliminate discrimination, harassment and victimisation, to advance equality of opportunity between people who share a relevant protected characteristic (as defined by the Equality Act 2010) and those who do not share it, and to foster good relations between people who share a relevant protected characteristic and those who do not share it

- given regard to the need to reduce inequalities between patients in access to, and outcomes from healthcare services and to ensure services are provided in an integrated way where this might reduce health inequalities.

Specialised services

NHS England is responsible for commissioning specialised services to meet a wide range of health and care needs. In 2018/19 NHS England's total spend on all specialised services was 17.2 billion.

Four factors determine whether a service is commissioned by NHS England as a prescribed specialised service (rather than by clinical commissioning groups [CCGs] as a non-specialised service). The four factors are:

- the number of individuals who require the service
- the cost of providing the service or facility
- the number of people able to provide the service or facility
- the financial implications for CCGs if they were required to arrange for provision of the service or facility themselves.

Highly specialised services

Within specialised services is a subset of services classified as 'highly specialised'.

Each highly specialised service is provided to a smaller number of patients compared to specialised services; usually no more than 500 patients per year.

Due to the small number of patients accessing such services, they are most appropriately delivered and coordinated nationally through a very small number of expert centres. This model of delivery makes it easier to recruit appropriately qualified professionals and to ensure that they receive the level of training needed to maintain their expertise. It also ensures the most effective use of resources through efficient management of patient care and ensuring access to the technology necessary to allow delivery of the services.

Planning highly specialised services on a national, rather than local, basis, provides a challenge for the Highly Specialised Commissioning Team (HSCT) to ensure equitable access to services, given the small number of expert centres and the fact that some patients may live a long way away from centres. It is also important to have a robust process for selecting and monitoring the centres which provide these services, given the very high level of expertise required.

The HSCT need to liaise closely with a range of stakeholders – within NHS England and in other legal entities – especially with colleagues in:

- regional specialised commissioning teams, who hold the budgets and contracts for the services
- the three devolved administrations (NHS Northern Ireland, NHS Scotland and NHS Wales) so that there is clarity about how patients from these countries may access the portfolio of services and so that services are planned UK-wide
- NHS Blood and Transplant as most solid organ transplants services are within the highly specialised portfolio.

Rare Diseases Advisory Group

The Rare Diseases Advisory Group (RDAG) is responsible for making recommendations to NHS England and the devolved administrations of NHS Scotland, NHS Wales and NHS Northern Ireland on the development of services for people with rare diseases and on highly specialised services.

RDAG makes recommendations to the Clinical Priorities Advisory Group (CPAG) about how highly specialised services should be commissioned, including providing advice on which services or technologies should be prioritised for investment. In addition, RDAG recommends the most appropriate model of provision for the service and which expert centres may (or may no longer) be nominated to deliver highly specialised services.

RDAG receives outcome information on the services and makes recommendations on any action required as a consequence of poor outcomes as well as ensuring proper provision of services commissioned, with equal access opportunities for patients across different geographies.

RDAG makes recommendations to NHS England and the devolved administrations on developing and implementing strategy for highly specialised services including making recommendations on how the UK Strategy for Rare Diseases should be implemented.

Expenditure figures

The expenditure figures for each service reflect NHS England's expenditure on that service in 2018/19. Expenditure is set out into the following categories:

- <£0.5 million
- >£0.5 million but <£1 million
- >£1 million but <£5 million
- >£5 million but <£10 million
- >£10 million but <£20 million
- >£20 million but <£30 million
- >£30 million but <£50 million
- >£50 million.

Clinical outcomes for highly specialised services

Monitoring of clinical outcomes is a key responsibility of the HSCT. Highly specialised services are unusual in the extent of clinical outcome monitoring in place, which for most services includes measures relating to all patients treated in the service.

The data for each centre providing a service are presented at the annual audit meeting for the service and provides a stimulus for challenge and learning (or confirmation of good practice).

In some services, outcome information cannot be published because the small numbers involved could potentially lead to the identification of individual patients. In some other cases, the data are too small to analyse. In these cases, the data is reviewed and held by the HSCT.

Geographical equity of access to highly specialised services

The central ethos of commissioning highly specialised services is to concentrate expertise in a small number of expert centres. The trade-off is that access may be difficult for patients who live a long way from their nearest expert centre. Hence a priority for the HSCT is to monitor the geographical access to highly specialised services.

The best measure for this is the standardised co-efficient of variation (SCV). An SCV above 20 indicates variation greater than expected by chance and an SCV below 20 can be considered to be random variation.

For each service, patients are mapped according to their home postcode, and rates per million population (child, adult or all-age as appropriate) calculated for each region or area. For most analyses, patients are mapped if they received the intervention (for example, transplant) or have a confirmed diagnosis of the relevant condition (for example, xeroderma pigmentosum).

Genuine clusters of disease can potentially explain observed variation that is unlikely not to have occurred by chance (SCV >20). For example, many genetic disorders are more common among highly consanguineous populations who themselves are unequally distributed in England. Conversely, there may be a genuine paucity of patients with a particular condition in particular areas. For example, patients with severe osteogenesis imperfecta (a condition that is unlikely to be missed) are fewer in number in the North East.

For those services where the SCV is above 20, the HSCT reviews the information in greater detail to understand the possible causes. They then explore options and take specific actions to reduce the inequalities, followed by a repeat analysis of the geographical data at an appropriate time.

The HSCT routinely analyses geographical equity for each service every three years, last doing so in 2017.

2. New highly specialised services commissioned during 2018/19

Auditory brainstem implant for patients with congenital abnormality of the auditory nerves or cochleae

In October 2018, a new highly specialised service providing auditory brainstem implantation (ABI) for children with profound deafness came into full operation, provided by two Trusts in England: Manchester University NHS Foundation Trust and Guy's and St Thomas' NHS Foundation Trust. The service was commissioned following approval of the associated policy (in December 2016) and service specification (in June 2018).

The surgery is for children who are profoundly deaf, aged five or under and who are unable to use conventional hearing aids or implants because their inner ear (cochlea) or auditory nerve did not develop properly. The highly complex procedure involves inserting a device directly into the brainstem to stimulate hearing pathways, bypassing the cochlea and auditory nerve.

It is estimated that about 15 children will be referred for assessment each year and up to 10 considered suitable for surgery.

The two Trusts were commissioned following a comprehensive procurement exercise, which included international clinical expertise on the selection panel.

During the period between policy approval and full-service commissioning, interim arrangements were agreed to give children access to this pioneering surgery as there is only a limited window of opportunity for achieving an optimal clinical outcome.

Theo was one of the children to receive an ABI during the interim arrangements. His mother, Imelda described the impact:

> "After discovering Theo couldn't have a cochlea implant, all we could think about was how would he hear a fire alarm, how could we protect him from danger?
>
> It's now two years since Theo's device was activated and he can hear me calling him from upstairs. His first word was 'more' and his second was 'mummy' – something I never thought I would hear. Every day he uses his voice more and more and now loves to try and sing.
>
> We are eternally grateful to the surgical and audiology teams in Manchester who have given our little boy the ability to hear and speak."

https://www.england.nhs.uk/2019/04/nhs-england-to-fund-pioneering-new-brain-surgery-for-children-who-are-deaf/

Balloon pulmonary angioplasty

The balloon pulmonary angioplasty (BPA) service started at Royal Papworth Hospital NHS Foundation Trust in April 2018 as a treatment for patients with chronic thromboembolic pulmonary hypertension (CTEPH).

CTEPH is a rare complication of pulmonary embolism and a major cause of pulmonary hypertension (PH) leading to right heart failure and premature death.

Pulmonary thromboendarterectomy (PEA) has been commissioned as a highly specialised surgical intervention for CTEPH since 2000. Before this the only cure for CTEPH was a heart and lung or lung transplant. PEA removes the scar tissue blocking or narrowing the pulmonary arteries but about 40% of patients are unsuitable for this surgery because the blockages are too far into their lung vessels. Previously, for these technically inoperable patients, PH-targeted medical therapy was recommended.

BPA improves blood flow through scarred pulmonary arteries by stretching them open with a balloon that is inflated. The procedure is performed in stages and several sessions are usually required to achieve therapeutic benefit. The patient is awake during the 'keyhole' BPA technique. After considering the growing evidence

for BPA, NHS England agreed a commissioning policy: this treatment would initially only be available at Royal Papworth Hospital given its expertise in managing patients with CTEPH and their expertise in deciding whether patients are suitable for PEA.

The service treated 29 patients in its first year. Outcomes are being closely monitored and the team at Papworth, in collaboration with the European BPA Registry, plan to publish outcome data on the first 1,000 patients treated across Europe.

High consequence infectious diseases, special isolation unit (airborne)

In 2015/16, NHS England established the High Consequence Infectious Disease (HCID) Programme to ensure that there was an agreed approach to managing the end-to-end patient pathway for suspected and confirmed HCIDs. This ensures that a sustainable response is in place that can be efficiently and effectively actionable should the need arise.

HCIDs are highly transmissible infections and patients require careful management to prevent the staff caring for them from becoming infected. This service is required to be in a heightened state of preparedness.

The main reason for establishing the programme was the continuing threat of ‘airborne’ diseases, such as Middle-East respiratory syndrome (MERS) and avian flu, and, in particular, the need for a clear treatment pathway for such patients.

NHS England already had services in place for system readiness and for the treatment of patients with ‘contact’ diseases including viral haemorrhagic fevers such as Ebola, but no specific contracts for the treatment of patients with high consequence ‘airborne’ diseases.

The service was commissioned from four providers in 2018: two in the north (The Newcastle upon Tyne Hospitals NHS Foundation Trust and The Royal Liverpool and Broadgreen University Hospitals NHS Trust [now Liverpool University Hospitals NHS Foundation Trust]); and two in the south (Royal Free London NHS Foundation Trust and Guy’s and Thomas’ NHS Foundation Trust).

The providers work collectively to support one another in a network model that has improved the co-ordination and delivery of care.

Multiple sclerosis management service for children

In April 2018, a new highly specialised service providing a multiple sclerosis (MS) management service for children began. By systematising care provision across England, it facilitates prompt referral of children suspected of having MS or an ‘MS-like’ demyelinating condition, reducing the time to diagnosis and enabling prompt access to disease-modifying drugs. This will reduce the rate of avoidable admissions and relapses and improve overall clinical outcomes for patients.

NHS England identified a small number of suitable providers in England to serve the patient population within their local geographies. The centres needed to have an established cohort of patients and staff with the expertise to make assessments of patients and meet their needs. A provider selection process took place between September 2017 and January 2018.

There are now five lead hub centres, including one in the North of England which is a collaboration of three units. These centres treat patients under 18 years of age and provide expert management, advice and support to referring clinicians, and develop shared care plans with local providers.

Around 80 children have been newly diagnosed; this is in addition to around 230 children in England who had already been diagnosed with MS, or with an ‘MS-like’ recurrent acquired demyelinating condition, which requires the same treatment.

Proton beam therapy

Proton beam therapy is a type of radiotherapy that uses a beam of high energy protons, which are small parts of atoms, rather than high energy x-rays (called ‘photons’) to treat specific types of cancer.

Proton beam therapy enables a dose of high energy protons to be precisely targeted at a tumour, reducing the damage to surrounding healthy tissues and vital

organs which is an advantage in certain groups of patients or where the cancer is close to a critical part of the body such as the spinal cord.

Proton beam therapy is only suitable for certain types of cancer, such as highly complex brain, head and neck cancers and sarcomas as it does not lead to better outcomes for many cancer cases than using high energy x-rays, which is still considered the most appropriate and effective treatment for the majority of cancers.

Like high energy x-ray radiotherapy, proton beam therapy is painless, but patients may experience side effects similar to those experienced from other forms of radiotherapy.

The first NHS PBT centre opened and started treating patients at The Christie NHS Foundation Trust in Manchester in December 2018. This honoured a Prime Ministerial pledge made in 2011 that the NHS would provide PBT by 2018. A second NHS PBT centre will be provided at University College London Hospitals NHS Foundation Trust.

Since 2008, almost 1,500 NHS patients have been referred overseas for PBT, the majority to providers in the USA. However, some have not been able to travel to the USA for clinical or social reasons. Therefore, in 2017, it was decided to procure another PBT provider closer to the UK. Three PBT centres entered formal bids. A team from NHS England made up of clinicians and managers visited each of them and a provider in Germany was chosen. This procurement won the 2018 Chartered Institute of Procurement and Supply (CIPS) Supply Management Award in the ‘International Procurement Project’ category. In addition, Arden and Gem CSU with NHS England were ‘highly commended’ at HSJ Value Awards in 2019 for increasing access to Proton Beam Therapy treatment to cancer patients.

The NHS PBT service is underpinned by a number of key documents, including a service specification, several clinical commissioning policies and a standard operating procedure.

3. Services and providers of highly specialised services for 2018/19

Alkaptonuria service (adults)

Alkaptonuria (AKU) is a rare inherited disorder that causes considerable morbidity in the peak of adulthood due to severe premature destruction of the joints and spine. Disability, often severe, is the norm for those over 30 years of age. There are around 50 people in England with AKU.

The service provides an inpatient-based assessment service for patients with AKU where patients are reviewed annually. It provides one-stop care to: assess and detect disease complications; prescribe and monitor drugs to arrest the progression of the disease; and formulate shared care management plans with local providers.

NHS centre	The Royal Liverpool and Broadgreen University Hospitals NHS Trust [now Liverpool University Hospitals NHS Foundation Trust]
Expenditure	Between £0.5 million and £1 million
Caseload	59
Outcomes collated	• Median quality of life (SF36) score for patients treated in the service for 12 months or longer: 50% SF36 stable • Median AKU severity score index measurement (AKUSSI) for patients treated in the service for 12 months or longer: 93% AKUSSI improved, ochronosis scores reversed • Note: ochronosis is a condition in which the body cannot break down the toxic acid homogentisic acid. This causes bones and cartilage to become black and brittle. The spine collapses and prostate and kidney stones appear. Heart valves become blocked and the patient needs heart surgery.

Geographical equity access	There is some evidence of geographical inequity so further analysis will be undertaken along with ongoing discussions with the service.

Alström syndrome service (adults and children)

Alström is a rare genetic syndrome that usually presents with blindness in childhood. Patients go on to develop insulin-resistant diabetes, fibrosing cardiomyopathy (where abnormal tissue growing in the heart stops it working effectively) and renal failure. They may also become Deaf. Fewer than 100 people are thought to be affected by Alström syndrome in England.

Both the adult and paediatric services run two-day clinics that undertake assessment of all patients in a multidisciplinary structure. Patients are assessed and reviewed by all the specialties appropriate to their needs during the clinic.

A management plan is agreed and communicated to local care providers to allow their healthcare professionals to implement the recommendations and monitor patients' progress. Alström Syndrome UK support workers attend the clinic to provide advocacy and guidance on the social care aspects of living with the condition.

NHS centres	Birmingham Children's Hospital NHS Foundation Trust [now Birmingham Women's and Children's Hospital NHS Foundation Trust] University Hospitals Birmingham NHS Foundation Trust
Expenditure	<£0.5 million
Caseload	85
Outcomes collated	• % of children in HbA1c target range: – Birmingham Children's Hospital 8% • % of adults with HbA1c <75 mmol/mol: – University Hospitals Birmingham 76% • Median age at death of patients on active caseload: • University Hospitals Birmingham: no deaths in 2018/19

	Note: glycated haemoglobin (HbA1c) is measured primarily to identify the 3-month average plasma glucose concentration.
Geographical equity access	Numbers too small to analyse.

Ataxia telangiectasia services for adults

Ataxia telangiectasia (AT) is a rare, neurodegenerative and progressive condition that starts in early childhood causing severe disability and premature death. It affects many parts of the body and a wheelchair is often needed by the age of 10. The average life expectancy is 25 years. During the adult stage of the condition, there is increased susceptibility to leukaemia, lymphoma, pneumonia, chronic lung disease and neurological decline. Fewer than 100 adults in England have AT.

The service undertakes annual multidisciplinary inpatient assessment for all diagnosed adult AT patients. This comprises a CT scan, video fluoroscopy, pulmonary function testing, sleep studies, brain imaging, neurophysiology and immunological blood testing. Following this review, a management plan for local care providers is agreed and communicated to allow the local healthcare professionals to implement the recommendations and monitor their progress.

NHS centre	Papworth Hospital NHS Foundation Trust [now Royal Papworth Hospital NHS Foundation Trust]
Expenditure	<£0.5 million
Caseload	80
Outcomes collated	• Median age at death: 40.5 years (median cited in medical publications is 20 years) • Median BMI: 22.1 interquartile range 18.4-25.9 kg/m^2 (an important measure because patients with the condition often do not achieve optimum BMIs).
Geographical equity access	There is some evidence of geographical inequity so further analysis will be undertaken along with ongoing discussions with the service

Ataxia telangiectasia services for children

Ataxia telangiectasia (AT) is a rare, neurodegenerative and progressive condition that starts in early childhood causing severe disability and premature death. It affects many parts of the body and a wheelchair is often needed by the age of 10. The average life expectancy is 25 years. Fewer than 150 children in England have AT.

This service provides outpatient clinics to patients with AT, which take place over two days with a multidisciplinary team of experts. Following this review, a management plan for local care providers is agreed and communicated to allow the local healthcare professionals to implement the recommendations and monitor their progress.

NHS centre	Nottingham University Hospitals NHS Trust
Expenditure	<£0.5 million
Caseload	132
Outcomes collated	• % of patients with previously unrecognised treatable or untreatable morbidity: 93% • % of patients for whom active intervention was undertaken in clinic or arranged locally: 95%
Geographical equity access	No evidence of geographical inequity

Atypical haemolytic uraemic syndrome (adults and children)

Atypical haemolytic uraemic syndrome (aHUS) can occur at any age. Onset in childhood is slightly more common than in adulthood (around 60% and 40% of all cases respectively). Most children (70%) who develop aHUS experience the disease for the first time before the age of two years. Worldwide, the prevalence of aHUS ranges from 2.7 to 5.5 per million population, with an incidence of about 0.40 per million population.

The aim of the service is to provide a national diagnostic and management advice for patients with aHUS. The service offers comprehensive diagnostic clinical and

pathological investigations and expert opinion, facilitating optimal patient management on a shared-care basis with referring clinicians and other specialist services.

NHS centre	The Newcastle upon Tyne Hospitals NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Caseload	154
Outcomes collated	• Number of deaths in patients with a diagnosis of complement mediated aHUS: no patient in England died of aHUS in 2018/19
Geographical equity access	Geographical analysis to be undertaken during 2020

Auditory brainstem implant for children with congenital abnormality of the auditory nerves or cochleae

The auditory brainstem (ABI) service is commissioned to provide services for children under the age of five years with no functional hearing as a result of congenital abnormalities affecting the auditory nerves or the cochleae, which renders them unable to gain adequate benefit from conventional well-fitted hearing aids or cochlear implants.

The service includes multidisciplinary assessment, surgical implantation and rehabilitation (including maintenance of the implant).

NHS centres	Manchester University NHS Foundation Trust Guy's and St Thomas' NHS Foundation Trust
Expenditure	New service so only available from next year
Surgical operations	Data suppressed to maintain patient confidentiality
Outcomes collated	Data suppressed to maintain patient confidentiality

Geographical equity access	Geographical analysis to be undertaken during 2020

Autologous intestinal reconstruction service for adults

Adult patients in the UK with chronic intestinal failure usually receive home parenteral nutrition (HPN). Autologous intestinal reconstruction in adults (AuGIR) is a surgical procedure in adult patients with short bowel syndromes who are on parenteral nutrition. Patients have insufficient bowel to take in enough food by mouth to provide adequate nutrition. The aim of the service is to employ surgical techniques for autologous intestinal reconstruction (from the patient's own intestine) and lengthening. If successful, this treatment allows the patient to gain nutritional autonomy and thus cease to require, or have a reduced requirement for, HPN. This is an established procedure in children.

NHS centre	Salford Royal NHS Foundation Trust
Expenditure	<£0.5 million
Caseload	Data suppressed to maintain patient confidentiality
Outcomes collated	Data suppressed to maintain patient confidentiality
Geographical equity access	Numbers too small to analyse

Bardet-Biedl syndrome service (adults and children)

Bardet-Biedl syndrome is a highly debilitating autosomal-recessive genetic disorder that causes early-onset blindness, renal failure, obesity, diabetes, Hirschsprung disease, urological problems and neurological deficits. About 1 in 100,000 babies are born each year with Bardet-Biedl syndrome, i.e. five or six each year in England.

Both the adult and paediatric services run dedicated clinics that undertake assessment of all patients in a multidisciplinary structure. Patients are assessed and reviewed by all the specialities appropriate to their needs during the clinic.

Following this review, a management plan for local care providers is agreed and communicated to allow the local healthcare professionals to implement the recommendations and monitor their progress. Bardet-Biedl Syndrome UK co-ordinates the clinics at the centres and provides advocacy and support to patients attending the clinics.

NHS centres	Birmingham Children's Hospital NHS Foundation Trust [now Birmingham Women's and Children's Hospital NHS Foundation Trust] Great Ormond Street Hospital for Children NHS Foundation Trust Guy's and St Thomas' NHS Foundation Trust University Hospitals Birmingham NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Assessments	325
Outcomes collated	• % of children with HbA1c <48 mmol/mol: – Birmingham Women's and Children's Hospital: 90% – Great Ormond Street Hospital: 78% – Guy's and St Thomas' 77% – % of adult patients with HbA1c <75 mmol/mol: – University Hospitals Birmingham: 22.5% – Great Ormond Street Hospital: 83% – Guy's and St Thomas': 83% • % of adult patients with a BMI <35: – University Hospitals Birmingham: 96% – Guy's and St Thomas': 56% Note: Glycated haemoglobin (HbA1c) is a form of haemoglobin that is measured primarily to identify the 3-month average plasma glucose concentration.
Geographical equity access	Data not available or not comparable

First Bardet-Biedl ‘virtual’ telemedicine clinic

During 2017, a business case was approved to provide innovative new telemedicine clinics in all four centres (supported by the stakeholder group BBS UK). This has proved successful. BWCH issued the following press release:

Two brothers with a rare genetic condition have become the first to benefit from a virtual telemedicine clinic at our Children’s Hospital.

The brothers aged 15 and 11, who both have Bardet-Biedl syndrome, recently ‘attended’ their consultation appointment with our expert multidisciplinary team from the comfort of their home in Wednesbury thanks to the use of the digital technology.

Their condition, which affects 1 in 300,000 people, causes a number of issues including retinal dystrophy (blindness), sensorineural deafness, infancy onset obesity, cystic kidneys, and development and behaviour issues. All this means that travelling to Birmingham from the family’s Black Country home can be difficult.

To avoid attending multiple appointments, the NHS England Bardet-Biedl multidisciplinary clinic set up a national service, delivered at our Trust in partnership with University Hospitals Birmingham, Great Ormond Street Hospital and Guy's and Thomas' to deliver combined clinics for young people and adults in optometry, ophthalmology, diabetes, endocrinology, nephrology, psychology, dietetics, genetics, and transitional care. This is a ‘one stop shop’, where families can see all the specialists in one visit.

However, as this clinic runs just eight times a year, our consultants worked with NHS commissioners and staff in our Waterfall House Rare Disease Centre to put in place our hospital’s first telemedicine clinic of its type for patients.

Through Zoom video conferencing, set up by our IT team, they were able to link with the whole family via an iPad on their kitchen table. Mum felt the virtual clinic brought real benefit. She said:

> *“‘The brothers attend a clinic, which is a whole day of appointments and it can be stressful for them and a tiring day for me.*
>
> *I felt the telemedicine clinic was really good for us as a family. We got as much done as we would have in clinic but in the comfort in our home. The boys loved it and they were also able to do some of it independently, which was brilliant.*

"We are so lucky to have such a great team of specialists and we'd like to say thanks to all involved."

One of our team that helped put the clinic in place was Professor Timothy Barrett, Consultant Paediatric Endocrinology and Diabetes at BWC. He said:

"The clinic was a huge success. It was delivered with specialists in endocrinology, diabetes, and dietetics, along with Ms Amy Clapp from the family support group BBS-UK.

Thanks to our NHS commissioners, and with great support from our fantastic IT department, we were able to see and hear the boys and their mum, which helped us have important discussions around their medical, emotional and educational needs.

This clinic model does not replace face to face appointments but does offer an option for families who find travelling to Birmingham difficult. It also has to potential to address NHS waiting times for outpatient appointments."

Barth syndrome service (male adults and children)

Barth syndrome is an X-linked disorder of lipid metabolism presenting as cardiac/skeletal myopathy, neutropenia (reduced white blood cell count leading to susceptibility to infection) and growth retardation and has with a high infant mortality rate. Patients present with frequent cardiac problems and, in two-thirds, neutropenia. When undiagnosed or treated by non-specialists, patients typically experience frequent hospital admissions for a range of diagnostic tests and treatment of severe infections. Infections are significantly reduced through protocol-driven prescription of granulocyte colony stimulating factor (G-CSF). About 30 people in England have Barth syndrome.

The service provides diagnostic testing, which includes cardiolipin (a lipid essential for the optimal functioning of enzymes involved in energy metabolism) testing and genetic testing. It also provides post-mortem cardiolipin testing and familial gene testing. Care is provided through a multidisciplinary team that: monitors cardiac function and other co-morbid factors; prescribes appropriate drugs; and develops management plans with local healthcare providers.

NHS centre	University Hospitals Bristol NHS Foundation Trust

Expenditure	>£0.5 million but <£1 million
Caseload	Data suppressed to maintain patient confidentiality
Outcomes collated	• Median age at diagnosis: one diagnosis made prenatally • Median age at death: no deaths
Geographical equity access	Numbers too small to analyse

Beckwith-Wiedemann syndrome with macroglossia service (children)

Beckwith-Wiedemann syndrome is a disorder present at birth, characterised by an increased risk of childhood cancer and certain congenital features. One of the congenital features is macroglossia (significant enlargement of the tongue), which causes: drooling; feeding, speech, orthodontic and dental problems; and devastating psychosocial consequences. About 1 in 15,000 babies are born each year with Beckwith-Wiedemann syndrome but only about half have macroglossia (about 15-20 babies each year).

The service provides multidisciplinary, centralised, expert clinical care for pre-operative assessment, surgical management and post-operative rehabilitation of this group of patients, including access to support and advice on the functional problems associated with the macroglossia.

NHS centre	Great Ormond Street Hospital for Children NHS Foundation Trust
Expenditure	<£0.5 million
Caseload	189
Outcomes collated	• % with improvement at the 3-6-month postoperative assessment of resting tongue position: 100% • % with improvement (reduction or cessation) at the 3-–6-month postoperative assessment of drooling: 100% • % with improvement (reduction or elimination) at the 3-6-month postoperative assessment of macroglossia-related errors: 100%

Geographical equity access	Numbers too small to analyse

Beckwith Wiedermann syndrome service telemedicine clinic piloted

In the Beckwith Wiedemann syndrome service, a telemedicine clinic was piloted for post-operative reviews of patients once they had been discharged back home. Feedback has been obtained systematically from families following these appointments which has been consistently positive.

Behçet’s syndrome service (adults and adolescents)

Behçet's syndrome is a chronic, inflammatory, multisystemic vasculitic disorder with a wide spectrum of clinical presentations that may include blindness, severe ulceration and cardiovascular problems. There are around 1,700 people in England that have Behçet's syndrome. The aim of the service is to ensure that patients of all ages suffering from Behçet’s syndrome can access timely definitive diagnosis, or exclusion, of Behçet’s syndrome and receive optimal treatment equitably across the country, usually in local centres.

NHS centres	Aintree University Hospital NHS Foundation Trust [now Liverpool University Hospitals NHS Foundation Trust] Barts Health NHS Trust Sandwell and West Birmingham Hospitals NHS Trust
Expenditure	>£1 million but <£5 million
Caseload	1,697
Outcomes collated	• Median number of flares per patient during the previous 12 months: – Aintree: 1 – Barts Health: 0.5 – Sandwell and West Birmingham: 1
Geographical equity access	Evidence of geographical inequity is being investigated, but the explanation is likely to be data quality issues

Bladder exstrophy service (children)

The service provides diagnosis, management advice and treatment for children with bladder exstrophy, primary epispadias, cloacal exstrophy and all variants. Expert management and appropriate surgical reconstruction can provide a child suffering from bladder exstrophy with near normal lifestyle. The goals of exstrophy reconstruction are:

- anatomic reconstruction of the bladder/urethra, bony pelvis, abdominal wall and external genitalia
- creation of urinary continence with preservation of renal function
- healthy psychological adjustment and adaptation to the condition throughout life
- support during adolescence.

Between 1 in 30,000 and 1 in 50,000 babies are born each year with bladder exstrophy, i.e. around 20 babies each year in England.

The service is provided by a multidisciplinary team including dedicated psychologists, clinical nurse specialists, input from nephrology and urodynamics and a specialist urology ward. One of the centres provides dedicated orthopaedic surgical input to address bony pelvis abnormalities.

NHS centres	Great Ormond Street Hospital for Children NHS Foundation Trust Manchester University NHS Foundation Trust
Expenditure	>£1 million but <£5 million
New babies	Data suppressed to maintain patient confidentiality
Outcomes collated	• % closure achieved without dehiscence: – Great Ormond Street: 93% – Manchester University: 100%
Geographical equity access	Data not available or not comparable

Breast radiotherapy injury rehabilitation service (a discrete cohort of adult females)

This service is for a discrete cohort of women who have severe, chronic and complex conditions arising from radiation-induced injuries. The women received a treatment regime for breast cancer in the 1970s and 1980s that is now known to be associated with a particular risk of damage to the nerves of the brachial plexus.

The service provides a specialist, multidisciplinary rehabilitation service. The lead centre provides an inpatient service.

NHS centre	Royal National Hospital for Rheumatic Diseases – Royal United Hospitals Bath NHS Foundation Trust
Expenditure	<£0.5 million
Caseload	65
Outcomes collated	• % Brief Pain Inventory Score: 13% at 3 months and 58% at 12 months • % Acceptance & Action Questionnaire Score: 2% at 3 months and 9% at 12 months Note: These are good scores for patients who have been treated for severe health problems.
Geographical equity access	Data not available or not comparable

Cardiothoracic transplantation service (paediatric)

The service provides a comprehensive transplantation service for referred infants and children who have not responded to maximum conventional treatment for cardiac or respiratory failure and who are therefore candidates for transplantation.

The service integrates seamlessly with services for heart failure, cystic fibrosis/respiratory medicine and pulmonary hypertension. It is closely integrated with the Ventricular Assist Devices (VADs) for Children as a Bridge to Heart Transplant service.

The demand for cardiothoracic transplant exceeds the supply of organs. Patients are listed for a heart transplant if they have no contraindications and this is likely to improve their quality of life and survival. Clinical outcomes are monitored by NHS England in collaboration with NHS Blood and Transplant. International benchmarking ensures that immunosuppression and surveillance are consistent with the best management internationally.

NHS centres	Great Ormond Street Hospital for Children NHS Foundation Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust
Expenditure	>£50 million (adults and children, heart and lung)
Number of transplants	26
Outcomes collated	• 30-day unadjusted patient survival rate after first paediatric heart only transplant: – Great Ormond Street: 98.3% – Newcastle upon Tyne: 92.3% • 1-year unadjusted patient survival rate after first paediatric heart only transplant: – Great Ormond Street: 96.7% – Newcastle upon Tyne: 87.8% • 5-year unadjusted patient survival after first paediatric heart only transplant: – Great Ormond Street: 82.8% – Newcastle upon Tyne: 83.1% • 90-day patient survival rate after first paediatric lung transplant: – Great Ormond Street: 88.5% – Newcastle upon Tyne: survival rates for groups with <30 patients are not presented due to small numbers
Geographical equity access	Data not available or not comparable

Choriocarcinoma service (adults and adolescents)

This service diagnoses and treats women with the different types of gestational trophoblastic disease including the following:

- Hydatidiform mole (also known as molar pregnancy): in this condition, the sperm and egg cells join together but a healthy fetus does not develop. The placenta grows to an abnormal size, requiring surgical evacuation of the uterus.
- Choriocarcinoma, which is an aggressive and malignant cancer that may spread from the uterus to other organs in the body, such as the lungs or brain. Each year about 10 women in England develop choriocarcinoma.
- Placental site trophoblastic tumour, a rare variant of choriocarcinoma. This cancer is able to spread through the body via the lymphatic system.

The service provides monitoring for all women who have a molar pregnancy through the regular measurement of hCG (human chorionic gonadotrophin). For those women who go on to develop gestational trophoblastic disease, the service provides a full inpatient and outpatient management service to treat the cancer.

NHS centres	Imperial College Healthcare NHS Trust Sheffield Teaching Hospitals NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Inpatient episodes	547
Outcomes collated	• Deaths as % of new cases each year: – Imperial College: 0 – Sheffield Teaching: 0
Geographical equity access	Data required from cancer register

Chronic pulmonary aspergillosis service (adults)

Chronic pulmonary aspergillosis (CPA) is a chronic, progressive infection of the lung with the fungus *Aspergillus fumigatus* that follows a lung insult (typically

sarcoidosis, atypical TB or recurrent pneumothoraces) and occurs in those with one or more innate genetic defects. The service is an assessment and long-term clinical management service for CPA. It diagnoses patients referred by appropriate hospital consultants with probable chronic aspergillus infection and classifies the specific nature of any detected aspergillus infection. Those patients confirmed to have CPA within the parameters of this specification are offered clinically appropriate treatment options.

NHS centre	Manchester University NHS Foundation Trust
Expenditure	>£5 million but <£10 million
Caseload	517
Outcomes collated	• % of patients showing a ≥12-point improvement in the St George's Respiratory Questionnaire (SGRQ) and ≥3 kg weight gain: 52% Note: This is a substantial improvement in quality of life in those with a condition that does not improve at all without treatment.
Geographical equity access	Strong evidence of long-standing geographical inequity; commissioning solutions are being explored.

Complex childhood osteogenesis imperfecta service

Osteogenesis imperfecta (OI) is a genetic condition characterised by bones that break easily, often from little or no apparent cause. The condition can vary quite significantly from one person to another: a person can have just a few or as many as several hundred fractures in a lifetime. About 300 children in England have severe or complex OI.

The service provides care for children whose OI meets a service definition of 'severe', 'atypical' or 'complex'. The service brings together surgery (opinion only), pharmacology, physiotherapy, occupational therapy, nursing and social work into a network model that aims to improve the diagnosis and management of under 16s who have this rare, genetic collagen deficiency.

NHS centres	Birmingham Children's Hospital NHS Foundation Trust {now Birmingham Women's and Children's Hospital NHS Foundation Trust Great Ormond Street Hospital for Children NHS Foundation Trust Sheffield Children's NHS Foundation Trust University Hospitals Bristol NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Caseload	306
Outcomes collated	• Median number of new non-vertebral fractures: – Birmingham Women's and Children's: 0 – Great Ormond Street: 0 – University Hospitals Bristol: 0 – Sheffield Children's: 0 • Median number of new vertebral fractures: – Birmingham Women's and Children's: 0 – Great Ormond Street: 0 – University Hospitals Bristol: 0 – Sheffield Children's: 0 • % patients with scoliosis and Cobb angle >45 degrees (the Cobb angle measures the degree of abnormal lateral spinal curvature): – Birmingham and Women's Children's: 4% – Great Ormond Street: 5% – University Hospitals Bristol: 0% – Sheffield Children's: 2%
Geographical equity access	No evidence of geographical inequity

Osteogenesis imperfecta care planning

In the OI service, the four centres work collaboratively to streamline current medical treatments as well as offering patients' opportunities to access innovative medical

treatments through enrolment in research studies. Through care planning, there is support for patients to be cared for in the local setting and Clinical Nurse Specialists support and provide education materials for school staff to enable children to access education.

Complex Ehlers-Danlos syndrome service (adults and children)

Ehlers-Danlos syndrome (EDS) is a group of heritable disorders of connective tissue. The main clinical features are hyperextensible skin, hypermobile joints and tissue fragility. In severe cases, patients can have life-threatening complications such as aortic dissection, where the layers of the aorta wall aorta separate. Each of the types of EDS has its own specific management.

The fully comprehensive service (under the auspices of the clinical genetics service) gives patients a precise clinical diagnosis and manages the subset in whom clinical diagnosis is not straight forward or diagnosis through laboratory testing needs to be confirmed with further clinical evaluation.

NHS centres	London North West University Healthcare NHS Trust Sheffield Children's NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Definitive diagnosis	EDS confirmed in 96 patients
Outcomes collated	• % patients with a definitive diagnosis or diagnosis ruled out: – London North West: 37% of patients with definitive EDS diagnosis and EDS ruled out in 60% – Sheffield Teaching: diagnosis or diagnosis ruled out 98% • % of patients with a genetic diagnosis: – London North West: 29% – Sheffield Teaching: 43%
Geographical equity access	Data not available or not comparable

Complex neurofibromatosis type I service (adults and children)

Neurofibromatosis type 1 (NF1) is an inherited genetic disorder characterised by the formation of neurofibromas (tumours involving nerve tissue) in the skin, subcutaneous tissue, cranial nerves and spinal root nerves. About 1 in 25,000 of the population has NF1. Those with complex NF1 have a high risk of developing rare complications, which may affect most of the body systems.

Complex NF1 is defined by the presence of these other conditions that can cause significant morbidity and mortality and which require integrated management by an expert team.

The service includes:

- specialist assessment of patients with suspected NFI and complex complications of the disease, to provide accurate diagnosis of unusual phenotypes and other diseases that can be mistaken for NF1. This is through genetic testing with support from genetic counselling
- co-ordination of care by a specialist multidisciplinary team (when NF1 complications mean the condition manifests differently from the usual clinical picture)
- monitoring the risk of NF1-related malignancy and tumour progression
- long-term monitoring to evaluate the need for surgery, e.g. Cervical cord compression.

NHS centres	Guy's and St Thomas' NHS Foundation Trust Manchester University NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Outpatient attendances	1,004
Outcomes collated	• Total number of interventions facilitated: – Guy's and St Thomas': 217 interventions in 445 patients – Manchester University: 377 interventions out of a total of 353 patients

Geographical equity access	There is evidence of geographical inequity and the establishment of a new shared care clinic should help resolve this issue

Support for NF1 patients

In the two national centres, clinical psychologist offers weekly clinics and telephone support to help illness adjustment and anxiety in adults with NF1. There are also neuro-disability paediatricians who reduce the paediatric waiting list and focus on children with chronic neurological problems.

There are weekly phone clinics held by Clinical Nurse Specialists to support complex NF1 patients in between clinic visits, lessening the need for people to travel to face to face clinics, helping people who live a long distance from the centres to access care and review locally and triaging sick patients for urgent assessment to the centres.

Complex tracheal disease service (children)

The complex tracheal disease service assesses and treats children with severe and rare conditions affecting the trachea (long segment tracheal stenosis). Patient selection is particularly complex.

A range of surgical procedures is offered, including slide tracheoplasty.

About 60 babies and children are referred to the service each year for assessment.

NHS centre	Great Ormond Street Hospital for Children NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Inpatient episodes	78
Outcomes collated	• 1-year survival: 98%
Geographical equity access	Numbers too small to analyse

Congenital hyperinsulinism service (children)

Congenital hyperinsulinism (CHI) is a condition characterised by excess insulin production, resulting in hypoglycaemia. The clinical presentation and progress of CHI lie on a spectrum, varying from those with transient hypoglycaemia to those unresponsive to medical treatment and requiring pancreatectomy. In the absence of expert management, children may show development delay because of brain injury in infancy from prolonged or recurrent hyperinsulinaemic hypoglycaemia.

The service diagnoses patients (usually in the new-born period) and refers them to one of the national centres. If immediate transfer cannot be arranged, then the national centre supports the referring unit to provide appropriate care for the patient. The national centre may also require the referring hospital to carry out investigations to confirm the diagnosis of CHI. The service liaises and works with a surgical team to manage those children whose condition and response to medical management indicates that surgery is a viable option.

NHS centres	Great Ormond Street Hospital for Children NHS Foundation Trust Manchester University NHS Foundation Trust and Alder Hey Children's NHS Foundation Trust, which together form 'NORCHI'
Expenditure	>£1 million but <£5 million
Caseload	1,083
Outcomes collated	• Incidence of deaths in patients with CHI as a consequence of CHI: – Great Ormond Street: 0 – NORCHI: 0 • Unplanned admissions due to CHI and admitted under the paediatric endocrine team, day cases excluded: – Great Ormond Street: 16 – NORCHI: 28
Geographical equity access	Data not available or not comparable

CHI holds joint multi-disciplinary team (MDT) videoconferencing meetings

In the congenital hyperinsulinism service, the two centres work together as a single team and have introduced joint MDT videoconference meetings to review the care of patients and ensure systematic reviews of patients. The service is also developing a prototype App for CHI patients and is using continuous glucose monitoring for patients with hypoglycaemia. The service's Clinical Nurse Specialists support school staff to ensure children are safe in the school setting.

Craniofacial service (adults and children)

This service provides assessment, surgical and non-surgical treatment, and follow-up of patients with severe congenital deformities of the skull and face.

NHS centres	Alder Hey Children's NHS Foundation Trust Birmingham Children's Hospital NHS Foundation Trust [now Birmingham Women's and Children's Hospital NHS Foundation Trust] Great Ormond Street Hospital for Children NHS Foundation Trust Oxford University Hospitals NHS Trust
Expenditure	>£10 million but <£20 million
Inpatient episodes	420
Outcomes collated	• Proportion of patients with level 4 surgical complications (should decrease or remain the same): – Alder Hey Children's: 1% – Birmingham Children's: 0% – Great Ormond Street: 0% – Oxford University: 1%
Geographical equity access	Data not available or not comparable

Clinical nurses liaise with local schools

Children who are diagnosed with one of the craniofacial conditions have a care plan set out for them and the service's Clinical Nurse Specialists liaise with local schools to make sure that children are assessed for and receive additional support depending on their changing needs.

Cryopyrin associated periodic syndrome service (adults)

Cryopyrin associated periodic syndrome (CAPS) is a very rare, lifelong inflammatory disease that interferes with growth and development that causes serious morbidity and is often fatal.

The service assesses patients and makes or confirms a diagnosis; drug treatment may be appropriate.

NHS centre	Royal Free London NHS Foundation Trust
Expenditure	>£5 million but <£10 million
Patients on high cost drugs	147
Outcomes collated	• Median 20-point CAPS activity score (a low CAPS score indicates symptom control): 2/20
Geographical equity access	Data not available or not comparable

Diagnostic service for amyloidosis (adults and children)

Amyloidosis is a condition in which abnormal protein deposits accumulate in many different organs.

The National Amyloidosis Centre provides diagnostic imaging (SAP scintigraphy – a technique for identifying amyloid deposits – and specialist echocardiography), histology and DNA analysis, genetic counselling, monitoring of amyloid proteins in

the blood, recommendations for treatment, and supports the evaluation of existing and new therapies.

The service provides a diagnostic service to about 1,400 new patients each year.

The centre's role is expanding as more therapies are developed to treat amyloidosis.

NHS centre	Royal Free London NHS Foundation Trust
Expenditure	>£5 million but <£10 million
First evaluations	1,435
Outcomes collated	• % of patients with a definitive diagnosis or diagnosis ruled out: 99%
Geographical equity access	Data not available or not comparable

Diagnostic service for primary ciliary dyskinesia (adults and children)

Primary ciliary dyskinesia is a genetic disorder of the air tubes of the lungs (the bronchi), which become infected and filled with pus due to abnormalities of the hair-like structure (cilia) of the cells lining the respiratory tract. This can lead to repeated infections and damage the lung, especially if the diagnosis is delayed. Around 100 children are diagnosed with PCD each year in England.

This service provides a diagnostic and advice service to patients who are referred with suspected PCD. It also supports and trains them in certain aspects of self-care treatment.

NHS centres	Royal Brompton & Harefield NHS Foundation Trust University Hospitals Southampton NHS Foundation Trust University Hospitals of Leicester NHS Trust

Expenditure	>£0.5 million but <£1 million (for management and diagnostic elements)
Number of positive samples	75
Outcomes collated	Outcomes captured by PCD management service
Geographical equity access	Data not available or not comparable

Support for patients with primary ciliary dyskinesia

Patients' GPs are sent comprehensive outcome letters following clinic review – the delivery of any requests for local clinical actions such as further local testing and review is followed up by the service's nursing staff to make sure that patients receive the management of care that is required. Patient's families have a direct phone number to the service's nursing staff to discuss any issues or concerns.

Diagnostic service for rare neuromuscular disorders (adults and children)

The aim of the service is to make a precise molecular or clinical diagnosis in patients with four rare neuromuscular conditions and to assess fully the extent of their disease:

- limb girdle muscular dystrophies
- congenital muscular dystrophies
- congenital myasthenic syndromes
- muscle channelopathies (also known as periodic paralysis).

This service provides a diagnostic, advisory and clinical service for patients with four groups of very rare inherited neuromuscular disorders. These conditions are all inherited, and the definitive diagnosis for a patient is made by identifying the primary gene defect. Each disease group involves multiple genes, and the decision as to which gene to search first for DNA mutations is arrived at by using a disease-

specific battery of techniques. These may include detailed clinical assessments, specialist neurophysiological tests, and immunological analyses on tissue biopsies.

NHS centres	Great Ormond Street Hospital for Children NHS Foundation Trust University College London Hospitals NHS Foundation Trust Oxford University Hospitals NHS Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust
Expenditure	>£5 million but <£10 million
Number of referrals	1,424
Outcomes collated	• % of patients with a genetic diagnosis: – Great Ormond Street Hospital: 25% – University College London: 64% – Oxford University: 68% – Newcastle upon Tyne: 41% Note: Centres diagnose and assess different conditions, so outcome measures vary.
Geographical equity access	No evidence of geographical inequity

Encapsulating peritoneal sclerosis treatment service (adults)

The encapsulating peritoneal sclerosis surgical service (EPS SS) provides surgical treatment for encapsulating peritoneal sclerosis (EPS). EPS, also referred to as sclerosing peritonitis, is a complication arising from long term use of peritoneal dialysis. EPS is characterised by marked sclerotic thickening of the peritoneal membrane, leading to encapsulation of the gut and sub-acute or acute bowel obstruction. As a chronic fibrosing process, it leads to abdominal pain, nausea, vomiting, weight loss, fever, malnutrition, anaemia, ascites and finally surgical peritonitis and mortality. EPS is a condition associated with significant morbidity and mortality and with poor outcomes if not recognised early and treated. With centralising treatment in specified national centres, experience has been consolidated leading to better patient outcomes, mirroring the best international experience.

NHS centres	Cambridge University Hospitals NHS Foundation Trust Manchester University NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Primary surgical procedures	Data suppressed to maintain patient confidentiality
Outcomes collated	• 1-year survival post operation for renal cases across the centres: 93% • % TPN free post operation across the centres: 100%
Geographical equity access	No evidence of geographical inequity

Epidermolysis bullosa service (adults and children)

Epidermolysis bullosa (EB) is the name given to a group of rare inherited disorders that cause lifelong blistering and ulceration of the skin and often the mucous membranes. Blistering is almost always apparent at or soon after birth, but the severity of the condition varies greatly, depending on the type of Epidermolysis bullosa present. The national EB service aims to provide diagnosis and assessment of infants, children, adolescents and adults with suspected or known EB, along with treatment and long-term support.

NHS centres	Birmingham Children's Hospital NHS Foundation Trust [now Birmingham Women's and Children's Hospital NHS Foundation Trust] Great Ormond Street Hospital for Children NHS Foundation Trust Guy's and St Thomas' NHS Foundation Trust Heart of England NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Caseload	509 severe cases (based on three centres)
Outcomes collated	• Median quality of life score at transition (adults only) (QoLEB): – Birmingham Women's and Children's: 12

	– Guy's and St Thomas': 12.4 • Proportion of unplanned admissions among patients with recessive dystrophic EB: – Birmingham Women's and Children's and Heart of England combined: 21.5% – Great Ormond Street: 35% – Guy's and St Thomas': 35% Note: Recessive dystrophic EB is the most severe type.
Geographical equity access	Data not available or not comparable

Extracorporeal membrane oxygenation service for adults

Extracorporeal membrane oxygenation (ECMO) supports adults with severe potentially reversible acute respiratory failure by oxygenating the blood through an artificial lung machine.

The specialist centres function as a national network, working closely with their local critical care networks. All centres provide a retrieval service that includes the capability to undertake 'mobile' ECMO when this is deemed clinically necessary. The service assesses about 1,000 patients for treatment each year and about 300 of these are treated with ECMO.

NHS centres	Guy's and St Thomas' NHS Foundation Trust Manchester University NHS Foundation Trust Papworth Hospital NHS Foundation Trust [now Royal Papworth Hospital NHS Foundation Trust] Royal Brompton & Harefield NHS Foundation Trust University Hospitals of Leicester NHS Trust
Expenditure	>£20 million but <£30 million (adults and children)
Starting treatment	332

Outcomes collated	• % survival at discharge: – Guy's and St Thomas': 71% – Manchester University: Data not available – Papworth: 76% – Royal Brompton & Harefield: 81% – Leicester: 82%
Geographical equity access	There is some evidence of geographical inequity in the service. The issues are understood, the inequity continues to be monitored and further analysis is anticipated in 2019/20

Extracorporeal membrane oxygenation service for neonates, infants and children with respiratory failure

Extracorporeal membrane oxygenation (ECMO) supports critically ill babies and children who have severe, potentially reversible acute respiratory failure by oxygenating their blood through an artificial lung machine.

NHS centres	Alder Hey Children's NHS Foundation Trust Birmingham Women's and Children's Hospital NHS Foundation Trust Great Ormond Street Hospital for Children NHS Foundation Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust University Hospitals of Leicester NHS Trust
Expenditure	>£0.5 million but <£1 million (adults and children)
Starting treatment	56
Outcomes collated	Data suppressed to maintain patient confidentiality
Geographical equity access	No evidence of geographical inequity

Ex-vivo partial nephrectomy service (adults)

Ex-vivo partial nephrectomy can be used to treat cancers in patients with a single kidney and offers the possibility of cancer cure and avoiding a life of dialysis.

The overall aim of the service is to provide patients with complex renal tumours in solitary kidneys or bilateral disease not suitable for conventional treatments, the possibility of cancer cure and avoidance of dialysis. The service provides; initial assessment and evaluation, surgery and postoperative recovery; and long-term follow-up.

NHS centre	Oxford University Hospitals NHS Trust
Expenditure	<£0.5 million
Patients accepted into service	Data suppressed to maintain patient confidentiality
Outcomes collated	• % 1-year survival post operation: 100%
Geographical equity access	Numbers too small to analyse

Gender identity development service for children and adolescents

The gender identity development service is a Tier 4 specialist multidisciplinary mental health service that provides support and therapeutic input for children and adolescents who have social and psychological difficulties with the development of their gender identity. Depending on need and subject to meeting strict criteria, the service works with paediatric endocrinology clinics who may prescribe and administer hormone therapy and early intervention from 12 years onwards.

The service provides outreach support to patients and families across the country. This network model of management for children struggling with the development of their gender identity involves close collaboration between the national service and local child and adolescent mental health services.

NHS centres	The Tavistock and Portman NHS Foundation Trust
Expenditure	>£5 million but <£10 million
Number of referrals	2,390
Outcomes collated	• Median change in Children's Global Assessment Scale (CGAS) score between first appointment and follow-up (increase indicates improvement): 2.2% when comparing the first and last measurements of the CGAS score. Note: CGAS recorded as generally positive changes in young people's functioning observed.
Geographical equity access	Data not available or not comparable

Hand and upper limb transplantation service (adults)

Hand and upper limb transplantation is possible following cadaveric donation. The surgery involved is extremely complex and recipients have, as with other cadaveric transplants, to take immunosuppressive drugs for life to prevent the transplanted organ being rejected.

This service provides assessment, transplantation and follow-up.

NHS centres	Leeds Teaching Hospitals NHS Trust
Expenditure	<£0.5 million
Number of referrals	4
Outcomes collated	Data suppressed to maintain patient confidentiality
Geographical equity access	Numbers too small to analyse

Heart transplantation service (adults)

The heart transplant service provides: assessment of adult patients who are eligible for a heart transplant; the transplant operation; and lifelong follow-up.

NHS centres	Manchester University NHS Foundation Trust Papworth Hospital NHS Foundation Trust [now Royal Papworth Hospital NHS Foundation Trust Royal Brompton & Harefield NHS Foundation Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust University Hospitals Birmingham NHS Foundation Trust Sheffield Teaching Hospitals NHS Foundation Trust (follow-up only)
Expenditure	>£50 million (adult and children, heart and lung)
Number of transplants	149
Outcomes collated	• 30-day risk-adjusted patient survival rates after first adult heart transplant: – Birmingham: 88.7% – Brompton & Harefield: 88.4% – Manchester University: 93.9% – Newcastle upon Tyne: 89.1% – Papworth: 94.2% • 1-year risk-adjusted patient survival rates after first adult heart transplant by centre: – Birmingham: 79.9% – Brompton & Harefield: 80.1% – Manchester University: 85.0% – Newcastle upon Tyne: 79.8% – Papworth: 88.4% • 5-year risk-adjusted patient survival rates from listing for first heart transplants:

	– Birmingham: 74.9%; – Brompton & Harefield: 65.8% – Manchester University: 61.1% – Newcastle upon Tyne: 66.9% – Papworth: 79.3%
Geographical equity access	Some evidence of geographical inequity which is thought to be of a temporary nature

High consequence infectious diseases units (adults and children) – airborne diseases

The purpose of a special isolation unit (airborne) is the safe and effective treatment of high consequence infectious diseases (HCIDs) that are known or suspected to be transmissible from person to person via the airborne route. Services are commissioned for readiness and resulting activity is very small.

NHS centres	Royal Free London NHS Foundation Trust Guy's and St Thomas' NHS Foundation Trust Liverpool University Hospitals NHS Foundation Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust
Expenditure	>£0.5 million but <£1 million
Caseload	Not applicable
Outcomes collated	• Number of cases where HCID infection has spread from the specialist isolation unit • Number of occasions where unit is unable to admit and start treatment of any patient with a confirmed diagnosis of airborne HCID within 6 hours (maximum) of notification
Geographical equity access	Not applicable

High consequence infectious diseases units (adults and children) – contact diseases

The purpose of a special isolation unit (contact) is the safe and effective treatment of high consequence infectious diseases (HCIDs) that are known or suspected to be transmissible from person to person via the contact route. Services are commissioned for readiness and resulting activity is very small.

NHS centres	Royal Free London NHS Foundation Trust Liverpool University Hospitals NHS Foundation Trust Sheffield Teaching Hospital NHS Foundation Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Caseload	Not applicable
Outcomes collated	• Not defined
Geographical equity access	Not applicable

Insulin resistant diabetes (adults and children)

Insulin-resistant diabetes occurs because of either a genetic condition or because the individual has developed antibodies to insulin. In addition to the usual complications of diabetes (renal failure, stroke, etc.) the condition can also affect the liver and can result in pancreatitis. The aim of the service is to provide diagnostic, therapeutic and educational support for both patients and their local healthcare professionals, and to establish and disseminate evidence-based recommendations for the therapy of this severe group of conditions.

NHS centre	Cambridge University Hospitals NHS Foundation Trust
Expenditure	<£0.5 million

Active caseload	204
Outcomes collated	• % of patients with specific diagnosis: 94% • % of patients maintaining HbA1c below 75 mmol/mol: 82% Note: glycated haemoglobin (HbA1c) is measured primarily to identify the 3-month average plasma glucose concentration
Geographical equity access	Strong evidence of geographical inequity, the reasons for which are not fully understood and will be explored

Islet transplantation service (adults)

Islet transplantation is of proven benefit for a very small group of eligible patients with Type 1 diabetes who suffer from recurrent episodes of severe hypoglycaemia. Successful transplantation can abolish episodes of hypoglycaemia unawareness and improve the quality of life of recipients, while also improving overall metabolic control. Patients who are already immunosuppressed for a kidney transplant may also benefit from islet transplantation through the improved metabolic control afforded by an islet after kidney transplant.

NHS centres	King's College Hospital NHS Foundation Trust Manchester University NHS Foundation Trust North Bristol NHS Trust Oxford University Hospitals NHS Trust Royal Free London NHS Foundation Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust
Expenditure	<£1 million but <£5 million
Number of transplants	Data suppressed to maintain patient confidentiality
Outcomes collated	• Median number of severe hypoglycaemic events between registration and transplant: before transplant was nine per year and at 1-year post transplant was no events per year. Of the

	patients for whom number of severe hypoglycaemic events at 1-year post-transplant was available, 85% experienced no events, 13% experienced one or two events and 2% experienced three or more events • For routine islet transplants between 1 April 2014 and 31 March 2018, median HbA1c dropped from 62 mmol/mol before transplant to 48 mmol/mol at 1-year post-transplant. Of those patients for whom HbA1c was reported at one year, 65% had an HbA1c of <53 mmol/mol Note: Glycated haemoglobin (HbA1c) is measured primarily to identify the 3-month average plasma glucose concentration.
Geographical equity access	Numbers too small to analyse

Liver transplantation service, including live liver transplantation (adults)

This service provides assessment, transplantation and lifelong follow-up for patients requiring liver transplant surgery, including from living donors. The three main indications for liver transplantations are primary and secondary biliary cirrhosis, chronic hepatitis and fulminant hepatic failure. There are about 900 liver transplants in the UK each year (adults and children).

NHS centres	Cambridge University Hospitals NHS Foundation Trust King's College Hospital NHS Foundation Trust Leeds Teaching Hospitals NHS Trust Royal Free London NHS Foundation Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust University Hospitals Birmingham NHS Foundation Trust
Expenditure	>£50 million (adults and children)
Number of transplants	787

Outcomes collated	• 1-year risk-adjusted patient survival for adult elective deceased donor first liver transplants: – Cambridge University: 95.2% – King's College: 95.1%; – Leeds Teaching: 92.4% – Royal Free: 94.3% – Newcastle upon Tyne: 88% – Birmingham: 93% • 5-year risk-adjusted patient survival for adult elective deceased donor first liver transplants: – Cambridge University: 88.1% – King's College: 83.7% – Leeds Teaching: excluded due to a lack of follow-up beyond 4 years – Royal Free: 82.9% – Newcastle upon Tyne: 77.2% – Birmingham: 81.2% • 5-year risk adjusted patient survival rates from listing for adult elective first liver registrations: – Cambridge University: 74% – King's College: 74% – Leeds Teaching: have been excluded due to a lack of follow-up beyond 4 years – Royal Free: 58% – Newcastle upon Tyne: 69% – Birmingham: 66% Note: The national survival rates after joining the transplant list for adult elective first liver only patients are 83% at 1, 70% at 5 and 57% at 10 years.
Geographical equity access	No evidence of geographical inequity

Liver transplantation service (children)

This service provides assessment, transplantation and lifelong follow-up for patients requiring liver transplant surgery, including from living donors. The main conditions for paediatric liver transplantation are biliary atresia, congenital metabolic conditions, other cirrhosis, mostly non-recurring, tumours and acute liver failure. There are about 100 paediatric liver transplants in England each year.

NHS centres	Birmingham Children's Hospital NHS Foundation Trust [now Birmingham Women's and Children's Hospital NHS Foundation Trust] King's College Hospital NHS Foundation Trust Leeds Teaching Hospitals NHS Trust
Expenditure	>£50 million (adults and children)
Number of transplants	91
Outcomes collated	• 1-year unadjusted patient survival for paediatric elective deceased donor first liver transplants: – Birmingham Women's and Children: 93.3% – King's College: 99.0% – Leeds Teaching: 97.7% • 5-year unadjusted patient survival for paediatric elective deceased donor first liver transplants: – Birmingham Women's and Children: 90.8% – King's College: 91.1% – Leeds Teaching: 93.9%
Geographical equity access	No evidence of geographical inequity

Lung transplantation service (adults)

The lung transplant service provides: assessment of adult patients who are eligible for a lung transplant; the transplant operation; and lifelong follow-up.

NHS centres	Manchester University NHS Foundation Trust Royal Brompton & Harefield NHS Foundation Trust Papworth Hospital NHS Foundation Trust [now Royal Papworth Hospital NHS Foundation Trust] The Newcastle upon Tyne Hospitals NHS Foundation Trust University Hospitals Birmingham NHS Foundation Trust
Expenditure	>£50 million (adults and children, heart and lung)
Number of transplants	142
Outcomes collated	• 90-day risk-adjusted patient survival rate after first adult lung transplant: – Brompton & Harefield: 88.8% – Manchester University: 95.4% – Papworth: 90.2% – Newcastle upon Tyne: 85.3% – Birmingham: 83.3% • 1-year risk-adjusted patient survival rate after first adult lung transplant: – Brompton & Harefield 85.3% – Manchester University: 87.4% – Papworth: 79.9% – Newcastle upon Tyne: 78.6% – Birmingham: 71.6% • 5-year risk-adjusted patient survival rate from listing for first lung only transplants: – Brompton & Harefield: 53.7% – Manchester University: 50.7% – Papworth: 47.9% – Newcastle upon Tyne: 47.2% – Birmingham: 32.1%

Geographical equity access	No evidence of geographical inequity

Lymphangioleiomyomatosis

Lymphangioleiomyomatosis (LAM) is a rare, progressive disease characterised by lung cysts, kidney tumours and lymphatic abnormalities. LAM occurs in a sporadic form, which affects females only, usually of childbearing age; LAM also occurs in patients who have tuberous sclerosis complex (TSC), a genetic condition that causes non-malignant tumours to grow in the brain and on other vital organs.

The service is delivered through outpatient assessment and management; and lung transplant referral.

NHS centre	Nottingham University Hospitals NHS Trust
Expenditure	<£0.5 million
Caseload	65
Outcomes collated	• % of patients having a pneumothorax: 0.04% • % of patients having a renal angiolipoma bleed: 0% • % of patients having an FEV_1 decline of >150 mL per annum: 0.07%
Geographical equity access	Evidence of geographical inequity. This may be due to the expert centre's ability to detect very mild forms of the disease which would be missed elsewhere. This requires further investigation.

Lysosomal storage disorders service (children and adults)

Lysosomal storage disorders (LSDs) are a group of rare genetic storage disorders, characterised by specific lysosomal enzyme deficiencies. Some LSDs can be treated using enzyme replacement therapies (ERTs), substrate reduction therapy (SRT) or other disease modifying drugs.

There are licensed disease-modifying treatments for eight LSDs:

- Gaucher’s disease
- Anderson-Fabry’s disease
- mucopolysaccharidosis type I (MPSI; which occurs as Hurler’s syndrome, Hurler-Scheie syndrome and Scheie syndrome)
- mucopolysaccharidosis type IVa (Morquio syndrome)
- mucopolysaccharidosis type VI (MPSVI or Maroteaux Lamy syndrome)
- Pompe’s disease
- mucopolysaccharidosis type II (MPSII)
- Niemann Pick type C.

NHS centres	Birmingham Children's Hospital NHS Foundation Trust [now Birmingham Women’s and Children's Hospital NHS Foundation Trust] Cambridge University Hospitals NHS Foundation Trust Great Ormond Street Hospital for Children NHS Foundation Trust Manchester University NHS Foundation Trust Royal Free London NHS Foundation Trust Salford Royal NHS Foundation Trust University College London Hospitals NHS Foundation Trust University Hospitals Birmingham NHS Foundation Trust
Expenditure	>£50 million
Active caseload	2,419 (of these, 1,046 received enzyme replacement therapy and substrate reduction therapy drugs)
Outcomes collated	• Fabry’s type 1: % of patients initiating renal replacement therapy among patients treated for 3 years or more: – Birmingham Women’s and Children’s: 3% – Cambridge University: 0% – Great Ormond Street: 0% – Manchester University: 0% – Royal Free: 1%

	– Salford: 1.5% – University College London: 8% – University Hospitals Birmingham: 3% • Fabry's type 2: % of patients treated for 3 years or more having a stroke: – Birmingham Women's and Children's: 0% – Cambridge University: 0% – Great Ormond Street: 0% – Manchester University: 0% – Royal Free: 0% – Salford: 1.5% – University College London: 0% – University Hospitals Birmingham: 0% • Fabry's type 3: % of patients treated for 3 years or more having a cardiac device implanted: – Birmingham Women's and Children's: 7% – Cambridge University: 0% – Great Ormond Street: 0% – Manchester University: 0% – Royal Free: 1% – Salford: 3.1% – University College London: 0% – University Hospitals Birmingham: 6% • Gaucher's: % of patients treated for 3 years or more having a hospital admission for bone crisis: – Birmingham Women's Children's: 0% – Cambridge University: 0% – Great Ormond Street: 14% – Manchester University: 0% – Royal Free: 0% – Salford: 0% – University College London: 0%

	• University Hospitals Birmingham: 0%MPS: % of patients treated for 3 years or more having a new cranio-cervical episode: – Birmingham Women's and Children's: 0% – Cambridge University: 0% – Great Ormond Street: 0% – Manchester University: 0% – Royal Free: 13% – Salford: 0% – University College London: 0% – University Hospitals Birmingham: 0%
Geographical equity access	New data definition proposed

McArdle's disease service

McArdle's disease is a condition caused by an inborn deficiency of muscle phosphorylase resulting in an abnormal accumulation of glycogen in muscle tissue, characterised by exercise intolerance, muscular pain, fatigability and muscle cramping. Rhabdomyolysis (the breakdown and death of muscle fibres and release of their contents into the bloodstream following a direct or indirect muscle injury) leading to renal failure is a particularly severe complication of McArdle's disease.

The service provides an accurate diagnosis and outpatient management of the condition.

NHS centre	University College London Hospitals NHS Foundation Trust
Expenditure	>£0.5 million but <£1 million
Caseload	211
Outcomes collated	• Median functional capacity – 12MWD: 815 m • Number of patients requiring hospital assessment: 7 admissions • Median quality of life (SF36) score: physical functioning: 46.0

	Note: 12MWD is a 12-minute walking distance test used to estimate functional exercise capacity.
Geographical equity access	No evidence of geographical inequity

Multiple sclerosis management service for children

The multiple sclerosis (MS) management service for children is commissioned to provide a service for children with MS, with suspected MS or the equally rare 'MS-like' recurrent acquired demyelinating syndromes, or who following a first demyelination episode have a high risk of relapse which will require similar treatments.

NHS centres	**North Hub Lead Centre (single centre with three units):** • Alder Hey Children's NHS Foundation Trust • Manchester University NHS Foundation Trust • The Newcastle upon Tyne Hospitals NHS Foundation Trust **Midland hubs:** • Birmingham Women's and Children's Hospital NHS Foundation Trust • Cambridge University Hospitals NHS Foundation Trust **London and the South hubs:** • Great Ormond Street Hospital for Children NHS Foundation Trust • Guy's and St Thomas' NHS Foundation Trust
Expenditure	New service – information not yet available
Caseload	247
Outcomes collated	Not yet available
Geographical equity access	Not yet available

Neurofibromatosis type 2 service (all ages)

Neurofibromatosis type 2 (NF2) is a genetic disorder characterised by the growth of non-cancerous tumours in the central nervous system. NF2 patients develop bilateral vestibular schwannomas (abnormal tissue growth originating in the cells of the sheath around the nerve), meningiomas (a type of benign brain tumour) and spinal tumours; usually causing deafness, balance problems, compression of the brain stem and premature death.

The service includes:

- outpatients: MDT outpatients and satellite outpatients
- mutation testing for NF2
- auditory brainstem implants and auditory implants
- vestibular schwannomas surgery
- stereotactic radiosurgery
- LINK's NF2 course (intensive rehabilitation programmes for adults with significant hearing impairment)
- drug treatment in line with agreed protocols.

NHS centres	Cambridge University Hospitals NHS Foundation Trust Guy's and St Thomas' NHS Foundation Trust Manchester University NHS Foundation Trust Oxford University Hospitals NHS Trust
Expenditure	>£5 million but <£10 million
Caseload	981
Outcomes collated	• Median length of time (>3 years) that useful hearing (measured by speech discrimination of >60%) is maintained in at least one ear from date of diagnosis: – Cambridge University: 44 months (range of time from diagnosis 1-95 months)

	– Guy's and St Thomas': for patients whose hearing was still preserved at the end of follow-up – for patients who lost hearing before the end of follow-up – 38 months – Manchester University: 1 in 66 lost hearing before a median of five years – Oxford University: of patients who had useful hearing when they joined the service (from 2010 onwards), only one lost useful hearing – Median duration for the groups combined: 40 months • % facial palsy rates of <20% 18 months post-surgery for vestibular schwannoma: – Cambridge University: 18% – Guy's and St Thomas': post-surgery facial palsy rate of 14% – Manchester University: 11% – Oxford University: among all patients who had good facial function pre-operatively, facial function had dropped in 20% 18 months post operatively
Geographical equity access	No evidence of geographical inequity

Neuromyelitis optica service (adults and children)

Neuromyelitis optica (NMO) (also known as Devic's disease) is a rare inflammatory demyelinating disorder of the central nervous system that typically presents as severe optic neuritis (inflammation of the optic nerve) and longitudinally extensive myelitis (inflammation of the spinal cord) often followed by further severe attacks, which usually result in permanent disability (visual loss, limb weakness, respiratory muscle weakness). There are high mortality and morbidity rates associated with the condition. About 1,000 people in England are living with NMO.

The service provides an accurate diagnosis, inpatient or outpatient assessment and review.

NHS centres	Oxford University Hospitals NHS Trust The Walton Centre NHS Foundation Trust

Expenditure	>£1 million but <£5 million –
First evaluations	179
Outcomes collated	• Annualised relapse rate: Oxford University: 8% • Walton Centre: 0%
Geographical equity access	Data not available or not comparable

Ocular oncology service (adults)

The ocular oncology service provides diagnosis and treatment of adults with suspected malignant tumours of the eye. Of the patients referred to the service one third (about 700 each year) are confirmed as having eye cancer.

There are a number of different treatment modalities:

- surgery
- radiotherapy
- phototherapy
- cryotherapy
- chemotherapy.

These aim wherever possible to preserve vision in the affected eye and can be used individually or in a combination. At present it is unclear if any of these treatments have better outcomes than the others. Follow-up care is provided for patients whose tumours recur or who have complications requiring treatment.

NHS centre	Moorfields Eye Hospital NHS Foundation Trust The Royal Liverpool and Broadgreen University Hospitals NHS Trust [now Liverpool University Hospitals NHS Foundation Trust] Sheffield Teaching Hospitals NHS Foundation Trust
Expenditure	>£5 million but <£10 million – same banding

Positive assessment	693
Outcomes collated	• % primary enucleation among patients with melanoma: – Liverpool University: 20% – Moorfields: 2.5% – Sheffield Teaching: 19% • % secondary enucleation among patients with melanoma: – Liverpool University: 1% – Moorfields 2% – Sheffield Teaching: 0% • % developing metastatic disease among patients with melanoma: – Liverpool University: 1% – Moorfields: 3% – Sheffield Teaching: 2%
Geographical equity access	Data required from cancer register

Ophthalmic pathology service (adults and children)

The National Specialist Ophthalmic Pathology Service (NSOPS) is the core national reference service for the specialist reporting of ophthalmic histopathology and cytology specimens.

This service includes diagnosis and advice relevant to the clinical management of eye conditions. The service provides a comprehensive diagnostic service for malignant and non-malignant conditions for the following specimen types: eyelid, conjunctiva, cornea, aqueous and vitreous humour, iris, ciliary body, retina, choroid, sclera and orbit (including lacrimal gland and optic nerve).

The service receives about 3,700 specimens each year.

NHS centres	Manchester University NHS Foundation Trust Sheffield Teaching Hospitals NHS Foundation Trust

	The Royal Liverpool and Broadgreen University Hospitals NHS Trust [now Liverpool University Hospitals NHS Foundation Trust] University College London Hospitals NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Total cases reviewed	4,022
Outcomes collated	• % of simple cases reported within 7 working days: – Manchester University: 91% – Sheffield Teaching: 92% – Liverpool University: %78% (calendar days) – University College London: 91% • % of complex cases reported within 10 working days: – Manchester University: 98% – Sheffield Teaching: 95% – Liverpool University: 92% (calendar days) – University College London: 92% • % of all cases reported within 21 working days: – Manchester University: 100% – Sheffield Teaching: 100% – Liverpool University: 100%. (calendar days) – University College London: 100%
Geographical equity access	Data not available or not comparable

Osteo-odonto-keratoprosthesis service for corneal blindness (adults)

Osteo-odonto-keratoprosthesis (OOKP) surgery is a specialist surgical intervention that can restore meaningful vision to patients suffering from end stage corneal blindness, and for whom conventional corneal surgery is not possible for reasons such as severe ‘dry eyes’ that causes heavy scarring of the cornea. OOKP is only contemplated in patients where no other treatments would restore sight.

During OOKP, patients are initially assessed by ophthalmic and maxillofacial consultants, involving examination of the eyes, teeth and mouth. OOKP is then a two-stage procedure that firstly involves the extraction of the patient's own tooth and bone, which are then fashioned into a 'bolt' and placed within the eye for supporting a synthetic optical cylinder.

The second stage of the procedure is performed about four months after the first stage. Each surgical procedure lasts about six hours and patients require lifelong follow-up.

NHS centre	Brighton and Sussex University Hospitals NHS Trust
Expenditure	<£0.5 million
Stage 2 surgery	Data suppressed to maintain patient confidentiality
Outcomes collated	• % patients with visual acuity 6/12 or better at 12 months post operation: 75%
Geographical equity access	Numbers too small to analyse

Paediatric intestinal pseudo-obstructive disorders service

Chronic intestinal pseudo-obstruction is an intestinal motility disorder. Impaired intestinal motor activity causes recurrent symptoms of intestinal obstruction in the absence of mechanical occlusion. The service provides expert, multidisciplinary diagnostic services for infants and children under five with congenital and acquired variations of the condition.

This service provides a prompt and accurate diagnosis leading to rapid access to definitive treatment. There is evidence of unnecessary investigation without a definitive diagnosis. The service treats children under the age of five.

The service treats about 20 children each year.

NHS centre	Great Ormond Street Hospital for Children NHS Foundation Trust

Expenditure	>£1 million but <£5 million
Number of new patient referrals	Data suppressed to maintain patient confidentiality
Outcomes collated	• % clinical phenotype defined within 2 weeks (should increase or stay the same): 11% • % pathological diagnosis within 2 weeks of phase 2 period (should increase or remain the same): data suppressed to maintain patient confidentiality
Geographical equity access	Data not available or not comparable

Pancreas transplantation service (adults)

This service provides assessment, transplantation and lifelong follow-up for diabetic patients requiring pancreas transplant surgery.

NHS centres	Cambridge University Hospitals NHS Foundation Trust Guy's and St Thomas' NHS Foundation Trust Imperial College Healthcare NHS Trust Manchester University NHS Foundation Trust Oxford University Hospitals NHS Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust
Expenditure	>£5 million but <£10 million
Number of transplants	148
Outcomes collated	• 1-year risk-adjusted patient survival from first transplant from deceased donors: – Cambridge University: 99%; – Guy's and St Thomas': 96%

	– Imperial College: 100 – Manchester University: 99% – Newcastle upon Tyne: 100% – Oxford University: 96% • 5-year risk-adjusted patient survival from first transplant from deceased donors: – Cambridge University: 93% – Guy's and St Thomas': 83% – Imperial College: 84% – Manchester University: 90% – Newcastle upon Tyne: 82% – Oxford University: 87% • 5-year risk-adjusted patient survival from listing from first deceased donor transplant: – Cambridge University: 87% – Guy's and St Thomas': 87% – Imperial College: 86% – Manchester University: 86% – Newcastle upon Tyne: 86% – Oxford University: 85%
Geographical equity access	No evidence of geographical inequity

Paroxysmal nocturnal haemoglobinuria

Paroxysmal nocturnal haemoglobinuria (PNH) is a rare disease in which red blood cells break down earlier than normal. Symptoms include abdominal pain, back pain, blood clots, dark urine, easy bruising or bleeding, headache and shortness of breath. About 650 people in England suffer from PNH.

This service provides diagnosis, clinical review and ongoing management for patients with the haemolytic form of paroxysmal nocturnal haemoglobinuria who are eligible for treatment with anti-complement targeted therapy.

Outreach clinics are held in locations outside of the centres.

NHS centres	Leeds Teaching Hospitals NHS Trust King's College Hospital NHS Foundation Trust
Expenditure	>£50 million
Caseload	239 receiving eculizumab 486 patients not receiving eculizumab 837 Total
Outcomes collated	• 5-year relative survival rate: – Leeds Teaching: 97% – King's College: 82% • Median transfusions per patient in previous 12 months: – Leeds Teaching: 0 – King's College: Data not available
Geographical equity access	No evidence of geographical inequity

Primary ciliary dyskinesia management service (children)

Primary ciliary dyskinesia (PCD) is a genetic disorder of the air tubes of the lungs (the bronchi), which become infected and filled with pus due to abnormalities of the hair-like structure (cilia) of the cells lining the respiratory tract. This can lead to repeated infections and damage the lung, especially if diagnosis is delayed.

This service provides a multidisciplinary outpatient-based diagnostic, advice and management service (including respiratory, ENT, audiology and physiotherapy) to patients who are referred with a confirmed diagnosis of primary ciliary dyskinesia (PCD). It also supports and trains them in aspects of self-care treatment. The service also provides support to local providers when managing patients within an inpatient setting. About 1 in 100,000 of the population has PCD, which equates to about 560 patients in England.

NHS centres	Leeds Teaching Hospitals NHS Trust

	Royal Brompton & Harefield NHS Foundation Trust University Hospital Southampton NHS Foundation Trust University Hospitals of Leicester NHS Trust
Expenditure	>£1 million but <£5 million (for management and diagnostic elements)
Caseload	845
Outcomes collated	• % of patients in the PCD management service offered an annual review appointment (consisting of the processes listed in the service specification: – Brompton & Harefield: 92%; – Leicester and Birmingham: 100% – Leeds Teaching: 84% – Southampton: 100% • % of patients seen by a physiotherapist at annual review: – Brompton & Harefield: 100% – Leicester and Birmingham: 95% – Leeds Teaching: 84% – Southampton: 86%
Geographical equity access	Data not available or not comparable

Primary malignant bone tumours service (adults and adolescents)

This service provides diagnosis and surgery for primary malignant bone cancers. Examples of conditions include osteosarcoma, chondrosarcoma and Ewing's sarcoma. The key aim is to avoid amputation if possible while ensuring complete removal of the cancer.

The service receives about 1,000 referrals of suspected primary malignant bone tumours (PMBT) each year, of which around 300 are confirmed as having a PMBT.

NHS centres	Oxford University Hospitals NHS Trust Royal National Orthopaedic Hospital NHS Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust The Robert Jones and Agnes Hunt Orthopaedic Hospital NHS Foundation Trust The Royal Orthopaedic Hospital NHS Foundation Trust
Expenditure	>£10 million but <£20 million
Number of confirmed cases	306
Outcomes collated	• % 3-year local recurrence among patients having limb salvage: – Oxford University: 73% – Robert Jones and Agnes Hunt: 3% – Royal Orthopaedic: 6% – Royal National Orthopaedic: 8% – Newcastle upon Tyne: 94%% • % limb salvage: – Oxford University: 13% – Robert Jones and Agnes Hunt: 91%; – Royal Orthopaedic: 72%; – Royal National Orthopaedic: 85%. – Newcastle upon Tyne: 83% • % 3-year prosthesis infection/loosening: – Oxford University: 16% – Robert Jones and Agnes Hunt: 0% – Royal Orthopaedic: 15% – Royal National Orthopaedic: 8%. – Newcastle: 10%
Geographical equity access	Data not available or not comparable

Proton beam therapy service (adults and children)

Proton beam therapy is a type of radiotherapy that uses a beam of high energy protons, which are small parts of atoms, rather than high energy x-rays (called ‘photons’) to treat specific types of cancer.

Proton beam therapy enables a dose of high energy protons to be precisely targeted at a tumour, reducing the damage to surrounding healthy tissues and vital organs which is an advantage in certain groups of patients or where the cancer is close to a critical part of the body such as the spinal cord.

Proton beam therapy is only suitable for certain types of cancer, such as highly complex brain, head and neck cancers and sarcomas as it does not lead to better outcomes for many cancer cases than using high energy x-rays, which is still considered the most appropriate and effective treatment for the majority of cancers.

Like high energy x-ray radiotherapy, proton beam therapy is painless, but patients may experience side effects similar to those experienced from other forms of radiotherapy.

Centres	The Christie NHS Foundation Trust University of Florida Health Proton Institute, Jacksonville, USA Westdeutsches Protonentherapiezentrum (WPE) Essen, Germany Paul Scherrer Institut, Villigen, Switzerland
Expenditure	Overseas providers: >£20 million but <£30 million The Christie NHS Foundation Trust: data not yet available
Number of patients approved for referral	Total patients overseas (all UK countries) 190 – paediatrics 130, Adult 60 Total patients The Christie (all UK countries) 46 – paediatrics 22, TYA 11, Adult 13
Outcomes collated	• 3-year combined English and overseas service unpublished actuarial overall survival and local control rates for adults and children with tumours of the central nervous system: 92% and 94% respectively
Geographical equity access	Geographical analysis to be undertaken during 2020

Pseudomyxoma peritonei service (adults)

Pseudomyxoma peritonei (PMP) is a rare, mucus-producing tumour, which spreads to compress the abdominal organs. PMP usually arises from a ruptured tumour of the appendix. The condition is of borderline malignancy in that it does not metastasise by the bloodstream or through lymphatic spread in the early stages. The tumour spreads locally within the peritoneal cavity and eventually compresses the abdominal organs. The disease is slow growing and is considered a relatively benign condition. However, without specialist cancer treatment, the majority of patients die either from complications of repeated surgery or from compression of the small bowel with resulting malnutrition.

The symptoms of PMP are varied with most patients complaining of gradual abdominal swelling over a period of time affecting their ability to eat normally.

Treatment options include:

- cytoreduction with HIPEC (hyperthermic intraperitoneal chemotherapy)
- debulking of the tumour is also an option.
- draining of the abdomen as part of supportive care.

About 200 new patients present each year in England with PMP.

NHS centre	Hampshire Hospitals NHS Foundation Trust The Christie NHS Foundation Trust
Expenditure	>£20 million but <£30 million
Major full cytoreduction	224
Outcomes collated	• 5-year patient survival – all operative cases: – Christie: 83% – Hampshire: 75% • 5-year patient survival – complete cytoreduction: – Christie: 93% – Hampshire: 91%

Geographical equity access	Data not available or not comparable

Pulmonary hypertension service for children

Paediatric pulmonary hypertension (PH) is a high pressure in the circulation of blood through the lungs, leading to progressive heart failure. The prognosis has improved with recently developed drugs. Some patients also need a lung (or heart and lung) transplant.

All patients are investigated, diagnosed, have their treatment for PH determined and their care package organised at the Highly Specialist Pulmonary Hypertension Centre by a multidisciplinary team.

The service provides care for patients with pulmonary hypertension including cardiac catheterisation, invasive radiology, echocardiography, non-invasive imaging (CT scanning, magnetic resonance imaging), exercise physiology and lung function testing. Patients may also need frequent access to microbiology, dental services, psychology, dietetics and other paediatric expertise.

The service cares for about 500 children with PH.

NHS centre	Great Ormond Street Hospital for Children NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Caseload	518
Outcomes collated	• % of patient followed up in the year with at least one functional class measure (denominator) who achieved a functional class of 2 or better (numerator): 73% • Proportion/% of children receiving epoprostenol who required a line change due to infection: 10% • Proportion of children receiving epoprostenol who experienced a line-related bloodstream infection: 0%
Geographical equity access	No evidence of geographical inequity

Pulmonary thromboendarterectomy service (adults and adolescents)

Pulmonary thromboendarterectomy (PTE) is complex surgery to remove blood clots and related material from the pulmonary artery of people with chronic thromboembolic pulmonary disease (CTEPH) (repeated episodes of blood clots travelling to the lung) that may cause life-threatening pulmonary hypertension (raised pressure in the artery that carries blood to the lung). The aim of the service is to treat all patients with operable CTEPH. Through the network of adult pulmonary hypertension units, all patients with a diagnosis of CTEPH are referred for consideration of surgery. A secondary aim is to help spread awareness of CTEPH and the success of pulmonary endarterectomy surgery.

NHS centre	Papworth Hospital NHS Foundation Trust [now Royal Papworth Hospital NHS Foundation Trust]
Expenditure	>£5 million but <£10 million
Caseload	157
Outcomes collated	• 90-day patient survival: 96% • 3-year patient survival: 93% • In-hospital mortality: 3%
Geographical equity access	No evidence of geographical inequity

Rare mitochondrial disorders service (adults and children)

Mitochondria are small organelles, present in every cell in the body – whose function is to process the cell's energy. They contain their own genetic complement, the mitochondrial genome, and their principal task is to provide the energy necessary for normal cell functioning and maintenance. Disruption of this energy supply can have devastating effects for the cell, organ and individual. One important consequence of mitochondrial involvement in all cell types is that mitochondrial disease can affect virtually any organ and present with a plethora of

symptoms and signs to a variety of specialties. These genuinely multi-system diseases are associated with significant morbidity and mortality.

The service provides diagnostic services for those patients with suspected rare mitochondrial disorders, which cannot be diagnosed by standard genetic tests available at Clinical Molecular Genetics Society-affiliated diagnostic laboratories.

The Highly Specialist Mitochondrial Disorders Centres provide:

- Specialist histochemical, biochemical and molecular genetics
- Multi-disciplinary outpatient assessment, including access to cardiology, ophthalmology, diabetology, neurology, genetics, physiotherapy, speech therapy

The service diagnoses about 280 new patients each year.

NHS centres	Oxford University Hospitals NHS Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust University College London Hospitals NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Outpatient referrals	461
Outcomes collated	• % of patients with a genetic diagnosis: – Oxford University: 7% – Newcastle upon Tyne: Data not available – University College London: 63% • % of patients given an alert card: – Oxford University: 90% – Newcastle upon Tyne: Data not available – University College London: 63%
Geographical equity access	Data not available or not comparable

Retinoblastoma service (children)

Retinoblastoma is a malignant tumour of the retina and usually presents in children under the age of two. It is an aggressive eye cancer which can result in the loss of vision and in extreme cases, death.

The treatment modalities are as follows:

- laser treatment – heat treatment to destroy the tumour
- cryotherapy – freezing treatment to destroy the tumour
- radiotherapy – external beam plaque brachytherapy to damage the tumour and control its growth
- chemotherapy – to shrink the tumour (often combined with laser treatment)
- enucleation – surgical removal of the eye in advanced cases.

About 50 children are diagnosed with retinoblastoma each year.

NHS centres	Barts Health NHS Trust Birmingham Children's Hospital NHS Foundation Trust [now Birmingham Women's and Children's Hospital NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Confirmed patients	50
Outcomes collated	• % 5-year survival: – Barts Health: 99% – Birmingham Women's and Children's: 100% • % primary enucleation: – Barts Health: 40% – Birmingham Women's and Children's: 59%
Geographical equity access	Data required from cancer register

Severe acute porphyria (adults and children)

Acute porphyrias are a rare, inherited disorder, typically presenting in young adults. Acute attacks can be life-threatening. The condition can result in permanent disability and even death due to progressive motor neuropathy.

The service comprises two elements:

- An acute support service to hospitals around the country. Advice is given on the treatment of the patient and the two centres also arrange for a stock of the drug, haem arginate, to be sent where appropriate.
- A structured multidisciplinary follow-up service for patients after acute attacks and severely affected patients with recurrent attacks, often complicated by paralysis, and increased risk of kidney disease and hypertension.

The service treats about 150 people per annum with acute porphyria who meet the definition of ‘severe’ disease.

NHS centres	King's College Hospital NHS Foundation Trust University Hospital of Wales
Expenditure	>£1 million but <£5 million
Active caseload	119
Outcomes collated	• % mortality rate: – King’s College: 0% – Wales: 0% • % of patients having four or more hospital admissions (porphyria-related) in the previous 12 months: – King’s College: 6% – Wales: 5%
Geographical equity access	Numbers too small to analyse

Severe combined immune deficiency and related disorders service (children)

Severe combined immunodeficiency disorders (SCID) is the term used to cover the most serious types of primary immunodeficiency where various components of the body's defence system are defective, leaving the child prone to unusual and/or frequent infections. In all forms of SCID, both T and B lymphocyte functions, the body's defence mechanisms, are defective from birth.

Treatment is usually through a bone marrow or stem cell transplant to boost the immune system. In some cases, gene therapy or thymus transplantation is appropriate.

There are about 60 referrals to the service each year and about the same number of transplants.

NHS centres	Great Ormond Street Hospital for Children NHS Foundation Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust
Expenditure	>£10 million but <£20 million
Number of transplants	66
Outcomes collated	• % 24-month patient survival post HSCT (or gene therapy): – Great Ormond Street: 81% – Newcastle upon Tyne: 100%
Geographical equity access	No evidence of geographical inequity

Small bowel transplantation service (adults)

This service provides assessment, transplantation and lifelong follow-up of adult patients requiring small bowel transplantation.

NHS centres	Cambridge University Hospitals NHS Foundation Trust Oxford University Hospitals NHS Trust

Expenditure	>£5 million but <£10 million (adults and children combined)
Number of transplants	Data suppressed to maintain patient confidentiality
Outcomes collated	• % unadjusted 90-day patient survival for elective first intestine transplants between 01/04/2007 and 31/03/2019: – Cambridge University: 92.7% – Oxford University: 88.6% • % unadjusted 1-year patient survival from elective first intestine transplants between 01/04/2007 to 31/03/2017: – Cambridge University: 77.7% – Oxford University: 80.5% • % unadjusted 5-year patient survival from elective first intestine transplants between 01/04/2007 and 31/03/2017: – Cambridge University: 48.3% – Oxford University: 56.9%
Geographical equity access	Numbers too small to analyse

Small bowel transplantation service (children)

This service provides assessment, transplantation and lifelong follow-up of paediatric patients requiring small bowel transplantation.

NHS centres	Birmingham Children's Hospital NHS Foundation Trust [now Birmingham Women's and Children's Hospital NHS Foundation Trust] King's College Hospital NHS Foundation Trust
Expenditure	>£5 million but <£10 million (adults and children combined)
Number of transplants	Data suppressed to maintain patient confidentiality
Outcomes collated	• % unadjusted 90-day patient survival for elective first intestine transplants between 01/04/2007 and 31/03/2019:

	– Birmingham Women's and Children: 91.6% – King's College: 100% • % unadjusted 1-year patient survival from elective first intestine transplants between 01/04/2007 and 31/03/2019: – Birmingham Women's and Children: 82.5% – King's College: 89.7% • % unadjusted 5-year patient survival from elective first intestine transplants between 01/04/2007 and 31/03/2019: – Birmingham Women's and Children: 55.9% – King's College: 71.3%
Geographical equity access	Numbers too small to analyse

Specialist paediatric liver disease service

This service provides a diagnostic, assessment and treatment service for paediatric liver disease. The major conditions covered by the service are:

- acute liver failure
- biliary atresia
- chronic liver disease
- hepatitis A, B and C
- metabolic liver disease
- neonatal hepatitis.

NHS centres	Birmingham Children's Hospital NHS Foundation Trust [now Birmingham Women's and Children's Hospital NHS Foundation Trust] King's College Hospital NHS Foundation Trust Leeds Teaching Hospitals NHS Trust
Expenditure	>£10 million but <£20 million

Inpatient episodes	1,132
Outcomes collated	Data suppressed to maintain patient confidentiality. The service covers a large number of diagnoses but only a few patients will have each one
Geographical equity access	Numbers too small to analyse (by individual condition)

Stickler syndrome diagnostic service (adults and children)

Stickler syndrome is an inherited disorder of connective tissue associated with cleft palate, deafness and arthropathy. It is the commonest inherited cause of rhegmatogenous retinal detachment in children (where fluid passes into the space between the retina and the retinal pigment layer). Although the systemic features are widespread, the sight-threatening complications are generally the most serious, particularly the risk of giant retinal tear, which is frequently bilateral and, if untreated, can lead to blindness.

The service is an outpatient diagnostic service that focuses on genetic testing to establish the patient's sub-classification of the disease. The service sees about 100 new patients each year and their families.

NHS centre	Cambridge University Hospitals NHS Foundation Trust
Expenditure	>£0.5 million but <£1 million
Index patients	61
Outcomes collated	• % of patients with a definitive diagnosis or diagnosis ruled out: 88% definitive diagnosis • % of patients with a genetic diagnosis: 85%
Geographical equity access	No evidence of geographical inequity

Vein of Galen malformation service (adults and children)

Vein of Galen malformations (VGMs) are extremely rare abnormalities in the blood vessels in the brain leading to excess blood flow which can result in cardiac problems.

VGMs usually occur in fetuses or new-born children, although sometimes these problems do not present until later in life.
Treatment for VGMs in children involves injecting acrylate or placing a coil into the blood vessels to restore arteriovenous equilibrium.

The service treats about 10 new babies and children each year.

NHS centres	Alder Hey Children's Hospital (interim contract from March 2017) Great Ormond Street Hospital for Children NHS Foundation Trust
Expenditure	<£0.5 million
Number of procedures	Data suppressed to maintain patient confidentiality
Outcomes collated	Data suppressed to maintain patient confidentiality
Geographical equity access	No evidence of geographical inequity

Ventricular assist device as a bridge to heart transplantation or myocardial recovery (all ages)

Ventricular assist devices (VADs) can be attached externally or implanted within the body to support the adult's failing heart until a donor heart becomes available for transplantation, a technique known as 'bridge to transplant'.

VADs work by supporting the pumping action of the left ventricle, which is the main pumping chamber of the heart. They sometimes also need to be implanted in the right ventricle.

The implantation of a VAD is only considered in patients with advanced heart failure who are listed for a transplant and who are deemed to be deteriorating so rapidly that they would not survive long enough to receive a heart via the urgent allocation scheme. Occasionally, a VAD enables the heart to recover sufficiently for the device to be removed (‘bridge to recovery’).

A small but increasing number of children requiring a heart transplant are supported with ventricular assist devices (VADs), mechanical devices that circulate blood outside the body to supports the failing heart. This is known as ‘bridge to transplant’ and supports the heart until a donor heart becomes available for transplantation.

NHS centres	Great Ormond Street Hospital for Children NHS Foundation Trust Manchester University NHS Foundation Trust Royal Papworth Hospital NHS Foundation Trust [now Royal Papworth Hospital NHS Foundation Trust Royal Brompton & Harefield NHS Foundation Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust University Hospitals Birmingham NHS Foundation Trust
Expenditure	Figure included in heart and lung transplant –
Number of procedures	169
Outcomes collated	• Patients given a long-term VAD as bridge to transplant who received a transplant within 3 years: 20% • 3-year survival rate in adults with long-term bridging devices: 58.4% (up from 52.1% in 2017/18 and 46.6% in 2016/2017) • Unadjusted patient survival rates after first adult DBD (donor after brain death) heart transplant, by mechanical support status: – 30 days: 76% – 90 days: 70% – one year: 67%
Geographical equity access	Numbers too small to analyse

Wolfram syndrome service (adults and children)

Wolfram syndrome is a very rare inherited disorder. It is a progressive neurodegenerative disorder with a debilitating and life-threatening association of diabetes, blindness, deafness and brain disease.

Both the adult and paediatric services run clinics that undertake assessment of all patients in a multidisciplinary structure. Patients are assessed and reviewed by all the specialities appropriate to their needs during the clinic.

NHS centres	Birmingham Children's Hospital NHS Foundation Trust [Birmingham Women's and Children's Hospital NHS Foundation Trust] University Hospitals Birmingham NHS Foundation Trust
Expenditure	<£0.5 million
Caseload	78
Outcomes collated	• % of children with HbA1c in target range: – Birmingham joint reporting: 77% • % of adults with HbA1c <75 mmol/mol: – Birmingham joint reporting: 49% • % of adults with a BMI <35: – Birmingham: 84%
Geographical equity access	Numbers too small to analyse

Xeroderma pigmentosum service (adults and children)

Xeroderma pigmentosum is a life-threatening inherited disorder affecting the skin, eyes and nervous system. A defect in the process of repairing ultraviolet-induced DNA damage results in: severe sunburn-type reactions to daylight; skin cancers in exposed skin from early childhood; eye disease; and progressive neurological degeneration in 20–30% of patients. There are about 120 people with the condition in the UK. The service provides a laboratory diagnostic service and 1-stop

multidisciplinary clinic for patients to advice on ongoing management and intervention necessary for their condition.

NHS centre	Guy's and St Thomas' NHS Foundation Trust
Expenditure	>£0.5 million but <£1 million
Caseload	125
Outcomes collated	• % of adults with window film at home: 63% • % of children with window film at home: 100% • % of children wearing sunglasses when outside (who do not wear a visor): 80% • % of adults wearing sunglasses when outside (who do not wear a visor): 71%
Geographical equity access	No evidence of geographical inequity

Award

Guy's & St Thomas' issued the following press release:

The National Xeroderma Pigmentosum service, based at Guy's and St Thomas' Hospitals in London, received the Dermatology Team of the Year award at the Annual BMJ awards, in recognition of the outstanding care it provides to patients with XP.

The service was set up in 2010 by Dr Robert Sarkany, Consultant Dermatologist at St John's Institute of Dermatology at Guy's and St Thomas', in close collaboration with the XP patient group and NHS England's Highly Specialised Services Team.

As part of the service patients attend an annual multidisciplinary clinic where they can see a dermatologist, dermatological surgeon, ophthalmologist, neurologist, neuropsychologist, geneticist and specialist nurses. The specialist nurses also visit patients at home and give advice on, for example, fitting ultraviolet protective window films.

The service has improved patient care, reducing the number of appointments and amount of surgery. It has proved a cost-effective model of care for patients and saved money for the wider NHS.

Dr Bob Sarkany said:

"The National XP team is honoured to have been received the BMJ Dermatology Team of the Year award.

The XP team is a diverse group of nurses and doctors from six specialties, laboratory scientists, research academics and patient representatives. What unites us is a commitment to making life easier for patients and their families. The team's work is leading to patients having a longer life expectancy.

Sandra Webb, founder of the XP Support Group, recently described the tragedy of children with XP dying of skin cancer before the service was established, and how no child with XP in Britain has died of skin cancer since the service started.

This award is recognition of the dedication and ingenuity of the XP team. Thanks go too to NHS England's Highly Specialised Services team, who have supported the patients, families and service since its inception, and who have had the vision to provide an environment in which innovative clinical services can thrive."

Appendix A: European reference networks

European Reference Networks

The UK is a recognised leader in research on rare diseases, their treatment and care for those affected. The diagnosis, treatment and management of rare diseases requires the highest level of partnership working to remove unnecessary barriers. NHS England has encouraged collaboration at all levels and wherever possible to build upon the best research, diagnosis and service provision that already takes place in the UK and elsewhere.

The establishment of European Reference Networks (ERN) supports these objectives. They encompass the principles of better access for patients to highly specialised, safe care of the highest quality, support European co-operation on highly specialised healthcare, knowledge pooling, improving diagnosis and care in medical domains where expertise is rare. This type of collaboration can maximise the speed and scale of adoption and spread of innovations in medical science and health technologies. ERNs can also be focal points for medical training and research, information dissemination and evaluation.

To ensure there is oversight at a UK level, it has been agreed that applications from recognised healthcare providers must be endorsed by the Rare Diseases Advisory Group (RDAG), which is led by NHS England with representation from across the UK.

1. The European Union have approved the following UK providers to become part of the networks. The UK are members of 23 out of the 24 ERN networks. The detail listed in Annex 1 provides the detail of all the UK endorsed ERN centres.

UK endorsed ERN centres and the UK co-ordinating centres

Official name of ERN	ERN code	NHS trust
European Reference Network on Rare Bone Diseases	BOND	Alder Hey Children's NHS Foundation Trust
European Reference Network on Rare Bone Diseases	BOND	Birmingham Women's and Children's Hospital NHS Foundation Trust
European Reference Network on Rare Bone Diseases	BOND	University Hospitals Bristol NHS Foundation Trust
European Reference Network on Rare Bone Diseases	BOND	Manchester University NHS Foundation Trust
European Reference Network on Rare Bone Diseases	BOND	Great Ormond Street Hospital for Children NHS Foundation Trust
European Reference Network on Rare Bone Diseases	BOND	Guy's and St Thomas' NHS Foundation Trust
European Reference Network on Rare Bone Diseases	BOND	The Newcastle upon Tyne Hospitals NHS Foundation Trust
European Reference Network on Rare Bone Diseases	BOND	NHS Greater Glasgow and Clyde (NHS Scotland)
European Reference Network on Rare Bone Diseases	BOND	Oxford University Hospitals NHS Foundation Trust
European Reference Network on Rare Bone Diseases	BOND	Royal National Orthopaedic Hospital NHS Trust
European Reference Network on Rare Bone Diseases	BOND	Sheffield Children's NHS Foundation Trust
European Reference Network on Rare Bone Diseases	BOND	Sheffield Teaching Hospitals NHS Foundation Trust
European Reference Network on Rare Bone Diseases	BOND	University Hospitals Southampton NHS Foundation Trust
European Reference Network on Rare craniofacial anomalies and ENT disorders	CRANIO	Alder Hey Children's NHS Foundation Trust

Official name of ERN	ERN code	NHS trust
European Reference Network on Rare craniofacial anomalies and ENT disorders	CRANIO	Birmingham Women's and Children's Hospital NHS Foundation Trust
European Reference Network on Rare craniofacial anomalies and ENT disorders	CRANIO	Manchester University NHS Foundation Trust
European Reference Network on Rare craniofacial anomalies and ENT disorders	CRANIO	Great Ormond Street Hospital for Children NHS Foundation Trust
European Reference Network on Rare craniofacial anomalies and ENT disorders	CRANIO	NHS Tayside (NHS Scotland)
European Reference Network on Rare craniofacial anomalies and ENT disorders	CRANIO	Oxford University Hospitals NHS Foundation Trust
European Reference Network on Rare craniofacial anomalies and ENT disorders	CRANIO	Royal Free London NHS Foundation Trust
European Reference Network on Rare Endocrine Conditions	ENDO	Alder Hey Children's NHS Foundation Trust
European Reference Network on Rare Endocrine Conditions	ENDO	Barts Health NHS Trust
European Reference Network on Rare Endocrine Conditions	ENDO	University Hospitals Bristol NHS Foundation Trust
European Reference Network on Rare Endocrine Conditions	ENDO	Manchester University NHS Foundation Trust
European Reference Network on Rare Endocrine Conditions	ENDO	Great Ormond Street Hospital for Children NHS Foundation Trust – joint application with University College London Hospitals NHS Foundation Trust
European Reference Network on Rare Endocrine Conditions	ENDO	NHS Greater Glasgow and Clyde (NHS Scotland)
European Reference Network on Rare Endocrine Conditions	ENDO	University College London Hospitals NHS Foundation Trust – joint application with Great Ormond Street Hospital for Children NHS Foundation Trust

Official name of ERN	ERN code	NHS trust
European Reference Network on Rare Endocrine Conditions	ENDO	University Hospitals Birmingham NHS Foundation Trust
European Reference Network on Rare Endocrine Conditions	ENDO	University Hospitals Southampton NHS Foundation Trust
European Reference Network on Rare and Complex Epilepsies	EpiCARE	Great Ormond Street Hospital for Children NHS Foundation Trust
European Reference Network on Rare and Complex Epilepsies	EpiCARE	NHS Greater Glasgow and Clyde (NHS Scotland)
European Reference Network on Rare and Complex Epilepsies	EpiCARE	Oxford University Hospitals NHS Foundation Trust
European Reference Network on Rare and Complex Epilepsies	EpiCARE	University College London Hospitals NHS Foundation Trust
European Rare Kidney Diseases Reference Network	ERKNet	Birmingham Women's and Children's Hospital NHS Foundation Trust
European Rare Kidney Diseases Reference Network	ERKNet	Manchester University NHS Foundation Trust
European Rare Kidney Diseases Reference Network	ERKNet	Great Ormond Street Hospital for Children NHS Foundation Trust
European Rare Kidney Diseases Reference Network	ERKNet	The Newcastle upon Tyne Hospitals NHS Foundation Trust
European Rare Kidney Diseases Reference Network	ERKNet	Royal Free London NHS Foundation Trust
European Reference Network on Rare Hepatological Diseases	ERN Liver	Birmingham Women's and Children's Hospital NHS Foundation Trust
European Reference Network on Rare Hepatological Diseases	ERN Liver	The Newcastle upon Tyne Hospitals NHS Foundation Trust

Official name of ERN	ERN code	NHS trust
European Reference Network on Rare Hepatological Diseases	ERN Liver	Royal Free London NHS Foundation Trust
European Reference Network on Rare Hepatological Diseases	ERN Liver	University Hospitals Birmingham NHS Foundation Trust
European Reference Network on Rare Hepatological Diseases	ERN Liver	The Newcastle upon Tyne Hospitals NHS Foundation Trust
European Reference Network on Rare Eye Diseases	ERN-EYE	Manchester University NHS Foundation Trust
European Reference Network on Rare Eye Diseases	ERN-EYE	Leeds Teaching Hospitals NHS Trust
European Reference Network on Rare Eye Diseases	ERN-EYE	Moorfields Eye Hospital NHS Foundation Trust
European Reference Network on Rare Eye Diseases	ERN-EYE	Oxford University Hospitals NHS Foundation Trust
European Reference Network on Rare inherited and congenital anomalies	ERNICA	Great Ormond Street Hospital for Children NHS Foundation Trust
European Reference Network on Rare Respiratory Diseases	ERN-LUNG	Belfast Health and Social Care Trust (NHS Northern Ireland)
European Reference Network on Rare Respiratory Diseases	ERN-LUNG	Imperial College Healthcare NHS Trust
European Reference Network on Rare Respiratory Diseases	ERN-LUNG	The Newcastle upon Tyne Hospitals NHS Foundation Trust
European Reference Network on Rare Respiratory Diseases	ERN-LUNG	Ninewells Hospital and Medical School, Dundee (NHS Scotland)
European Reference Network on Rare Respiratory Diseases	ERN-LUNG	Royal Papworth Hospital NHS Foundation Trust
European Reference Network on Rare Respiratory Diseases	ERN-LUNG	Royal Brompton & Harefield Foundation Trust
European Reference Network on Rare Respiratory Diseases	ERN-LUNG	Royal Infirmary of Edinburgh (NHS Scotland)

Official name of ERN	ERN code	NHS trust
European Reference Network on Rare Respiratory Diseases	ERN-LUNG	University Hospitals Southampton NHS Foundation Trust
European Reference on Rare Neurological Diseases	ERN-RND	University College London Hospitals NHS Foundation Trust
European Reference Network on Rare and Undiagnosed Skin Disorders	ERN-SKIN	Barts Health NHS Trust
European Reference Network on Rare and Undiagnosed Skin Disorders	ERN-SKIN	Birmingham Women's and Children's Hospital NHS Foundation Trust
European Reference Network on Rare and Undiagnosed Skin Disorders	ERN-SKIN	Cardiff and Vale University Health Board (NHS Wales)
European Reference Network on Rare and Undiagnosed Skin Disorders	ERN-SKIN	Great Ormond Street Hospital for Children NHS Foundation Trust
European Reference Network on Rare and Undiagnosed Skin Disorders	ERN-SKIN	Guy's and St Thomas' NHS Foundation Trust
European Reference Network on Rare and Undiagnosed Skin Disorders	ERN-SKIN	Leeds Teaching Hospitals NHS Trust
European Reference Network on Rare and Undiagnosed Skin Disorders	ERN-SKIN	NHS Tayside (NHS Scotland)
European Reference Network on Rare Adult Cancers (solid tumours)	EURACAN	Imperial College Healthcare NHS Trust
European Reference Network on Rare Adult Cancers (solid tumours)	EURACAN	Oxford University Hospitals NHS Foundation Trust
European Reference Network on Rare Adult Cancers (solid tumours)	EURACAN	Royal Free London NHS Foundation Trust

Official name of ERN	ERN code	NHS trust
European Reference Network on Rare Adult Cancers (solid tumours)	EURACAN	Royal Marsden NHS Foundation Trust
European Reference Network on Rare Adult Cancers (solid tumours)	EURACAN	Sheffield Teaching Hospitals NHS Foundation Trust
European Reference Network on Rare Adult Cancers (solid tumours)	EURACAN	University College London Hospitals NHS Foundation Trust
European Reference Network on Rare Adult Cancers (solid tumours)	EURACAN	University Hospitals Coventry and Warwickshire NHS Trust
European Reference Network for Rare Neuromuscular Diseases	EURO NMD	Great Ormond Street Hospital for Children NHS Foundation Trust
European Reference Network for Rare Neuromuscular Diseases	EURO NMD	The Newcastle upon Tyne Hospitals NHS Foundation Trust
European Reference Network for Rare Neuromuscular Diseases	EURO NMD	Oxford University Hospitals NHS Foundation Trust
European Reference Network for Rare Neuromuscular Diseases	EURO NMD	University College London Hospitals NHS Foundation Trust
European Reference Network on Rare Haematological Diseases	EuroBloodNet	Barts Health NHS Trust
European Reference Network on Rare Haematological Diseases	EuroBloodNet	Guy's and St Thomas' NHS Foundation Trust
European Reference Network on Rare Haematological Diseases	EuroBloodNet	Imperial College Healthcare NHS Trust
European Reference Network on Rare Haematological Diseases	EuroBloodNet	Oxford University Hospitals NHS Foundation Trust

Official name of ERN	ERN code	NHS trust
European Reference Network on Rare Haematological Diseases	EuroBloodNet	Sheffield Teaching Hospitals NHS Foundation Trust
European Reference Network on Rare Haematological Diseases	EuroBloodNet	University College London Hospitals NHS Foundation Trust
European Reference Network for Rare and Complex Urogenital Diseases and Conditions	eUROGEN	Guy's and St Thomas' NHS Foundation Trust
European Reference Network for Rare and Complex Urogenital Diseases and Conditions	eUROGEN	King’s College Hospital NHS Foundation Trust
European Reference Network for Rare and Complex Urogenital Diseases and conditions	eUROGEN	Sheffield Teaching Hospitals NHS Foundation Trust
European Reference Network for Rare and Complex Urogenital Diseases and Conditions	eUROGEN	St George's University Hospitals NHS Foundation Trust
European Reference Network for Rare and Complex Urogenital Diseases and Conditions	eUROGEN	University College London Hospitals NHS Foundation Trust
European Reference Network on Genetic Tumour Risk Syndromes	GENTURIS	Cambridge University Hospitals NHS Foundation Trust
European Reference Network on Genetic Tumour Risk Syndromes	GENTURIS	Manchester University NHS Foundation Trust
European Reference Network on Genetic Tumour Risk Syndromes	GENTURIS	Guy's and St Thomas' NHS Foundation Trust
Gateway to Uncommon and Rare Diseases of the Heart	GUARD Heart	Barts Health NHS Trust

Official name of ERN	ERN code	NHS trust
Gateway to Uncommon and Rare Diseases of the Heart	GUARD Heart	Great Ormond Street Hospital for Children NHS Foundation Trust
Gateway to Uncommon and Rare Diseases of the Heart	GUARD Heart	St George's University Hospitals NHS Foundation Trust
European Reference Network on Rare Congenital Malformations and Rare Intellectual Disability	ITHACA	Birmingham Women's and Children's Hospital NHS Foundation Trust
European Reference Network on Rare Congenital Malformations and Rare Intellectual Disability	ITHACA	Manchester University NHS Foundation Trust
European Reference Network on Rare Congenital Malformations and Rare Intellectual Disability	ITHACA	Great Ormond Street Hospital for Children NHS Foundation Trust
European Reference Network for Rare Hereditary Metabolic Disorders	MetabERN	Birmingham Women's and Children's Hospital NHS Foundation Trust
European Reference Network for Rare Hereditary Metabolic Disorders	MetabERN	Bristol Royal Hospital for Children
European Reference Network for Rare Hereditary Metabolic Disorders	MetabERN	Manchester University NHS Foundation Trust
European Reference Network for Rare Hereditary Metabolic Disorders	MetabERN	Great Ormond Street Hospital for Children NHS Foundation Trust
European Reference Network for Rare Hereditary Metabolic Disorders	MetabERN	University Hospitals Birmingham NHS Foundation Trust
European Reference Network for Paediatric Cancer (haemato-oncology)	PaedCan	Birmingham Women's and Children's Hospital NHS Foundation Trust
European Reference Network for Paediatric Cancer (haemato-oncology)	PaedCan	Great Ormond Street Hospital for Children NHS Foundation Trust

Official name of ERN	ERN code	NHS trust
European Reference Network for Paediatric Cancer (haemato-oncology)	PaedCan	Royal Manchester Children's Hospital
Rare Immunodeficiency, Autoinflammatory and Autoimmune Diseases Network	RITA	Liverpool University Hospitals NHS Foundation Trust
Rare Immunodeficiency, Autoinflammatory and Autoimmune Diseases Network	RITA	Barts Health NHS Trust
Rare Immunodeficiency, Autoinflammatory and Autoimmune Diseases Network	RITA	Leeds Teaching Hospitals NHS Trust
Rare Immunodeficiency, Autoinflammatory and Autoimmune Diseases Network	RITA	The Newcastle upon Tyne Hospitals NHS Foundation Trust
Rare Immunodeficiency, Autoinflammatory and Autoimmune Diseases Network	RITA	Royal Free London NHS Foundation Trust
Rare Immunodeficiency, Autoinflammatory and Autoimmune Diseases Network	RITA	Sandwell and West Birmingham Hospitals NHS Trust
European Reference Network on Transplantation in Children (including HSCT, heart, kidney, liver, intestinal, lung and multi-organ)	TRANSCHILD	King’s College Hospital NHS Foundation Trust
European Reference Network on Rare Multisystemic Vascular Diseases	VASCern	Derby Teaching Hospitals NHS Foundation Trust
European Reference Network on Rare Multisystemic Vascular Diseases	VASCern	Guy's and St Thomas' NHS Foundation Trust

Official name of ERN	ERN code	NHS trust
European Reference Network on Rare Multisystemic Vascular Diseases	VASCern	Imperial College Healthcare NHS Trust
European Reference Network on Rare Multisystemic Vascular Diseases	VASCern	St George's University Hospitals NHS Foundation Trust

Appendix B: UK-wide commissioning arrangements for highly specialised services during 2018/19

Name of service	NHS England commissioning arrangements on behalf of the devolved administrations
Alkaptonuria service (adults)	Fully commissioned on behalf of England and Scotland
Alström syndrome service (adults and children)	Fully commissioned on behalf of England and Scotland
Ataxia telangiectasia services for adults	Fully commissioned on behalf of England and Scotland
Ataxia telangiectasia services for children	Fully commissioned on behalf of England and Scotland
Atypical haemolytic uraemic syndrome (adults and children)	Fully commissioned on behalf of England only
Auditory brainstem implant for patients with congenital abnormality of the auditory nerves or cochleae	Fully commissioned on behalf of England only
Autologous intestinal reconstruction service for adults	Fully commissioned on behalf of England only
Bardet–Biedl syndrome service (adults and children)	Fully commissioned on behalf of England and Scotland
Barth syndrome service (adults and children)	Fully commissioned on behalf of England and Scotland
Beckwith–Wiedemann syndrome with macroglossia service (children)	Fully commissioned on behalf of England and Scotland

Name of service	NHS England commissioning arrangements on behalf of the devolved administrations
Behçet's syndrome service (adults and adolescents)	Fully commissioned on behalf of England only
Bladder exstrophy service (children)	Fully commissioned on behalf of England and Scotland
Breast radiotherapy injury rehabilitation service (a discrete cohort of adult females)	Fully commissioned on behalf of England only
Cardiothoracic transplantation service (paediatrics)	Fully commissioned on behalf of England, in-part for Scotland by arrangement and in full for Northern Ireland
Choriocarcinoma service (adults and adolescents) gestational trophoblastic disease	Fully commissioned on behalf of UK (pre-1991)
Chronic pulmonary aspergillosis service (adults)	Fully commissioned on behalf of England and Scotland
Complex childhood osteogenesis imperfecta service	Fully commissioned on behalf of England only
Complex Ehlers Danlos syndrome service (adults and children)	Fully commissioned on behalf of England and Scotland
Complex neurofibromatosis type I service (adults and children)	Fully commissioned on behalf of England only
Complex tracheal disease service (children)	Fully commissioned on behalf of England and Scotland
Congenital hyperinsulinism service (children)	Fully commissioned on behalf of England and Scotland
Craniofacial service (adults and children)	Fully commissioned on behalf of UK (pre-1991)
Cryopyrin associated periodic fever syndromes (CAPS), also known as autoinflammatory diseases, treated with IL blockers	Fully commissioned on behalf of England and Scotland
Diagnostic service for amyloidosis (all ages)	Fully commissioned on behalf of England and Scotland
Diagnostic service for primary ciliary dyskinesia (adults and children)	Fully commissioned on behalf of England and Scotland

Name of service	NHS England commissioning arrangements on behalf of the devolved administrations
Diagnostic service for rare neuromuscular disorders (adults and children)	Fully commissioned on behalf of England and Scotland
Encapsulating peritoneal sclerosis treatment service (adults)	Fully commissioned on behalf of England only
Epidermolysis bullosa service (adults and children)	Fully commissioned on behalf of England and Scotland
Extracorporeal membrane oxygenation service for adults	Fully commissioned on behalf of England and Scotland
Extracorporeal membrane oxygenation service for neonates, infants and children with respiratory failure	Fully commissioned on behalf of England and Scotland
Ex-vivo partial nephrectomy service (adults)	Fully commissioned on behalf of England only
Gender identity development service for children and adolescents	Fully commissioned on behalf of England and Scotland
Heart transplantation service (adults)	Fully commissioned on behalf of England, in-part for Scotland by arrangement and in full for Northern Ireland
High consequence infectious diseases special isolation unit (airborne) (adults)	Fully commissioned on behalf of England, Scotland and Northern Ireland
High consequence infectious diseases, special isolation unit (airborne) (children aged 16 and under)	Fully commissioned on behalf of England, Scotland and Northern Ireland
Insulin resistant diabetes (adults and children)	Fully commissioned on behalf of England only
Islet transplantation service (adults)	Fully commissioned on behalf of England only
Live liver transplantation (adults)	Fully commissioned on behalf of England only
Liver transplantation service (adults)	Fully commissioned on behalf of England, Northern Ireland and Wales and by exception for Scotland
Liver transplantation service children	Fully commissioned on behalf of UK (pre-1991)

Name of service	NHS England commissioning arrangements on behalf of the devolved administrations
Lung transplantation service (adults)	Fully commissioned on behalf of England, in-part for Scotland by arrangement and in full for Northern Ireland
Lymphangioleiomyomatosis	Fully commissioned on behalf of England and Scotland
Lysosomal storage disorders service (children and adults)	Fully commissioned on behalf of England, and in-part for Scotland (service only not drugs) and for Northern Ireland (not ERT drugs)
McArdle's disease service (children)	Fully commissioned on behalf of England and Scotland
Multiple sclerosis management service for children	Fully commissioned on behalf of England only
Neurofibromatosis type 2 service (all ages)	Fully commissioned on behalf of England and Scotland
Neuromyelitis optica service (adults and children)	Fully commissioned on behalf of England and Scotland
Ocular oncology service (adults and adolescents)	Fully commissioned on behalf of England, from devolved administrations for Scotland
Ophthalmic pathology service (adults and children)	Fully commissioned on behalf of England, from devolved administrations for Scotland
Osteo-odonto-keratoprosthesis service for corneal blindness (adults)	Fully commissioned on behalf of England only
Pulmonary hypertension service for children	Fully commissioned on behalf of England and in part for Scotland (service only not drugs)
Pulmonary thromboendarterectomy service (adults and adolescents)	Fully commissioned on behalf of England and Scotland
Rare mitochondrial disorders service (adults and children)	Fully commissioned on behalf of England and Scotland
Retinoblastoma service (children)	Fully commissioned on behalf of UK (pre-1991)
Severe acute porphyria	Fully commissioned on behalf of England and Scotland

Name of service	NHS England commissioning arrangements on behalf of the devolved administrations
Severe combined immune deficiency and related disorders service (children)	Fully commissioned on behalf of England and in-part for Scotland
Small bowel transplantation service (adults)	Fully commissioned on behalf of England only
Small bowel transplantation service (children)	Fully commissioned on behalf of England and Scotland
Specialist paediatric liver disease service	Fully commissioned on behalf of UK (pre-1991)
Stickler syndrome diagnostic service (adults and children)	Fully commissioned on behalf of England and Scotland
Vein of Galen malformation service (adults and children)	Fully commissioned on behalf of England and Scotland
Ventricular assist devices as a bridge to heart transplantation or myocardial recovery (all ages)	Fully commissioned on behalf of England and in-part for Scotland
Wolfram syndrome service (adults and children)	Fully commissioned on behalf of England and Scotland