NHS England

HIGHLY SPECIALISED SERVICES 2018

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Description	This Highlight Report provides detailed information on selected services and gives summary information on all the services in the Highly Specialised services portfolio. This includes: which NHS providers deliver the services; the funding, activity, clinical outcomes and geographical equity access and the NHS England commissioning arrangements on behalf of the devolved nations.
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Highly Specialised Services 2018

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From 1 March 2018, information can be made available in alternative formats, such as easy read or large print, and may be available in alternative languages, upon request.

Contents

1 Introduction

1. The purpose of this document

The primary purpose of this document is to provide key information about highly specialised services. 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 of access to the service

In a small number of cases, some additional information is provided about the service in relation to service innovation, improvement and listening to and acting on patient feedback.

Appendix A details membership of European Reference Networks.

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

2. Equality Statement

Promoting equality and addressing health inequalities are at the heart of NHS England'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.

3. Specialised services

NHS England is responsible for commissioning £16.4 billion (2017/18 budget) of specialised services to meet a wide range of health and care needs.

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; and
- The financial implications for CCGs if they were required to arrange for provision of the service or facility themselves.

4. 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 permits the most effective use of resources by 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 particular 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.

A key task for the HSCT is to liaise closely with a range of stakeholders both within NHS England and with other legal entities, especially:

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

5. Rare Diseases Advisory Group (RDAG)

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.

6. Expenditure figures

The expenditure figures for each service reflect NHS England's expenditure on that service in 2017/18. Expenditure is set out into the following categories:

- Less than £0.5m
- More than £0.5m but less than £1m
- More than £1m but less than £5m
- More than £5 million but less than £10m
- More than £10 million but less than £20m
- More than £20 million but less than £30m
- More than £30 million but less than £50m
- More than £50m

7. 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 unit 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.

8. 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 implied by this ethos is that access may be difficult for patients who live remotely from the expert centres. Hence it is incumbent on the HSCT to monitor the geographical access to highly specialised services.

The best measure is a metric known as the standardised coefficient of variation (SCV). An SCV above 20 indicates variation greater than expected by chance. Where the SCV is below 20, variation can be considered random. For each service, patients have been 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 (e.g. transplant) or have a confirmed diagnosis of the relevant condition (e.g. Xeroderma pigmentosum).

Where variation is observed which is likely not due to chance (SCV greater than 20), it is possible that there are genuine clusters of disease. For example, many genetic disorders are commoner among highly consanguineous populations which are themselves unequally distributed in England. Conversely, there may be a genuine lack of patients with a particular condition – for example severe osteogenesis imperfecta is unlikely to be missed and there are fewer patients identified with this condition in the North East of England.

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

The HSCT routinely undertakes an analysis of geographical equity for each service every three years.

2 Services and Providers of Highly Specialised Services for 2017/18

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
Expenditure	Between £0.5m and £1m
Caseload	53
Outcomes collated:	Median SF36 score for patients treated in the service for 12 months or longer: • SF36 stable Median AKU Severity Score for patients treated in the service for 12 months or longer: • AKUSSI improved, Ochronosis [i] scores reversed. NB: Ochronosis is a condition in which the body cannot break down a toxic acid called homogentisic acid. This causes bones and cartilage to become black and brittle. The spine collapses. Prostate and kidney stones appear. Heart valves get blocked and heart surgery is needed.
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 grows in the heart and stops it working effectively) and renal failure. They may also become Deaf. There are thought to be fewer than 100 people affected by Alström syndrome in England.

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

Following this review, a management plan for local care providers is agreed and communicated to local health care professionals to allow them to implement the recommendations and monitor 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; University Hospitals Birmingham NHS Foundation Trust
Expenditure	Less than £0.5m
Caseload	78
Outcomes collated	Percentage of children in HbA1C in target range; • Birmingham Children's Hospital 80% Percentage of adults with Hba1C <75 mmol/mol; • University Hospitals Birmingham 80% Median age at death of patients on active caseload • University Hospitals Birmingham: no deaths in 2017/18 NB: Glycated haemoglobin (HbA1C) is a form of haemoglobin that is measured primarily to identify the three-month average plasma glucose concentration
Geographical equity access	Numbers too small to analyse

Improvements – listening to and acting on feedback

In the Alström clinic in Birmingham, young people and their families spend a whole day in clinic, being reviewed and tested by a multidisciplinary team of specialists.

Whilst this means that the young people see all their specialists on the same day, the experience can be stressful and the tests can sometimes be painful and uncomfortable. Previously young people and families spent a long and unpredictable

amount of time in the ophthalmology department at the beginning of their consultation day. This was exacerbating their discomfort and having a negative impact on their other appointments throughout the day.

Following feedback from the young patients and their parents, the ophthalmologist listened and worked with colleagues to change the schedule, allocating their afternoon to the Alström clinic; and conducting consultations alongside other specialists within a designated area for the clinic.

This simple change to times and location within the hospital has significantly reduced the amount of waiting around time, leading to young people being less anxious and saying that their experience in clinic has improved dramatically.

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 leukaemias, lymphoma, pneumonia, chronic lung disease and neurological decline. Fewer than 100 adults in England have AT.

The service undertakes annual multi-disciplinary 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 health care professionals to implement the recommendations and monitor their progress.

NHS Centre	Papworth Hospital NHS Foundation Trust
Expenditure	Less than £0.5m
Caseload	75
Outcomes collated	Median age at death: 35 (this is higher than the median cited in medical publications of 20 years) Median BMI: 22.5 interquartile range 18.4-25.9 kg/m2 (this is an important measure because the nature of the condition often means that patients do not achieve optimum BMIs).
Geographical equity access	No evidence of geographical inequity

Innovation

The ataxia telangiectasia (AT) service for adults has introduced an MRI breast screening programme for female AT patients in liaison with the breast screening service at Addenbrooke's Hospital. This started in autumn 2017.

All female AT adults over 25 are offered annual screening as recommended by national breast screening guidance because AT patients are at very high risk of developing malignancy, especially breast tumours.

Many patients were having difficulty accessing this service locally and it made sense to make it part of their annual review at the specialist centre.

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 health care professionals to implement the recommendations and monitor their progress.	
NHS Centre	Nottingham University Hospitals NHS Trust
Expenditure	Less than £0.5m
Caseload	128
Outcomes collated	Percentage of patients with previously unrecognised treatable or untreatable morbidity: • 85.3% Percentage of patients for whom active intervention was undertaken in clinic or arranged locally: • 52.9%
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 appears slightly more frequently 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 2.7–5.5 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	More than £1m but less than £5m
Caseload	143
Outcomes collated	Number of deaths in patients with a diagnosis of complement mediated aHUS: • No patient in England died of aHUS in 2017/18
Geographical equity access	New service

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	Less than £0.5m
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 multi-disciplinary 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 health care 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; Great Ormond Street Hospital for Children NHS Foundation Trust; Guy's and St Thomas' NHS Foundation Trust; University Hospitals Birmingham NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Assessments	240
Outcomes collated	Percentage of children with HbA1C in target range: • Birmingham Children's Hospital: 95% • Great Ormond Street Hospital: 98% Percentage of adult patients with Hba1C <75 mmol/mol; • University Hospitals Birmingham: 94.5% • Guy's and St Thomas': 84% Percentage of adult patients with a BMI <35; • University Hospitals Birmingham: 46% • Guy's and St Thomas': 55.6% NB: Glycated haemoglobin (HbA1C) is a form of haemoglobin that is measured primarily to identify the three-month average plasma glucose concentration
Geographical equity access	Data not available or not comparable

Barth syndrome service (male adults and children)	
Barth syndrome is an x-linked disorder of lipid metabolism presenting as cardiac/skeletal myopathy, neutropenia and growth retardation with a high infant mortality rate. Patients with Barth Syndrome present with frequent cardiac problems and, in two-thirds of patients, neutropenia (reduced white blood cell count leading to susceptibility to infection). 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 multi-disciplinary 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	More than £0.5m but less than £1m
Caseload	Data suppressed to maintain patient confidentiality
Outcomes collated	Median age at diagnosis: • Six weeks Median age at death: • Three years
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, i.e. about 15-20 babies each year. The service provides multi-disciplinary, 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	Less than £0.5m
Caseload	179
Outcomes collated	% of improvement at the 3 - 6 months post-operative assessment of resting tongue position: • 100% % of improvement at the 3-6 month post-operative assessment in the reduction or cessation of drooling: • 100% % of improvement at the 3-6 month post-operative assessment in the reduction or elimination of macroglossia-related errors: • 100%
Geographical equity access	Numbers too small to analyse
Innovation In the Beckwith Wiedemann syndrome service, a telemedicine clinic was piloted for the post-operative patient reviews. Feedback has been obtained systematically from families following these appointments, which has been consistently positive. Key benefits are time and travel cost savings for families. Clinicians have conducted successful reviews and given key management advice at this stage of the children's recovery. Given the success and benefits of these clinics, the service plans to implement them on an ongoing basis.	

Behçet's syndrome service (adults and adolescents)	
Behçet's syndrome is a chronic, inflammatory, multi-systemic vasculitic disorder with a wide spectrum of clinical presentations that may include blindness, severe ulceration and cardiovascular problems. There are around 1,500 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; Barts Health NHS Trust; Sandwell & West Birmingham Hospitals NHS Trust
Expenditure	More than £1m but less than £5m
Caseload	1,494
Outcomes collated	Median number of flares per patient during the previous 12 months • Aintree: 1 • Barts Health: 0.5 • Sandwell & West Birmingham: 0
Geographical equity access	Evidence of geographical inequity is being investigated but is likely due to data quality issues

Bladder exstrophy service (children)	
The service provides diagnostic, 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 a 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 multi-disciplinary 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	More than £1m but less than £5m
New babies	Data suppressed to maintain patient confidentiality
Outcome collated	Percentage closure achieved without dehiscence: • Great Ormond Street Hospital: 90% • Manchester University Hospitals: 100% Percentage continent (dry by day) at age five years - target 50% • Great Ormond Street Hospital: 45% • Manchester University Hospitals: 89%
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, multi-disciplinary rehabilitation service. A lead centre provides an inpatient service.	
NHS Centre	Royal National Hospital for Rheumatic Diseases – Royal United Hospitals Bath NHS Foundation Trust
Expenditure	More than £0.5m but less than £1m
Caseload	71
Outcomes collated	Improvement in mood and in upper limb: • 30% improvement Median Brief Pain Inventory Score: • 0 Median Acceptance & Action Questionnaire Score: • 22 Median Upper Extremity Function Index • 40 These are good scores for patients who have been treated for severe health problems.
Geographical equity access	Data not available or not comparable

Innovation

The Royal National Hospital for Rheumatic Diseases in Bath has developed special expertise in treating complex pain syndromes. This service is primarily focused on a small group of women who received a particular regime of radiotherapy for breast cancers in the 1990s. Although this regime had certain advantages, over time it became clear that a small but important group of women who received it developed complex pain syndromes, and other disabilities. This was due to nerve damage in structures adjacent to the radiotherapy target area.

The highly specialised service has offered assessment and treatment options to all women who suffer from this injury syndrome. Some women suffer considerably from restricted use of their arm on the affected side; expert physiotherapy and occupational therapy can improve function and reduce the associated disability. As with many highly specialised services, the model of care is based on a close partnership between the expert centres and ongoing care from the local service where the patient lives.

Cardiothoracic transplantation service (paediatrics)	
The heart and lung transplant service provides: assessment of patients who are eligible for a heart transplant; the transplant operation; and lifelong follow up.	
NHS Centres	Great Ormond Street Hospital for Children NHS Foundation Trust; The Newcastle Upon Tyne Hospitals NHS Foundation Trust
Expenditure	More than £50m (adults and children, heart and lung)
Number of transplants	34
Outcomes collated	30-day unadjusted patient survival rates after first paediatric heart only transplant: • GOSH: 96.8% • Newcastle: 92.3% One-year unadjusted patient survival rates after first paediatric heart only transplant by centre: • GOSH: 93.3% • Newcastle: 89.1% Five-year unadjusted patient survival after first paediatric heart only transplant by centre: • GOSH: 81.3% • Newcastle: 83.8% 90-day patient survival rates after first paediatric lung transplants by centre • GOSH: 93.1% Newcastle: survival rates for groups with less than 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 foetus 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	More than £1m but less than £5m
Inpatient Episodes	547
Outcomes collated	Deaths as percentage of new cases each year • Imperial College: 0 • Sheffield Teaching Hospitals: 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	More than £5 million but less than £10m
Caseload	473
Outcomes collated	• Percentage of patients showing a 12-point improvement or more in the SGRQ AND 3KG weight gain or more:43.6% NB: This is a substantial improvement in quality of life in a condition which, if untreated, shows no improvement at all.
Geographical equity access	Strong evidence of geographical inequity which is long-standing and 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 drastically 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 16's who have this rare, genetic collagen deficiency.	
NHS Centres	Birmingham 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	More than £1m but less than £5m
Caseload	307
Outcomes collated	Median number of new non-vertebral fractures; • Birmingham Children's: 2 • GOSH: 2 • Bristol: 1 • Sheffield: 2 Median number of new vertebral fractures: • Birmingham Children's: 1 • GOSH: 2.5 • Bristol: 2 • Sheffield: 3 % patients with scoliosis and Cobb angle> 45 degrees (Cobb angle is defined as a measurement for determining the degree of an abnormal lateral spinal curve): • Birmingham Children's: 5.3% • GOSH: 11% • Bristol: 5.3% • Sheffield: 8.7%
Geographical equity access	No evidence of geographical inequity

Innovation
During 2017/18, all four designated providers receiving recognition as Centres of Excellence within the European Reference Network for Rare Bone Disease (ERN BOND), creating opportunities to share expertise and improve the quality of care and outcomes for patients with OI.

Across the 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.

Complex Ehlers Danlos syndrome service (adults and children)	
Ehlers-Danlos syndrome (EDS) is a group of heritable disorders of connective tissue. The major 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 wall of the aorta separate. There are several types of EDS each with their own specific management. The service diagnoses about 200 new patients each year with the rare forms of EDS. The service provides a fully comprehensive service under the auspices of the clinical genetics service for the precise clinical diagnosis and management of a sub-set of patients with all types of EDS in whom either the clinical diagnosis is not straight forward, or the clinical diagnosis is one of EDS but laboratory testing has not confirmed the diagnosis and further clinical evaluation is necessary. The service diagnoses about 200 new patients each year with the rare forms of EDS.	
NHS Centres	London North West Healthcare NHS Trust; Sheffield Children's NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Definitive diagnosis	EDS confirmed in 196 patients
Outcomes collated	% patients with a definitive diagnosis or diagnosis ruled out: • North West Healthcare: 40% of patients with definitive EDS diagnosis and 60% EDS ruled out • Sheffield: diagnosis or diagnosis ruled out 99% % patients with a genetic diagnosis: • North West Healthcare: 40% • Sheffield: 45%
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 formation of neurofibromas (tumours involving nerve tissue) in the skin, subcutaneous tissue, cranial nerves and spinal root nerve tissue. About 1 in 25,000 of the population has NF1. With *complex* NF1 there is a high risk of developing rare complications, which may affect most of the body systems.

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

The service includes:

- Specialist assessment of NF1 patients with suspected complex complications of the disease to provide accurate diagnosis of unusual phenotypes and other disease that can be mistaken for NF1. This is through genetic testing with support from genetic counselling
- Co-ordination of care by a specialist multi-disciplinary team when the NF1 complications mean the condition manifests differently to other NF1 patients
- Monitoring the risk of NF1-related malignancy and tumour progression
- Long-term monitoring to evaluate the need for surgery, for example, cervical cord compression

NHS Centres	Guy's and St Thomas' NHS Foundation Trust; Manchester University NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Outpatient Attendances	898
Outcomes collated	Total number of interventions facilitated: • Guys: 125 interventions out of a total of 442 patients • Manchester: 62 interventions out of a total of 314 patients
Geographical equity access	There is evidence of geographical inequity and the establishment of a new shared care clinic should help to resolve this issue.

Innovation

In the London centre of the neurofibromatosis type 1 (NF1) service, a clinical psychologist offers weekly clinics and telephone support to help illness adjustment and anxiety in adults with NF1. There is also a new neuro-disability paediatrician who is reducing the paediatric waiting list and focusing on children with chronic neurological problems.

There are weekly clinical nurse specialist phone clinics to support complex NF1 patients in between clinic visits, lessening the need for face to face clinics, helping people who live a long distance from the Trust and triaging sick patients for urgent

assessment.

The Trust has expanded transition days for young people for information, peer mentoring and networking.

The Trust are involved in international engagement through; sharing their template of care to help colleagues set up an NF1 clinic in Sydney, Australia and also co-chaired an international NF conference in Washington USA to improve clinical care and develop targeted therapy.

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	More than £1m but less than £5m
Inpatient Episodes	72
Outcomes collated	Percentage at one-year survival: • 100%
Geographical equity access	Numbers too small to analyse

Congenital hyperinsulinism service (children)

Congenital hyperinsulinism (CHI) is a condition characterised by excess insulin production from the pancreas, resulting in hypoglycaemia. The clinical presentation and progress of CHI lies on a spectrum, varying between those with transient hypoglycaemia to those unresponsive to medical treatment and requiring pancreatectomy. In the absence of expert management, children who have prolonged or recurrent hyperinsulinaemic hypoglycaemia in infancy can suffer harm to their brains and may be developmentally delayed.

The service diagnoses patients (usually in the newborn period) and patients are referred to one of the national centres. If immediate transfer cannot be arranged, then the provider supports the referring unit to provide appropriate care for the patient. The nationally designated provider 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 which works jointly with Alder Hey Children's NHS Foundation Trust which together form 'NORCHI'
Expenditure	More than £1m but less than £5m
Caseload	1148
Outcome collated	Incidence of deaths in patients with CHI as a consequence of CHI: • GOSH: 0 • NORCHI: 0 Unplanned admissions due to CHI and admitted under paediatric endocrine team, day cases excluded: • GOSH: 20 • NORCHI: 30
Geographical equity access	Data not available or not comparable

Innovation

In the congenital hyperinsulinism service, the two North centres work together as a single team (known as NORCHI) and the service, has introduced joint multi-disciplinary team videoconference meetings across all three units.

The service is also developing a prototype App for CHI patients and their families and using continuous glucose monitoring for patients with hypoglycaemia.

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; Great Ormond Street Hospital for Children NHS Foundation Trust Oxford University Hospitals NHS Trust
Expenditure	More than £10 million but less than £20m
Assessments	143
Outcomes collated	Percentage of patients with grade 3 or 4 surgical complications (should decrease or remain the same): • Alder Hey Children's: 5% • Birmingham Children's: 3.4% • GOSH: Grade 3: 2.6% and Grade 4: 0% • Oxford: 0%
Geographical equity access	Data not available or not comparable

Innovation

The Oxford craniofacial unit speech and language team (SaLT) applied and received funding from The Health Foundation for the 'Sing and Say' project. 'Sing and Say' provides a specialist set of language resources for children with craniofacial conditions and their families to use, including those with complex difficulties with communication, or with eating, drinking and swallowing.

Using an original song by Andy Stevens, Registered Music Therapist, SaLT colleagues invited children with craniosynostosis to star in the video of the song. This can be found here: https://www.ouh.nhs.uk/singandsay/shooting-star/default.aspx

These resources are now being used across all four centres providing complex craniofacial surgery as part of the scale and spread of this innovation.

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. The service treats a caseload of about 115 patients.	
NHS Centre	Royal Free London NHS Foundation Trust
Expenditure	More than £5 million but less than £10m
Patients on high cost drugs	128
Outcomes collated	Median 20-point CAPS activity score (should decrease or remain the same): • 1/20
Geographical equity access	Data not available or not comparable

Diagnostic service for amyloidosis (adults)	
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 supporting the evaluation of existing and new therapies. The service provides a diagnostic service to about 1,200 new patients each year.	
NHS Centre	Royal Free London NHS Foundation Trust
Expenditure	More than £5m but less than £10m
First evaluations	1,318
Outcomes collated	Percentage of patients with a definitive diagnosis or diagnosis ruled out: • 99.9%
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 Hospital Southampton NHS Foundation Trust; University Hospitals of Leicester NHS Trust
Expenditure	More than £1m but less than £5m (for management and diagnostic elements)
Number of positive samples	68
Outcomes collated	Outcomes captured by PCD management service
Geographical equity access	Data not available or not comparable

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. There are about 1,600 referrals to the service each year.	
NHS Centres	Great Ormond Street Hospital for Children NHS Foundation Trust; Oxford University Hospitals NHS Trust; The Newcastle Upon Tyne Hospitals NHS Foundation Trust; University College London Hospital NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Number of referrals	1,320
Outcomes collated	Percentage of patients with a genetic diagnosis: • GOSH & UCLH combined: 31% • Oxford: 70.1% • Newcastle: 41% NB: To note that 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	More than £1m but less than £5m
Primary surgical procedures	Data suppressed to maintain patient confidentiality
Outcomes collated	94% one-year survival post-operation for renal cases across the centres 100% of patients TPN free post-operation across the centres
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; Great Ormond Street Hospital for Children NHS Foundation Trust; Guy's and St Thomas' NHS Foundation Trust; Heart Of England NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Caseload	675 (caseload based on three centres)
Outcomes collated	Median quality of life score at transition (adults only) (QoLEB); • Guy's: 12 Mean number of unplanned admissions among patients with recessive dystrophic EB: • Birmingham Children's and Heart of England combined: 1/20 • GOSH: 31/19 • Guy's: 10/72 NB: Recessive dystrophic EB is the most severe types of dystrophic epidermolysis bullosa.
Geographical equity access	Data not available or not comparable

Improvements - listening to and acting on feedback

Epidermolysis bullosa (EB) is a rare genetic condition in which the skin is excessively fragile and easily blisters in response to trauma. Starting nursery or a new school is naturally an anxious time for all concerned.

Parents have concerns about their child being exposed to the rough-and-tumble of nursery and school and want to know that nursery and school staff are competent in these procedures.

School staff share similar concerns as parents and difficulties include identifying an appropriate staff member, finding a clean, safe area to perform dressings and integrating the child into all activities.

The child will need to learn to trust school staff managing their skin care and wants

to be treated normally and to be allowed to play and participate fully without being singled out.

The EB Team, with an ever-growing case-load, cannot visit all nurseries and schools individually.

The solution is to hold a study day for nursery and school staff three times a year to coincide with key times during the academic year. This is substantially more time and cost-efficient, enabling the nurses to provide more direct patient care. It also acts as a useful resource for new EB team staff. The event is free to attend and includes:

- Overview of EB and other skin fragility conditions
- Management of the child in an educational setting
- Awareness of risks and assessments
- Practical sessions that are enhanced using video presentations on, for example, how to lance blisters
- Written guidance on how to, for example, write a care plan to form part of the Educational Health Care Plan

Extra corporeal membrane oxygenation service for adults	
Extra corporeal 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; Royal Brompton & Harefield NHS Foundation Trust; University Hospitals of Leicester NHS Trust
Expenditure	More than £30m but less than £50m (adults and children)
Starting treatment	395
Outcomes collated	Percentage survival at discharge: • Guy's: 71% • Manchester: 62% • Papworth: 78% • Royal Brompton: 75.6% • Leicester: 83.3%
Geographical equity access	There is some evidence of geographical inequity in the service. The issues are understood, and the inequity continues to be monitored and it is anticipated that the analysis will be re-run in 2018/19.

Extra corporeal membrane oxygenation service for neonates, infants and children with respiratory failure	
Extra corporeal membrane oxygenation (ECMO) supports critically ill babies and children who have severe potentially reversible acute respiratory failure by oxygenating the 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	More than £30m but less than £50m (adults and children)
Starting treatment	77
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 post-operative recovery; and long-term follow-up.	
NHS Centre	Oxford University Hospitals NHS Trust
Expenditure	Less than £0.5m
Patients Accepted into service	Data suppressed to maintain patient confidentiality
Outcomes collated	Percentage one-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, the service works with paediatric endocrinology clinics to 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; Leeds Teaching Hospitals NHS Trust
Expenditure	More than £5m but less than £10m
Number of referrals	2,233
Outcomes collated	Median change in Clinical Global Assessment Score between first appointment and follow up (increase indicates improvement) • Median change of 1% when comparing the first and last measurements of the Children's Global Assessment Scale score. NB Children's global assessment scale 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 Centre	Leeds Teaching Hospitals NHS Trust
Expenditure	Less than £0.5m
Outpatient assessments	81
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; 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	More than £50m (adult and children, heart and lung)
Number of transplants	149
Outcomes collated	30-day risk-adjusted patient survival rates after first adult heart transplant by centre: • Birmingham 88.3% • Brompton & Harefield: 90.2% • Manchester: 93.1% • Newcastle: 88.1% • Papworth: 95.3% One-year risk-adjusted patient survival rates after first adult heart transplant by centre: • Birmingham: 78.8% • Brompton & Harefield: 82.1% • Manchester: 83.8% • Newcastle: 79.9% • Papworth: 90.0% Five-year risk-adjusted patient survival rates from listing for first heart transplants: • Birmingham: 61.5%; • Brompton & Harefield:66.8%; • Manchester: 73.4%; • Newcastle: 54.2%; • Papworth: 70.7%
Geographical equity access	Some evidence of geographical inequity which are thought to be of a temporary nature

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 clinical carers, 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	Less than £0.5m
Active caseload	184
Outcomes collated	Percentage of patients with specific diagnosis: • 87% Percentage of patients maintaining HbA1c below 75 mmol/mol: • 75.9% NB: Glycated haemoglobin (HbA1C) is a form of haemoglobin that is measured primarily to identify the three-month average plasma glucose concentration
Geographical equity access	Strong evidence of geographical inequity the reasons for which are not fully understood so 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, whilst 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	More than £1m but less than £5m
Number of Transplants	Data suppressed to maintain patient confidentiality
Outcomes collated	Median episodes of severe hypoglycaemia events in patients who were transplanted in the previous 12 months: • Median number of severe hypoglycaemia events between registration and transplant is zero compared to seven events in the 12 months pre-transplant Percentage of patients with HbA1c <75 mmol/mol who were transplanted in the previous 12 months: • Reduction in median HbA1c (mmol/mol) for routine islet transplants dropped from 64mmol/mol prior to transplant to 51mmol/mol at one-year post transplant NB: Glycated haemoglobin (HbA1C) is a form of haemoglobin that is measured primarily to identify the three-month average plasma glucose concentration
Geographical equity access	Numbers too small to analyse

Liver and live liver transplantation service (adults)	
This service provides assessment, transplantation and lifelong follow up for patients requiring liver transplant surgery, including from living donors. The three main conditions 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	More than £50m (adults and children)
Number of transplants	632
Outcomes collated	One-year risk-adjusted patient survival for adult elective deceased donor first liver transplants: • Cambridge: 94.9% • King's: 94.6%; • Leeds: 92.2% • Royal Free: 94.3% • Newcastle: 92.0% • Birmingham: 92.3% Five-year risk-adjusted patient survival for adult elective deceased donor first liver transplants: • Cambridge: 87.2% • King's: 83% • Leeds: excluded due to a lack of follow up beyond 12 months • Royal Free: 81.3% • Newcastle: 76.7% • Birmingham: 78.3% Five-year risk adjusted patient survival rates from listing for adult elective first liver registrations: • Cambridge: 74% • King's: 74% • Leeds: have been excluded due to a lack of follow up beyond 12 months • Royal Free: 70% • Newcastle: 68% • Birmingham: 67%

	The national rates of patient survival after joining the transplant list for adult elective first liver only patients is 83% at one, 70% at five and 57% at ten years post-registration.
Geographical equity access	No evidence of geographical inequity

Innovation

Liver transplantation is a life-saving procedure for people with liver failure; the median age at transplant is 55 years old, and a successful transplant gives on average an extra 20 years of life. A wide variety of diseases may result in liver failure but the commonest causes of liver failure requiring transplant are alcoholic liver disease and cancer.

Unlike kidney failure, where dialysis provides an alternative, the only treatment option for liver failure is a transplant. In the UK, the eligibility rules for transplant are designed to produce a broad balance between the number of donated organs and the number of patients listed. Although this means more restrictive criteria than used, for example, in the USA, it means patient on the waiting list stand a very good chance of actually receiving an organ.

Transplant centres in the UK pioneered the development of DCD or 'deceased cardiac death' organs as an alternative to organs donated after brain death (DBD). Almost 20% of liver transplants nowadays are from DCD donors. There is also a small programme of adult-to-adult living donor transplant.

Transplant surgery is a complex multi-disciplinary effort requiring close collaboration between surgeons, anaesthetists, hepatologists and many others; the national programme also requires strong organisational collaboration between NHS England, NHS Blood and Transplant and the providers.

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; King's College Hospital NHS Foundation Trust; Leeds Teaching Hospitals NHS Trust
Expenditure	More than £50m (adults and children)
Number of transplants	99
Outcomes collated	One-year unadjusted patient survival for paediatric elective deceased donor first liver transplants: • Birmingham: 96.1% • King's: 97.2% • Leeds: 100% Five-year unadjusted patient survival for paediatric elective deceased donor first liver transplants: • Birmingham: 89.0% • King's: 94.5% • Leeds: 91.4%
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; The Newcastle Upon Tyne Hospitals NHS Foundation Trust; University Hospitals Birmingham NHS Foundation Trust
Expenditure	More than £50m (adult and children, heart and lung)
Number of transplants	142
Outcomes collated	90-day risk-adjusted patient survival rates after first adult lung transplant by centre: • Brompton & Harefield 90.1% • Manchester: 92.8% • Papworth: 92.9% • Newcastle: 87.1% • Birmingham: 81.6% One-year risk-adjusted patient survival rates after first adult lung transplant by centre: • Brompton & Harefield 81.9% • Manchester: 85.1% • Papworth: 79.8% • Newcastle: 79.2 • Birmingham: 68.6% Five-year risk-adjusted patient survival rates from listing for first lung only transplants: • Brompton & Harefield 52.8% • Manchester: 48.7% • Papworth: 44.2% • Newcastle: 47.1% • Birmingham: 30.1
Geographical equity access	No evidence of geographical inequity

Lymphangioleiomyomatosis (LAM) is a rare, progressive disease characterised by lung cysts, kidney tumours and lymphatic abnormalities. LAM occurs in a sporadic form, which affects only females, 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
- Inpatient care: for management of complications and in some cases diagnostic workup
- Evaluation of patients with TSC or suspected TSC: support for a small number of patients who may require further genetics investigations including genotyping
- Surgical treatment: when clinically indicated, video assisted thorascopic lung biopsy is utilised. Liaison with surgical colleagues is also required for effective management of pneumothorax and pleural effusion
- Management of renal angiomyolipoma (benign tumour of the kidney): case discussion and surgery where appropriate [This service is provided outside of the Highly Specialist Lymphangioleiomyomatosis Centres as part of a sub-contracting arrangement.]
- Lung transplant referral

Fewer than 100 women in England have LAM.

NHS Centre	Nottingham University Hospitals NHS Trust
Expenditure	Less than £0.5m
Caseload	60
Outcomes collated	Percentage of patients having a pneumothorax: • 2.27% Percentage of patients having a renal angiolipoma bleed: • 0% Percentage of patients having an FEV1 decline of more than 150 ml per annum: • 15.3%
Geographical equity access	Evidence of geographical inequity. This is caused by the expert centre's ability to detect very mild forms of the disease which would be missed elsewhere.

Lysosomal storage disorders service (children & 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; 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 Hospital NHS Foundation Trust; University Hospitals Birmingham NHS Foundation Trust
Expenditure	More than £50m
Active Caseload	2,328 (Of these, the total number of patients receiving enzyme replacement therapy and substrate reduction therapy drugs is 1,046)
Outcomes collated	Fabry 1 Percentage of patients initiating renal replacement therapy among patients treated for 3 years or more: • Birmingham Children's: 0% • Cambridge: 0% • GOSH: 0% • Manchester: 0% • Royal Free: 4.1% • Salford: 0% • UCLH: 1% • University Hospitals Birmingham: 0% Fabry 2 Percentage of patients having a new stroke among patients treated for 3 years or more:

	• Birmingham Children's: 0% • Cambridge: 1% • GOSH: 0% • Manchester: 0% • Royal Free: 11% • Salford: 2.58% • UCLH: 2% • University Hospitals Birmingham: 3% Fabry 3 Percentage of patients having a cardiac device implanted among patients treated for 3 years or more: • Birmingham Children's: 0% • Cambridge: 2% • GOSH: 0% • Manchester: 0% • Royal Free: 19.9% • Salford: 3.32% • UCLH: 1% • University Hospitals Birmingham: 5% Gaucher Percentage of patients having a hospital admission for bone crisis among patients treated for 3 years or more: • Birmingham Children's: 0% • Cambridge: 0% • GOSH: 0% • Manchester: 0% • Royal Free: 1.7% • Salford: 0% • UCLH: 0% • University Hospitals Birmingham: 0% MPS Percentage of patients having a new cranio cervical episode among patients treated for 3 years or more: • Birmingham Children's: 0% • Cambridge: 0% • GOSH: 0% • Manchester: 0% • Royal Free: 0% • Salford: 0% • UCLH: 0% • University Hospitals Birmingham: 5%
Geographical equity access	New data definition proposed

McArdle's disease service (children)	
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 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. There are fewer than 200 people in England with McArdle's disease.	
NHS Centre	University College London Hospital NHS Foundation Trust
Expenditure	More than £0.5m but less than £1m
Caseload	201
Outcomes collated	Median functional capacity - 12MWD: • Median: 805m Number of patients requiring hospital assessment requiring hospital assessment: • 3 admissions out of 55 patients on follow-up Median Quality of Life (SF36) score: • Physical Functioning: 34.6 NB: 12MWD is a twelve-minute walking distance test used to estimate functional exercise capacity.
Geographical equity access	No evidence of geographical inequity

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 There are about 900 people in England who have NF2.	
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	More than £5 million but less than £10m
Caseload	978
Outcomes collated	Median length of time that useful hearing as measured by speech discrimination (>60%) is maintained in at least one ear from date of diagnosis for greater than three years • Cambridge: 44 months (time from diagnosis ranges from 1 – 95 months) • Guys: the median figure for those patients preserving hearing to the end of follow up is 40.5 months. The median duration for which hearing was preserved in those patients who lost hearing was 38 months. • Manchester: 1/66 lost hearing before a median of 5 years • Oxford: of all the patients who had useful hearing upon joining the service (from 2010 onwards), only one person has lost useful hearing since 2010. The median duration for the groups combined is 40 months. Percentage facial palsy rates of <20% as measured by House Brackmann scores of 4-6 18 months post-surgery for vestibular

	schwannoma: • Cambridge: 10% • Guys: there is a post-surgery facial palsy rate of 33% • Manchester: 38%Oxford: of all patients who started with good facial function pre-operatively for a vestibular schwannoma, 42% have had a drop in facial function and have a facial palsy 18 months post operatively
Geographical equity access	No evidence of geographical inequity

Innovation

Type 2 neurofibromatosis (NF2) is a genetic disorder characterised by the growth of tumours in nerve cells throughout the body, but particularly in the auditory (hearing) nerves. Surgery to remove the tumour may be necessary but a key aim is to avoid unnecessary surgical treatment. Removal of a tumour on the auditory nerve results in deafness and, in NF2 tumours, are often bilateral. The NF2 service has pioneered the use of drug treatment to slow the growth of tumours; where bilateral removal of tumours has resulted in complete hearing loss, some restoration of hearing is possible by inserting a device directly into the brainstem.

Highly specialised services offer the opportunity to develop a much greater understanding of the disease and the service has been at the forefront of research into genetics and other aspects of NF2.

An innovation in genetic testing (for the LZTR1 gene) has led to the correct reclassification of seven patients who, although would meet the criteria for NF2, would not have been previously identified.

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 and longitudinally extensive myelitis 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. The service provides an accurate diagnosis, inpatient or outpatient assessment and review. About 1,000 people in England are living with NMO.	
NHS Centres	Oxford University Hospitals NHS Trust; The Walton Centre NHS Foundation Trust
Expenditure	More than £1m but less than £5m
First Evaluations	189
Outcomes collated	Annualised relapse rate: pre (before treatment by the service) 0.37; post 0.22 (after treatment by the service)
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 eye cancer treatments can be used individually or in a combination and at present it is not clear if any particular approach has any advantages. The treatment aims, whenever possible, to preserve vision in the affected eye. Follow up care is provided for patients whose tumours recur or who have complications receive further treatment.

NHS Centres	Moorfields Eye Hospital NHS Foundation Trust; The Royal Liverpool and Broadgreen University Hospitals NHS Trust; Sheffield Teaching Hospitals NHS Foundation Trust
Expenditure	More than £5m but less than £10m
Positive assessment	650
Outcomes collated	Percentage primary enucleation amongst patients with melanoma: • Liverpool: 23% • Moorfields: 31.6% • Sheffield: 29% Percentage secondary enucleation amongst patients with melanoma: • Liverpool: 0% • Moorfields 0% • Sheffield: 1% Percentage developing metastatic disease amongst patients with melanoma: • Liverpool: 0% • Moorfields: 3.9% • Sheffield: data not available
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; University College London Hospital NHS Foundation Trust,
Expenditure	More than £1m but less than £5m
Total cases received	3,713
Outcomes collated	Percentage simple cases reported within 7 working days: • Manchester: 75% • Sheffield: 70% • Liverpool: 73% • UCLH: 95% Percentage complex cases reported within 10 working days: • Manchester: 93% • Sheffield: 93% • Liverpool: 82% • UCLH: 95% Percentage all cases reported within 21 working days: • Manchester: 100% • Sheffield: 99.79% • Liverpool: 97%. • UCLH: 99.9%
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.	
NHS Centre	Brighton and Sussex University Hospitals NHS Trust
Expenditure	Less than £0.5m
Stage 2 surgery	Data suppressed to maintain patient confidentiality
Outcomes collated	Percentage patients with visual acuity 6/12 or better at 12 months post operation: • 100%
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, multi-disciplinary 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	More than £1m but less than £5m
Number of new patient referrals	Data suppressed to maintain patient confidentiality
Outcomes collated	Percentage clinical phenotype defined within 2 weeks (should increase or stay the same): • 43% Percentage 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
Innovation A key priority for the service is to distinguish villous disease (disease of the finger-like tentacles that line the wall of the intestine) from disease of the nerve or muscle; it is also important to know whether or not the whole of the small bowel is affected. If only a short section is affected, it can be cut out and the remaining bowel joined together again to restore continuity. If the whole bowel is affected, small bowel transplant may be the only treatment option. GOSH probably has now more experience with this group of conditions than any other centre in the world. This concentration of expertise has allowed the service to make substantial contributions to the understanding of the disorders, particularly their genetic basis.	

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	More than £5 million but less than £10m
Number of transplants	148
Outcomes collated	One-year risk-adjusted patient survival from first transplant from deceased donors: • Cambridge: 99%; • Guy's: 96% • Imperial: 100% • Manchester: 97% • Newcastle: 100% • Oxford: 98% Five-year risk-adjusted patient survival from first transplant from deceased donors: • Cambridge: 91% • Guy's: 88% • Imperial: 73% • Manchester: 89% • Newcastle: 81% • Oxford: 87% Five-year risk-adjusted patient survival from listing from first deceased donor transplant: • Cambridge: 89% • Guy's: 87% • Imperial: 87% • Manchester: 85% • Newcastle: 84% • Oxford: 83%
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	King's College Hospital NHS Foundation Trust; Leeds Teaching Hospitals NHS Trust
Expenditure	More than £50 million
Caseload	239 receiving eculizumab for PNH 486 patients are not receiving eculizumab)
Outcomes collated	Five-year relative survival rate; • Leeds: 87.6% • King's: data not available Median transfusions per patient in previous 12 months: • Leeds: 2 • King's 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 multi-disciplinary outpatient-based diagnostic, advice and management service (including respirology, 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	More than £1m but less than £5m (for management and diagnostic elements)
Caseload	583
Outcomes collated	Percentage of patients in the PCD management service offered an annual review appointment. The annual review will consist of the processes listed in the service specification: • Brompton & Harefield: 99%; • Leicester and Birmingham: 100% • Leeds: 100% Percentage of patients seen by a physiotherapist at annual review: • Brompton & Harefield: 99% • Leicester and Birmingham: 100% • Leeds: 100%
Geographical equity access	Data not available or not comparable
Innovation The cilia are structures on the cell surface which play an important role in the lungs, ears, sperm and elsewhere. Electron microscopy reveals their beauty and complexity. This complex structure allows the cilia to wag like a dog's tail: this action provides motility to sperm, resonance to particular frequencies of sound in the inner ear, and clearance of mucous substances from the inner surface of the lung.	

The cilia may be malformed due to genetic defects, or more commonly they may be damaged by noxious substances, especially tobacco smoke in the lungs. Damage to the cilia leads to: frequent infections of the lung which take a long time to clear up; recurrent ear problems; and male infertility.

The basic test for abnormal motion of the cilia (dyskinesia) is simple but distinguishing genetic (primary) ciliary dyskinesia from dyskinesia secondary to other causes requires expert laboratory investigation, including genetic tests, electron microscopy and high resolution video of living cilia.

Another function of cilia is to give the body its sense of left and right during embryonic development, ensuring that the heart is on the left, the liver on the right and so on. If there is a ciliary abnormality, these organs develop laterality at random, so that 50% of patients with PCD have their heart on the right, liver on the left and so on. This *situs inversus* is of course a strong clue to diagnosis.

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	More than £5 million but less than £10m
Number of confirmed cases	312
Outcomes collated	Percentage 3-year local recurrence among patients having limb salvage: • Oxford: 12% • Robert Jones: 2% • Royal Orthopaedic: 12.8% • Royal National Orthopaedic: 9% • Newcastle: 8.57% Percentage limb salvage: • Oxford: 98% • Robert Jones: 91%; • Royal Orthopaedic: 80%; • Royal National Orthopaedic: 80%. • Newcastle: data not available Percentage 3-year prosthesis infection/loosening: • Oxford: 21% • Robert Jones: 0% • Royal Orthopaedic: 2.4% • Royal National Orthopaedic: 4%. • Newcastle: 7.69%
Geographical equity access	Data not available or not comparable

Proton beam therapy overseas service (adults and children)	
Proton beam therapy provides radiation by delivering a beam of proton particles, rather than x-Rays. The physical properties of protons, results in almost no radiation dose being deposited in the normal tissue beyond the tumour. This is in contrast to x-rays where there is dose extension beyond the tumour. One centre in the UK provides low-energy proton beam therapy for patients who have eye tumours. In 2018, high energy proton beam therapy will be available in an NHS centre. Patients requiring high energy proton beam therapy can be referred overseas or to the NHS centre for treatment depending on need. A national Proton Therapy Clinical Reference Panel considers cases, decides whether they are eligible for proton beam therapy and can therefore be referred. Treatment takes 8-10 weeks.	
NHS Centres	Three overseas providers commissioned by NHS England
Expenditure	More than £20 million but less than £30m
Number of patients approved for referral (excludes patients from Wales)	216
Outcomes collated	The three-year actuarial overall survival and local control rates for children with tumours of the central nervous system were 96% and 91% respectively.
Geographical equity access	No evidence of geographical inequity

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 blood stream 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 Centres	Hampshire Hospitals NHS Foundation Trust; The Christie NHS Foundation Trust
Expenditure	More than £20 million but less than £30m
Major full cytoreduction	214
Outcomes collated	Five-year patient survival – all operative cases: • Christie: 72.28% • Hampshire: 67% Five-year patient survival – complete cytoreduction: • Christie: 85.34% • Hampshire: 79%
Geographical equity access	Data not available or not comparable

Innovation

A pilot between the Oxford small bowel transplant centre and the PMP centre in Basingstoke demonstrated that multivisceral small bowel transplantation is technically feasible for end stage PMP, the first in the world to demonstrate the possibility of this approach.

It is life transforming in survivors with excellent quality of life. Long term outcomes remain to be determined.

Pulmonary hypertension service for children	
Paediatric pulmonary hypertension 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 multi-disciplinary 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	More than £1m but less than £5m
Caseload	519
Outcomes collated	Percentage 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): • 1/24 Proportion/percentage of children receiving epoprostenol who required a line change due to infection: • 4.1% Proportion of children receiving epoprostenol who experienced a line related blood stream 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 pulmonary thrombo-embolic disease (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
Expenditure	More than £5m but less than £10m
Surgical operations	175
Outcomes collated	90-day patient survival: • 95% 3-year patient survival: • 89% In-hospital mortality: • 2.1%
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 NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Outpatient referrals	392
Outcomes collated	Percentage of patients with a genetic diagnosis: • 54 – 87% across the centres Percentage of patients given an alert card: • 100% across all centres
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 is a combination of surgery, chemotherapy or radiotherapy dependent on the needs of the individual child. 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
Expenditure	More than £1m but less than £5m
Confirmed patients	63
Outcomes collated	Percentage 5-year survival: • Barts: 98% • Birmingham Children's: 100% Percentage primary enucleation: • Barts: 36.9% • Birmingham Children's: 41%
Geographical equity access	Data required from cancer register
Innovation In Birmingham the team has been identifying the genetic basis of individuals with retinoblastoma as this has a profound effect on many aspects of care of the patient and their family. Free fetal DNA is also available in the maternal circulation during pregnancy. The Team have completed a pilot of six non-invasive prenatal tests on maternal samples and confirmed the correct result post-natally in all pregnancies. This has greatly increased family choice and has been very positively received in allowing the family to receive an early diagnostic answer to pre-delivery without an associated miscarriage risk. This has the potential to avoid the cost and inconvenience of bringing new-born babies and their families to a clinic potentially hundreds of miles away if cord bloods are not available and can be set against the additional cost of	

the non-invasive pre-natal test.

In the last year, the London centre has seen the development and delivery of new group interventions that RB patients and their families have accessed such as Sib Squad (a group for siblings of children with cancer) and Health Heroes (a group for children and young people living with chronic illness, and their carers).

The centre has finalised a booklet outlining the psychosocial support available to families via the RB service and has started a weekly 'drop-in' service where families can meet the team's psychologist during 'examination under anaesthetic' appointments. The percentage of children and young people whose psychosocial needs are being assessed has increased by 59% on last year, and by 29% for families and carers. An increase of 46.5% of children and young people are accessing psychosocial support compared to last year, and an increase of 21% of families and carers. 100% of families (N=10) contacted reported feeling supported during their care and we have had extremely positive qualitative feedback.

With regards to orthoptics, there has been more of a pastoral key-worker involvement in patient care. It has been ensured that close-links have been kept with visual impairment teams, teachers and other professionals to ensure that children's visual needs are correctly met. Visits have also been made to some individuals' schools in order to ensure that appropriate low vision aids and strategies have been used.

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 three centres also arrange for a stock of the drug, haem arginate, to be sent where appropriate • A structured multi-disciplinary 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 with acute porphyria who meet the definition of 'severe' disease.	
NHS Centres	King's College Hospital NHS Foundation Trust; University Hospital of Wales
Expenditure	More than £0.5m but less than £1m
Active caseload	155
Outcomes collated	Percentage mortality rate: • King's: 0% • Wales: 2.4% Percentage of patients having four or more hospital admissions (porphyria-related) in the previous 12 months: • King's: 3.6% • Wales: 4.8%
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	More than £10 million but less than £20m
Number of transplants	48
Outcomes collated	Percentage 24-month patient survival post HSCT (or gene therapy): • GOSH: 80% • Newcastle: 89%
Geographical equity access	No evidence of geographical inequity

Innovation

Babies born with severe combined immune deficiency (SCID) lack both components of the body's defence against infection – B cells and T cells. If untreated, early death results from multiple infections with bacteria, viruses and other organisms. But in the highly specialised service, based at Newcastle and Great Ormond Street Hospital, treatment is able to achieve a complete cure in 90% of patients. This astonishing achievement is the result of gradual refinement over many decades of the treatment regimes. A key focus in the modern era is to reduce the toxicity of the regimes.

Although initially SCID was considered to be a single clinical entity, genetic and biochemical analysis has shown that it is the end result of many different genetic faults, each with their own characteristics.

The mainstay of treatment is haemopoietic stem cell transplant (HSCT), but the service has also developed (for appropriate patients) gene therapies and thymus (a small organ that forms part of the immune system) transplant.

Late diagnosis affects prognosis – an infant who already has multiple infections

before the diagnosis is made is likely to have a difficult treatment path. Public Health England is working up a pilot scheme for testing all babies at birth. This requires very careful planning as the aim is to detect 10-15 affected babies among the 600,000 born in England each year.

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	More than £1m but less than £5m
Number of transplants	Data suppressed to maintain patient confidentiality
Outcomes collated	Percentage unadjusted 90-day patient survival for elective first intestine transplants between 01/04/2007 to 31/03/2017: • Cambridge: 91.3% • Oxford: 86.8% Percentage unadjusted one-year patient survival from elective first intestine transplants between 01/04/2007 to 31/03/2017: • Cambridge: 79.9% • Oxford: 78.2% Percentage unadjusted five-year patient survival from elective first intestine transplants between 01/04/2007 to 31/03/2017: • Cambridge: 49.1% • Oxford: 61.9%
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; King's College Hospital NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Number of transplants	Data suppressed to maintain patient confidentiality
Outcomes collated	Percentage unadjusted 90-day patient survival for elective first intestine transplants between 01/04/2007 to 31/03/2017: • Birmingham:92.7% • King's 100% Percentage unadjusted one-year patient survival from elective first intestine transplants between 01/04/2007 to 31/03/2017: • Birmingham: 82.9% • King's: 93.8% Percentage unadjusted five-year patient survival from elective first intestine transplants between 01/04/2007 to 31/03/2017: • Birmingham: 53.4% • King's: 72.5%
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; King's College Hospital NHS Foundation Trust; Leeds Teaching Hospitals NHS Trust
Expenditure	More than £10m but less than £20m
Inpatient episodes	1,085
Outcomes collated	Data suppressed to maintain patient confidentiality on the basis that the service covers a large number of diagnoses but there are only a small number of patients who have each of the conditions
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	More than £0.5m but less than £1m
Index patients	81
Outcomes collated	Percentage of patients with a definitive diagnosis or diagnosis ruled out: • 47% definitive diagnosis Percentage of patients with a genetic diagnosis: • 32%
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 occur 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; NHS Greater Glasgow & Clyde (until July 2016)
Expenditure	Less than £0.5m
Number of procedures	Data suppressed to maintain patient confidentiality
Outcomes collated	Data suppressed to maintain patient confidentiality
Geographical equity access	Numbers too small to analyse

Ventricular assist devices (VADs) 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. In 2017/18 there were 103 long-term VADs in adults and children.	
NHS Centres	Great Ormond Street Hospital for Children NHS Foundation Trust; Manchester University NHS Foundation Trust: 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	187
Outcomes collated	27% of patients given a long term VAD as bridge to transplant had been transplanted within 3 years
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 multi-disciplinary structure. Patients are assessed and reviewed by all the specialities appropriate to their needs during the clinic. The service cares for about 70 people with Wolfram syndrome. Following this review, a management plan for local care providers is agreed and communicated to allow the local health care professionals to implement the recommendations and monitor their progress.	
NHS Centres	Birmingham Children's Hospital NHS Foundation Trust; University Hospitals Birmingham NHS Foundation Trust
Expenditure	Less than £0.5m
Caseload	79
Outcomes collated	Percentage of children with HbA1C in target range: • Birmingham joint reporting: 75% Percentage of adults with Hba1C <75 mmol/mol: • Birmingham joint reporting: 66.7% Percentage of adults with a BMI <35: • Birmingham: 94.1%
Geographical equity access	Numbers too small to analyse

Listening to and acting on feedback

In the Wolfram service, there was feedback that 'the clinic was very busy with all clinicians from the multi-disciplinary team booked to see the patients during the full day clinic appointment. As a result of the feedback, the Clinic Administrator changed the way the clinic works and now coordinates the timetable for each patient to ensure that they are at their appointments at the scheduled times and holds the master copy of the timetable which all staff adhere to and update. –
Other feedback was that families felt that they 'didn't know what appointment was next' so they are now talked through their timetable with different clinicians when they arrive at the hospital on the Monday morning.

Families also reported that they 'waited too long in the eye department' so this has been improved.

Finally, families reported that 'getting to the hospital for 8am for the blood test was stressful', so, at the last clinic, families were invited to arrive later instead (between 8.30-9am).

Xeroderma pigmentosum service (adults and children)	
Xeroderma pigmentosum is a life-threatening inherited disorder affecting 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 one-stop 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	More than £0.5m but less than £1m
Caseload	119
Outcomes collated	Percentage of adults with window film at home: • 60% Percentage of children with window film at home: • 97% Percentage of children wearing sunglasses when outside (who do not wear a visor): • 78% Percentage of adults wearing sunglasses when outside (who do not wear a visor): • 88%
Geographical equity access	No evidence of geographical inequity

Appendix A: European Reference Networks

1. 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. ERN 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.

The European Union have approved a number of UK providers to become part of the networks. The UK are members of 23 out of the 24 ERN networks and are lead coordinating centres for six of the ERNs. The detail is listed in list in Annex 1 provides the detail of all the UK endorsed ERN centres and the UK coordinating centres:

2. Annex 1: UK endorsed ERN centres and the UK co-ordinating centres

Official name of ERN	ERN code	NHS Trust	Approved as the Coordinat-ing EU centre
European Reference Network on Rare Bone Diseases	BOND	Alder Hey Children's NHS Foundation Trust	
European Reference Network on Rare Bone Diseases	BOND	Birmingham Children's Hospital NHS Foundation Trust	
European Reference Network on Rare Bone Diseases	BOND	Bristol University Hospital 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	Newcastle upon Tyne Hospitals NHS Foundation Trust	
European Reference Network on Rare Bone Diseases	BOND	NHS Greater Glasgow and Clyde	
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	
European Reference Network on Rare craniofacial anomalies and ENT disorders	CRANIO	Birmingham 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	

Official name of ERN	ERN code	NHS Trust	Approved as the Coordinat -ing EU centre
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	Bristol University Hospital 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 UCLA	
European Reference Network on Rare Endocrine Conditions	ENDO	NHS Greater Glasgow and Clyde	
European Reference Network on Rare Endocrine Conditions	ENDO	University College London Hospitals NHS Foundation Trust - joint application with GOSH	
European Reference Network on Rare Endocrine Conditions	ENDO	University Hospitals Birmingham NHS FT	
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	yes
European Reference Network on Rare and Complex Epilepsies	EpiCARE	NHS Greater Glasgow and Clyde	
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 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	Newcastle upon Tyne Hospitals NHS Foundation Trust	
European Rare Kidney Diseases Reference Network	ERKNet	Royal Free London, NHS Foundation Trust	

Official name of ERN	ERN code	NHS Trust	Approved as the Coordinat-ing EU centre
European Reference Network on Rare Hepatological Diseases	ERN Liver	Birmingham Children's Hospital NHS Foundation Trust	
European Reference Network on Rare Hepatological Diseases	ERN Liver	Newcastle upon Tyne Hospitals NHS Foundation Trust	yes
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 FT	
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 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	
European Reference Network on Rare Respiratory Diseases	ERN-LUNG	Imperial College Healthcare NHS Trust	
European Reference Network on Rare Respiratory Diseases	ERN-LUNG	Imperial College Healthcare NHS Trust	
European Reference Network on Rare Respiratory Diseases	ERN-LUNG	Newcastle upon Tyne Hospitals NHS Foundation Trust	
European Reference Network on Rare Respiratory Diseases	ERN-LUNG	Ninewells Hospital and Medical School, Dundee	
European Reference Network on Rare Respiratory Diseases	ERN-LUNG	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	
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 Children's Hospital NHS Foundation Trust	

Official name of ERN	ERN code	NHS Trust	Approved as the Coordinat-ing EU centre
European Reference Network on Rare and Undiagnosed Skin Disorders	ERN-SKIN	Cardiff and Vale University Health Board	
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	
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 & 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	Newcastle upon Tyne Hospitals NHS Foundation Trust	yes
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	Approved as the Coordinat-ing EU centre
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	yes
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	
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 Children's Hospital NHS Foundation Trust	
European Reference Network on Rare Congenital Malformations and Rare Intellectual Disability	ITHACA	Manchester University NHS Foundation Trust	yes
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 Children's Hospital NHS Foundation Trust	
European Reference Network for Rare Hereditary Metabolic Disorders	MetabERN	Bristol Royal Hospital for Children	

Official name of ERN	ERN code	NHS Trust	Approved as the Coordinat-ing EU centre
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 FT	
European Reference Network for Paediatric Cancer (haemato-oncology)	PaedCan	Birmingham Children's Hospital NHS Foundation Trust	
European Reference Network for Paediatric Cancer (haemato-oncology)	PaedCan	Great Ormond Street Hospital for Children NHS Foundation Trust	
European Reference Network for Paediatric Cancer (haemato-oncology)	PaedCan	Royal Manchester Children's Hospital	
Rare Immunodeficiency, Autoinflammatory and Autoimmune Diseases Network	RITA	Aintree University Hospitals 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	Newcastle upon Tyne Hospitals NHS Foundation Trust	yes
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 Hospital NHS Trust	
European Reference Network on Transplantation in Children (incl. 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	
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 of Highly Specialised Services during 2017/18

Name of Service	NHS England commissioning arrangements on behalf of devolved administrations – See Key
Alkaptonuria service (adults)	2
Alström syndrome service (adults and children)	2
Ataxia telangiectasia services for adults	2
Ataxia telangiectasia services for children	2
Atypical haemolytic uraemic syndrome (adults and children)	6
Autologous intestinal reconstruction service for adults	6
Bardet Biedl syndrome service (adults and children)	2
Barth syndrome service (male adults and children)	2
Beckwith-Wiedemann syndrome with macroglossia service (children)	2
Behçet's syndrome service (adults and adolescents)	6
Bladder exstrophy service (children)	2
Breast radiotherapy injury rehabilitation service (a discrete cohort of adult females)	6
Cardiothoracic Transplantation service (Paediatrics)	12
Choriocarcinoma service (adults and adolescents)	1
Chronic pulmonary aspergillosis service (adults)	2
Complex childhood osteogenesis imperfecta service	6
Complex Ehlers Danlos syndrome service (adults and children)	2
Complex neurofibromatosis type I service (adults and children)	6
Complex tracheal disease service (children)	2

Name of Service	NHS England commissioning arrangements on behalf of devolved administrations – See Key
Congenital hyperinsulinism service (children)	2
Craniofacial service (adults and children)	1
Cryopyrin associated periodic syndrome service (adults)	2
Diagnostic service for amyloidosis (adults)	2
Diagnostic service for primary ciliary dyskinesia (adults and children)	2
Diagnostic service for rare neuromuscular disorders (adults and children)	2
Encapsulating peritoneal sclerosis treatment service (adults)	6
Epidermolysis bullosa service (adults and children)	2
Extra corporeal membrane oxygenation service for adults	2
Extra corporeal membrane oxygenation service for neonates, infants and children with respiratory failure	2
Ex-vivo partial nephrectomy service (adults)	6
Gender identity development service for children and adolescents	2
Heart Transplantation service (adults)	12
Insulin Resistant Diabetes (Adults and Children)	6
Islet transplantation service (adults)	6
Liver transplantation service - ADULTS	8
Liver transplantation service CHILDREN	1
Lung Transplantation service (Adults)	12
Lymphangioleiomyomatosis	2
Lysosomal storage disorders service (Children & Adults)	9
McArdle disease service (children)	2

Name of Service	NHS England commissioning arrangements on behalf of devolved administrations – See Key
Neurofibromatosis type 2 service (All Ages)	2
Neuromyelitis optica service (adults and children)	2
Ocular oncology service (adults)	10
Ophthalmic pathology service (adults and children)	10
Osteo-odonto-keratoprosthesis service for corneal blindness (adults)	6
Paediatric intestinal pseudo-obstructive disorders service	2
Pancreas transplantation service (adults)	10
Paroxysmal nocturnal haemoglobinuria	11
Primary ciliary dyskinesia management service (children)	2
Primary malignant bone tumours service (adults and adolescents)	6
Proton beam therapy overseas service (adults and children)	13
Pseudomyxoma peritonei service (adults)	2
Pulmonary hypertension service for children	11
Pulmonary thromboendarterectomy service (adults and adolescents)	2
Rare mitochondrial disorders service (adults and children)	2
Reconstructive surgery service for adolescents with congenital malformation of the female genital tract	6
Retinoblastoma service (children)	1
Severe acute porphyria	2
Severe combined immune deficiency and related disorders service (children)	3
Small bowel transplantation service (adults)	6
Small bowel transplantation service (children)	2

Name of Service	NHS England commissioning arrangements on behalf of devolved administrations – See Key
Specialist paediatric liver disease service	**1**
Stickler syndrome diagnostic service (adults and children)	**2**
VADs as bridge to transplant	**3**
Vein of Galen malformation service (adults and children)	**2**
Wolfram syndrome service (adults and children)	**2**
Xeroderma pigmentosum service (adults and children)	**2**

Key:
1-Fully commissioned on behalf of UK (Pre-1991)
2-Fully commissioned on behalf of England & Scotland
3-Fully commissioned on behalf of England & in-part for Scotland
4-Fully commissioned on behalf of England & in-part for NI
5-Fully commissioned on behalf of England, in-part for Scotland & in-part for NI
6-Fully commissioned on behalf of England only
7-Commissioned on behalf of England service only not drugs
8-Fully commissioned on behalf of England, NI & Wales and by exception for Scotland
9-Fully commissioned on behalf of England, in-part for Scotland (service only not drugs) & in-part for NI (not ERT drugs)
10-Fully commissioned on behalf of England, from devolved administrations for Scotland
11-Fully commissioned on behalf of England and in part for Scotland (service only not drugs)
12-Fully commissioned on behalf of England, in-part for Scotland by arrangement, in full for NI
13-Fully commissioned on behalf of England, Scotland and NI