Classification: Official
Publication approval reference: PAR1769

Highly specialised services 2019/20

Highly Specialised Services 2019/20

Version number: 1

Prepared by: Highly Specialised Commissioning Team (HSCT)

Classification: OFFICIAL

Publishing Approval Reference Number: PAR1769

The information in this document can be made available in alternative formats, such as easy read or large print, and may be available in alternative languages, on request.

Contents

1. Introduction

Purpose of this document

The primary purpose of this document is to provide key information about highly specialised services during 2019/20. In summary, the information comprises:

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

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

Equality statement

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

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

services are provided in an integrated way where this might reduce health inequalities.

Specialised services

NHS England is responsible for commissioning specialised services to meet a wide range of health and care needs. In 2019/20 NHS England's total spend on all specialised services was £18.5 billion.

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

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

Highly specialised services

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

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

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

Planning highly specialised services on a national, rather than local, basis, provides a challenge for the Highly Specialised Commissioning Team (HSCT) to ensure

equitable access to services, given the small number of expert centers and the fact that some patients may live a long way away from centres. It is also important to have a robust process for selecting and monitoring the centres which provide these services, given the very high level of expertise required.

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

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

Rare Diseases Advisory Group

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

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

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

RDAG makes recommendations to NHS England and the devolved administrations on developing and implementing strategy for highly specialised services including

making recommendations on how the UK Strategy for Rare Diseases should be implemented.

Expenditure figures

The expenditure figures for each service reflect NHS England's expenditure on that service in 2019/20. Expenditure is set out into the following categories:

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

Clinical outcomes for highly specialised services

Monitoring of clinical outcomes is a key responsibility of the HSCT. Within highly specialised services there is a high level of clinical outcome monitoring in place. The HSCT work closely with the services and the NHSE Quality colleagues to ensure high levels of data completeness.

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

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

Geographical equity of access to highly specialised services

The central ethos of commissioning highly specialised services is to concentrate expertise in a small number of expert centres. The trade-off implied by this ethos is that access may be difficult for patients who need to travel long distances to access care from the expert centres. Hence it is incumbent on the HSCT to monitor the geographical access to highly specialised services.

The best way to measure the distribution of patients accessing a service across England is by using a metric known as the systematic component of variation (SCV). This compares how many patients per region are accessing a service against the number that would be expected, based on regional population data. The higher the SCV, the greater the discrepancy between the number of patients accessing the service and the number which would be expected for each region.

Patients are mapped to a region according to the postcode of the GP surgery with whom they are registered.

It is unlikely that the observed and expected figures will match completely as some variation will occur by chance. Where the SCV is below 0.2, variation can be considered random and further investigation is not required. An SCV above 0.2 (or 20%) indicates variation greater than expected by chance and requires further review.

Where variation is observed which is likely not due to chance (SCV greater than 0.2), 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 seems to be genuinely fewer patients in the North East of England.

For those services where the SCV is above 0.2, the HSCT is reviewing 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 variation at an appropriate time, The HSCT routinely undertakes an analysis of geographical variation for each service every three years.

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).

In some services the number of patients being treated is too small to allow for meaningful analysis or else the data is not available or comparable.

2. New highly specialised services commissioned during 2019/20

DNA nucleotide excision repair disorders service

In April 2019, the long established highly specialised service for Xeroderma pigmentosum (XP) incorporated patients with Cockayne syndrome (CS) and trichothiodystrophy (TTD) to form the new DNA nucleotide excision repair disorders service provided in the Rare Disease Centre at Guy's and St Thomas' NHS Foundation Trust.

These are rare inherited multi-organ disorders and patients have specific, complex and specialist needs. Although the underlying diseases are not curable at present, there is potential to significantly improve health and quality of life through a comprehensive, expert patient focussed service.

This is provided by a rare disease centre, with a multidisciplinary clinical and molecular diagnostic service to co-ordinate the care and management of children and adults, and those young people transitioning between paediatric and adult services.

Primary ciliary dyskinesia (PCD) management service for adults

In November 2019, a new highly specialised service for the management of adults with PCD was commissioned. Primary ciliary dyskinesia (PCD) is a genetic condition in which the microscopic cells in the respiratory system called cilia do not function normally. Ciliary dysfunction prevents the clearance of mucus from the lungs, paranasal sinuses and ears. Recurring respiratory infections can lead to an irreversible scarring and obstruction in the bronchi (bronchiectasis) and severe lung damage. Cilia are also present in the ventricles of the brain and in the reproductive system so ciliary dysfunction can also affect other parts of the body.

Primary ciliary dyskinesia management services are provided by expert centres and include outreach services delivered as part of a provider network. Patients will transition into the service from the paediatric HSS PCD service.

Open fetal surgery to treat fetuses with open spina bifida

Spina bifida is an in-utero condition where the spinal column and cord are not fully formed. Babies born with this condition are often unable to walk, incontinent of urine and faeces, may develop hydrocephalus due to incomplete closure of the spinal canal and often require postnatal neurosurgical interventions.

For a carefully selected group of women and their babies open fetal surgery (operating on the baby while it is still in the womb) can be used to successfully close the spinal defect and achieve good clinical outcomes for the baby. In September 2019 a new highly specialised service was established to perform open fetal surgery to treat foetuses with open spina bifida.

The service is provided by two fetal surgery centres that provide assessment, open fetal surgery and supporting medical services. The service is delivered by an expert MDT in a shared care pathway with existing local maternity units / Regional Fetal Medicine Units and regional neurosurgery centres.

Total pancreatectomy with islet autotransplant

Chronic pancreatitis is chronic inflammation of the pancreas characterised by an irreversible, permanent and progressive destruction of pancreatic tissue. It may be hereditary or acquired. It is a disabling condition with symptoms including severe, persistent, intractable abdominal pain and diabetes.

In April 2019, a new highly specialised service started which treats patients with chronic pancreatitis by undertaking a total pancreatectomy with islet autotransplantation. Total pancreatectomy with islet auto transplant surgery involves removal of the pancreas followed by retransplantation of the patient’s own islet cells, which are isolated from the pancreas and infused into their liver.

3. Services and providers of highly specialised services for 2019/20

Alkaptonuria service (adults)

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

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

NHS centre	The Royal Liverpool and Broadgreen University Hospitals NHS Trust [now Liverpool University Hospitals NHS Foundation Trust]
Expenditure	Between £0.5 million and £1 million
Caseload	61
Outcomes collated	• Median quality of life (SF36) score for patients treated in the service for 12 months or longer: 36% SF36 stable • Median AKU severity score index measurement (AKUSSI) for patients treated in the service for 12 months or longer: 16% AKUSSI improved, ochronosis scores reversed Note: ochronosis is a condition in which the body cannot break down the toxic acid homogentisic acid. This causes bones and cartilage to become black and brittle. The spine collapses and prostate and kidney stones appear. Heart valves become blocked and the patient needs heart surgery.

Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions.

Alström syndrome service (adults and children)

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

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

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

NHS centres	Birmingham Children's Hospital NHS Foundation Trust [now Birmingham Women's and Children's Hospital NHS Foundation Trust] University Hospitals Birmingham NHS Foundation Trust
Expenditure	<£0.5 million
Caseload	89
Outcomes collated	• % of children with HbA1C less than 48 mmol/mol: - Birmingham Women and Children's Hospital: 74% • % of adults with HbA1c <75 mmol/mol: - University Hospitals Birmingham: 73% • Median age at death of patients on active caseload: - University Hospitals Birmingham: 25 • Proportion of patients with a BMI <35

	- Birmingham Women and Children's Hospital: 95% - University Hospitals Birmingham: 79% Note: glycated haemoglobin (HbA1c) is measured primarily to identify the 3-month average plasma glucose concentration.
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Ataxia telangiectasia services for adults

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

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

NHS centre	Papworth Hospital NHS Foundation Trust [now Royal Papworth Hospital NHS Foundation Trust]
Expenditure	<£0.5 million
Caseload	85
Outcomes collated	• Median BMI: 21.8 interquartile range 18.4-25.9 kg/m^2 (an important measure because patients with the condition often do not achieve optimum BMIs).
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Bardet-Biedl syndrome service (adults and children)

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

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

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

NHS centres	Birmingham Women's and Children's Hospital NHS Foundation Trust Great Ormond Street Hospital for Children NHS Foundation Trust Guy's and St Thomas' NHS Foundation Trust University Hospitals Birmingham NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Assessments	382
Outcomes collated	• % of children with HbA1c <48 mmol/mol: - Birmingham Women's and Children's Hospital: 94% - Great Ormond Street Hospital: 100% - Guy's and St Thomas': 100% • % of adult patients with HbA1c <75 mmol/mol: - Guy's and St Thomas': 95% - University Hospitals Birmingham: 96% • % of adult patients with a BMI <35: - Birmingham Women's and Children's Hospital: 80% - Guy's and St Thomas' 69%

	- University Hospitals Birmingham: 47% Note: Glycated haemoglobin (HbA1c) is a form of haemoglobin that is measured primarily to identify the 3-month average plasma glucose concentration.
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Ataxia telangiectasia services for children

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

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

NHS centre	Nottingham University Hospitals NHS Trust
Expenditure	<£0.5 million
Caseload	126
Outcomes collated	• % of patients with previously unrecognised treatable or untreatable morbidity: 89% • % of patients for whom active intervention was undertaken in clinic or arranged locally: 97% • Median QOL score at transition (using PedsQL Version 4): Data not submitted
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Atypical haemolytic uraemic syndrome (adults and children)

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

The aim of the service is to provide a national diagnostic and management advice for patients with aHUS. The service offers comprehensive diagnostic clinical and pathological investigations and expert opinion, facilitating optimal patient management on a shared-care basis with referring clinicians and other specialist services.

NHS centre	The Newcastle upon Tyne Hospitals NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Caseload	130
Outcomes collated	• Number of deaths in patients with a diagnosis of complement mediated aHUS: no patient in England died of aHUS in 2019/20
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

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

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

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

NHS centres	Manchester University NHS Foundation Trust Guy's and St Thomas' NHS Foundation Trust
Expenditure	<£0.5 million
Surgical operations	The number of surgical operations is fewer than five, so the data has been suppressed to maintain patient confidentiality
Outcomes collated	• Mean length of stay in hospital (days): - Guy's and St Thomas': 6 - Manchester University: 7 • % patients with improved soundfield hearing at 24 months - Manchester University: 50 - Guy's and St Thomas': data suppressed to maintain patient confidentiality • % patients with improved CAP score at 24 months - Manchester University: 50 - Guy's and St Thomas': data suppressed to maintain patient confidentiality
Geographical equity access	Numbers too small to analyse

Autologous intestinal reconstruction service for adults

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

NHS centre	Salford Royal NHS Foundation Trust

Expenditure	<£0.5 million
Caseload	No new cases in 2019/20
Outcomes collated	• Proportion of patients not needing parenteral nutrition at 24 months post operation: 0 • Proportion of patients alive 1-year post operation: 0 Note: The outcomes results are zero as there were no new cases in 2019/20
Geographical equity access	Numbers too small to analyse

Barth syndrome service (male adults and children)

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

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

NHS centre	University Hospitals Bristol NHS Foundation Trust [now University Hospitals Bristol and Weston NHS Foundation Trust]
Expenditure	>£0.5 million but <£1 million
Caseload	27

Outcomes collated	• Median age at diagnosis: data suppressed to maintain patient confidentiality • Median age at death: No deaths • % of hospital admissions for bacterial infections for patients on G-CSF: 5% • Number of hospital admissions for bacterial infections for patients on G-CSF: 20
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Beckwith-Wiedemann syndrome with macroglossia service (children)

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

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

NHS centre	Great Ormond Street Hospital for Children NHS Foundation Trust
Expenditure	<£0.5 million
Caseload	209
Outcomes collated	• % patients achieving improvement at the 3 - 6 months post-operative assessment of resting tongue position: 100%

	• % patients achieving improvement at the 3 - 6 month post-operative assessment in the reduction or cessation of drooling: 100% • % patients achieving improvement at the 3 - 6 month post-operative assessment in the reduction or elimination of Macroglossia related errors: 100% • % patients achieving improvements at the 3 - 6 month post-operative assessment in the reduction or elimination of oral stage difficulties related to the macroglossia: 100% • % patients achieving improvements at the 3 - 6 month post-operative assessment in the reduction or elimination of parental concerns related to the macroglossia: 100%
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Behçet’s syndrome service (adults and adolescents)

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

NHS centres	Barts Health NHS Trust Liverpool University Hospitals NHS Foundation Trust Sandwell and West Birmingham Hospitals NHS Trust
Expenditure	>£1 million but <£5 million
Caseload	1,880
Outcomes collated	• Median number of flares per patient during the previous 12 months:

	- Barts Health: 1 - Liverpool: 1 - Sandwell and West Birmingham: 0
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Bladder exstrophy service (children)

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

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

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

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

NHS centres	Great Ormond Street Hospital for Children NHS Foundation Trust Manchester University NHS Foundation Trust
Expenditure	>£1 million but <£5 million
New babies	14

Outcomes collated	• % closure achieved without dehiscence: - Great Ormond Street: 67%Manchester University: 100% • % patients' continent (dry by day) at age five years: - Great Ormond Street: 33% - Manchester University: 78% Note: For GOSH, this figure describes the percentage of children who turned five in the financial year 2019/20 and were continent (dry by day) with no augmentation/catheterisation to empty the bladder. The number of children in this group is extremely low which means small changes in the numerator and denominator can significantly impact on the overall percentage. It is expected that continence will increase with age. Note: For Manchester, this figure describes the experience of the entire patient cohort at age 5 years who were continent (dry by day). Some of these children may use augmentation/catheterisation to empty the bladder.
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

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

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

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

NHS centre	Royal National Hospital for Rheumatic Diseases – Royal United Hospitals Bath NHS Foundation Trust
Expenditure	<£0.5 million
Caseload	24

Outcomes collated	• % improvement in mood: 63% • % improvement in upper limb function: 83% Note: These are good scores for patients who have been treated for severe health problems.
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Cardiothoracic transplantation service (paediatric)

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

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

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

NHS centres	Great Ormond Street Hospital for Children NHS Foundation Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust
Expenditure	>£50 million (adults and children, heart and lung)
Number of transplants	23
Outcomes collated	• 30-day unadjusted patient survival rate after first paediatric heart only transplant: - Great Ormond Street: 98.3%

	- Newcastle upon Tyne: 93.7% • 1-year unadjusted patient survival rate after first paediatric heart only transplant: - Great Ormond Street: 98.3% - Newcastle upon Tyne: 88.9% • 5-year unadjusted patient survival after first paediatric heart only transplant: - Great Ormond Street: 82% - Newcastle upon Tyne: 79.3% • 90-day patient survival rate after first paediatric lung transplant: - Great Ormond Street: 87% - Newcastle upon Tyne: 87.5% • 1-year unadjusted patient survival rate after first paediatric lung only transplant - Great Ormond Street: 82.6% - Newcastle Upon Tyne: Data not submitted • 5-year unadjusted patient survival after first paediatric lung only transplant: - Great Ormond Street: 78.3% - Newcastle upon Tyne: Data not submitted
Geographical equity access	Data not available or not comparable

Choriocarcinoma service (adults and adolescents)

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

- Hydatidiform mole (also known as molar pregnancy): in this condition, the sperm and egg cells join together but a healthy fetus does not develop. The placenta grows to an abnormal size, requiring surgical evacuation of the uterus.

- Choriocarcinoma, which is an aggressive and malignant cancer that may spread from the uterus to other organs in the body, such as the lungs or brain. Each year about 10 women in England develop choriocarcinoma.
- Placental site trophoblastic tumour, a rare variant of choriocarcinoma. This cancer is able to spread through the body via the lymphatic system.

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

NHS centres	Imperial College Healthcare NHS Trust Sheffield Teaching Hospitals NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Inpatient episodes	614
Outcomes collated	• Deaths as % of new cases each year: - Imperial College: 0% - Sheffield Teaching: 0%
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Chronic pulmonary aspergillosis service (adults)

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

NHS centre	Manchester University NHS Foundation Trust
Expenditure	>£5 million but <£10 million
Caseload	495
Outcomes collated	• % of patients showing a ≥12-point improvement in the St George's Respiratory Questionnaire (SGRQ) and ≥3 kg weight gain: 43% Note: This is a substantial improvement in quality of life in those with a condition that does not improve at all without treatment.
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Complex childhood osteogenesis imperfecta service

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

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

NHS centres	Birmingham Women's and Children's Hospital NHS Foundation Trust Great Ormond Street Hospital for Children NHS Foundation Trust Sheffield Children's NHS Foundation Trust University Hospitals Bristol NHS Foundation Trust
Expenditure	>£1 million but <£5 million

Caseload	325
Outcomes collated	• Median number of new non-vertebral fractures: - Birmingham Women's and Children's: 21* - Great Ormond Street: 0 - Sheffield Children's: 0 - University Hospitals Bristol: 0 • Median number of new vertebral fractures: - Birmingham Women's and Children's: 27* - Great Ormond Street: 0 - Sheffield Children's: 0 - University Hospitals Bristol: 0 • % patients with scoliosis and Cobb angle >45 degrees (the Cobb angle measures the degree of abnormal lateral spinal curvature): - Birmingham and Women's Children's: 4% - Great Ormond Street: 5% - Sheffield Children's: 5% - University Hospitals Bristol: 0% *All outcome measures are discussed at the annual clinical meeting including reasons for apparent variation
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Complex Ehlers-Danlos syndrome service (adults and children)

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

The fully comprehensive service (under the auspices of the clinical genetics service) gives patients a precise clinical diagnosis and manages the subset in

whom clinical diagnosis is not straight forward or diagnosis through laboratory testing needs to be confirmed with further clinical evaluation.

NHS centres	London North West University Healthcare NHS Trust Sheffield Children's NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Definitive diagnosis	119
Outcomes collated	• % patients with a definitive diagnosis or diagnosis ruled out: - London North West: 44.2%* - Sheffield Teaching: 100% • % of patients with a genetic diagnosis: - London North West: 32.5% - Sheffield Teaching: 40.4% *All outcome measures are discussed at the annual clinical meeting including reasons for apparent variation
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Complex neurofibromatosis type I service (adults and children)

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

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

The service includes:

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

NHS centres	Guy's and St Thomas' NHS Foundation Trust Manchester University NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Outpatient attendances	966
Outcomes collated	• Total number of interventions facilitated: - Guy's and St Thomas': 121 - Manchester University: 193 • Total number of interventions avoided: - Guy's and St Thomas': 28 - Manchester University: 17 • Mean age at death in the previous 10 years of patients with NFI: - Guy's and St Thomas': 38 - Manchester University: 44
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Complex tracheal disease service (children)

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

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

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

NHS centre	Great Ormond Street Hospital for Children NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Inpatient episodes	20
Outcomes collated	• 1-year survival: 100%
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Congenital hyperinsulinism service (children)

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

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

NHS centres	Great Ormond Street Hospital for Children NHS Foundation Trust Manchester University NHS Foundation Trust and Alder Hey Children's NHS Foundation Trust, which together form 'NORCHI'
Expenditure	>£1 million but <£5 million
Caseload	1,173
Outcomes collated	• Incidence of deaths in patients with CHI as a consequence of CHI: - Great Ormond Street: data suppressed to maintain patient confidentiality - NORCHI: 0 • Unplanned admissions due to CHI and admitted under the paediatric endocrine team, day cases excluded: - Great Ormond Street: 17 - NORCHI: 43
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

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 Women's and Children's Hospital NHS Foundation Trust Great Ormond Street Hospital for Children NHS Foundation Trust Oxford University Hospitals NHS Trust
Expenditure	>£10 million but <£20 million
Inpatient episodes	248

Outcomes collated	• Proportion of patients with level 4 surgical complications: - Alder Hey Children's: 0% - Birmingham Women's and Children's: 0% - Great Ormond Street: 0% - Oxford University: 1% • Proportion of patients with level 5 surgical complications: - Alder Hey Children's: 0% - Birmingham Women's and Children's: 0% - Great Ormond Street: 0% - Oxford University: 0%
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Cryopyrin associated periodic syndrome service (adults)

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

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

NHS centre	Royal Free London NHS Foundation Trust
Expenditure	>£5 million but <£10 million
Patients on high cost drugs	146
Outcomes collated	• Median 20-point CAPS activity score (a low CAPS score indicates symptom control): 2/20
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Diagnostic service for amyloidosis (adults and children)

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

The National Amyloidosis Centre provides diagnostic imaging (SAP scintigraphy – a technique for identifying amyloid deposits – and specialist echocardiography), histology and DNA analysis, genetic counselling, monitoring of amyloid proteins in the blood, recommendations for treatment, and supports the evaluation of existing and new therapies.

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

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

NHS centre	Royal Free London NHS Foundation Trust
Expenditure	>£5 million but <£10 million
First evaluations	1,457
Outcomes collated	• % of patients with a definitive diagnosis or diagnosis ruled out: 99.7% • % of patients with a genetic diagnosis: 8.6% Note: Amyloidosis can be genetic or acquired
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Diagnostic service for primary ciliary dyskinesia (adults and children)

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

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

NHS centres	Royal Brompton & Harefield NHS Foundation Trust University Hospitals Southampton NHS Foundation Trust University Hospitals of Leicester NHS Trust
Expenditure	>£0.5 million but <£1 million (for management and diagnostic elements)
Number of positive samples	78
Outcomes collated	• Paediatric outcomes collected by the paediatric PCD management service see page number 66. • Adult outcomes will be collected by the adult PCD management service which as a newly commissioned service will report its first set of measures in 2022.
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Diagnostic service for rare neuromuscular disorders (adults and children)

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

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

This service provides a diagnostic, advisory and clinical service for patients with four groups of very rare inherited neuromuscular disorders. These conditions are all inherited, and the definitive diagnosis for a patient is made by identifying the primary gene defect. Each disease group involves multiple genes, and the decision as to which gene to search first for DNA mutations is arrived at by using a disease-specific battery of techniques. These may include detailed clinical assessments, specialist neurophysiological tests, and immunological analyses on tissue biopsies.

NHS centres	Great Ormond Street Hospital for Children NHS Foundation Trust University College London Hospitals NHS Foundation Trust Oxford University Hospitals NHS Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust
Expenditure	>£5 million but <£10 million
Number of referrals	1,322
Outcomes collated	• % of patients with a genetic diagnosis: - Congenital muscular dystrophies: Great Ormond Street Hospital: 29% - Congenital myasthenic syndromes: Oxford University: 92% - Limb girdle muscular dystrophies: Newcastle upon Tyne: 55% - Muscle channelopathies University College London: 58% Note: Centres diagnose and assess different conditions, so outcome measures not comparable
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

DNA nucleotide excision repair disorders

DNA nucleotide excision repair disorders include patients with Xeroderma Pigmentosa (XP), Cockayne Syndrome (CS) and Trichothiodystrophy (TTD. These are rare inherited multi-organ disorders and patients have specific, complex and specialist needs. Although the underlying diseases are not curable at present, there is potential to significantly improve health and quality of life through a comprehensive, expert patient focussed service.

This is provided by a rare disease centre, with a multidisciplinary clinical and molecular diagnostic service to co-ordinate the care and management of children and adults, and young people transitioning between paediatric and adult services.

NHS centres	**Guy's and St Thomas' NHS Foundation Trust**
Expenditure	More than £1m but less than £5m
Caseload	186
Outcomes collated	The outcome measures for this service will be reported in the 2020/21 report
Geographical equity access	Numbers insufficient for robust analysis due to being a newly commissioned service.

Encapsulating peritoneal sclerosis treatment service (adults)

The encapsulating peritoneal sclerosis surgical service (EPS SS) provides surgical treatment for encapsulating peritoneal sclerosis (EPS). EPS, also referred to as sclerosing peritonitis, is a complication arising from long term use of peritoneal dialysis. EPS is characterised by marked sclerotic thickening of the peritoneal membrane, leading to encapsulation of the gut and sub-acute or acute bowel obstruction.

As a chronic fibrosing process, it leads to abdominal pain, nausea, vomiting, weight loss, fever, malnutrition, anaemia, ascites and finally surgical peritonitis and mortality. EPS is a condition associated with significant morbidity and mortality and with poor outcomes if not recognised early and treated. With centralising treatment in specified national centres, experience has been consolidated leading to better patient outcomes, mirroring the best international experience.

NHS centres	Cambridge University Hospitals NHS Foundation Trust Manchester University NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Primary surgical procedures	12
Outcomes collated	• 1-year survival rate for patient's post-operation for all cases (renal and non-renal): - Cambridge University Hospitals NHS Foundation Trust: 50%Manchester University NHS Foundation Trust: 100% • Proportion of patients TPN free post-operation: - Cambridge University Hospitals NHS Foundation Trust: 75% - Manchester University NHS Foundation Trust: 100%
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

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 Women's and Children's Hospital NHS Foundation Trust Great Ormond Street Hospital for Children NHS Foundation Trust Guy's and St Thomas' NHS Foundation Trust University Hospitals Birmingham NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Caseload	500 severe patients 1154 mild patients
Outcomes collated	• Median quality of life score at transition (QoLEB): - Birmingham Women's and Children's: 12.5 - Guy's and St Thomas': 17 - University Hospitals Birmingham NHS Foundation Trust: 16 • Proportion of unplanned admissions among patients with recessive dystrophic EB: - Birmingham Women's and Children's and Heart of England combined: 48% - Great Ormond Street: 47% - Guy's and St Thomas': 13% - University Hospitals Birmingham NHS Foundation Trust: 0 Note: Recessive dystrophic EB is the most severe type of EB.
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Extracorporeal membrane oxygenation service for adults

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

The specialist centres function as a national network, working closely with their local critical care networks. All centres provide a retrieval service that includes the

capability to undertake ‘mobile’ ECMO when this is deemed clinically necessary. The service assesses about 1,000 patients for treatment each year and about 300 of these are treated with ECMO.

NHS centres	Guy's and St Thomas' NHS Foundation Trust Manchester University NHS Foundation Trust Royal Brompton & Harefield NHS Foundation Trust Royal Papworth Hospital NHS Foundation Trust University Hospitals of Leicester NHS Trust
Expenditure	>£20 million but <£30 million (adults and children)
Starting treatment	292
Outcomes collated	• % survival at discharge: - Guy’s and St Thomas’: 82% - Manchester University: 65% - Royal Brompton & Harefield: 80% - Royal Papworth: 69%* - Leicester: 81% *All outcome measures are discussed at the annual clinical meeting including reasons for apparent variation These survival figures are not adjusted for case mix
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

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

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

NHS centres	Alder Hey Children's NHS Foundation Trust

	Birmingham Women's and Children's Hospital NHS Foundation Trust Great Ormond Street Hospital for Children NHS Foundation Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust University Hospitals of Leicester NHS Trust
Expenditure	>£10 million but <£20 million
Starting treatment	50
Outcomes collated	• % survival at discharge (neonatal): - Alder Hey Children's NHS Foundation Trust: 83% - Birmingham Women's and Children's: 60% Great Ormond Street: 67% - The Newcastle Upon Tyne: 50% - Leicester: 89% • % survival at discharge (children): - Alder Hey Children's: 20% - Birmingham Women's and Children's: 67% - Great Ormond Street: 80% - The Newcastle Upon Tyne: 57% - Leicester: 100%
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Ex-vivo partial nephrectomy service (adults)

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

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

NHS centre	Oxford University Hospitals NHS Trust
Expenditure	<£0.5 million
Patients accepted into service	11
Outcomes collated	• % 1-year survival post operation: 100% • Proportion of patient's dialysis free 1 year post-operation: 88%
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 service that provides support and therapeutic input for children and adolescents who have social and psychological difficulties with the development of their gender identity. Depending on need and subject to meeting strict criteria, the service may refer some children and young people to paediatric endocrinology clinics who may prescribe and administer hormone therapy.

NHS centres	The Tavistock and Portman NHS Foundation Trust Leeds Teaching Hospitals NHS Trust (satellite provider)
Expenditure	>£5 million but <£10 million
Number of referrals	2,586
Outcomes collated	Outcome measures to be revised

Geographical equity access	The geographical spread of patients was not assessed for this service

Hand and upper limb transplantation service (adults)

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

This service provides assessment, transplantation and follow-up.

NHS centres	Leeds Teaching Hospitals NHS Trust
Expenditure	<£0.5 million
Number of referrals	The number of transplants is less than five, so the data has been suppressed to maintain patient confidentiality
Outcomes collated	New outcome measures agreed which will be reported in 2020/21
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 Royal Brompton & Harefield NHS Foundation Trust Royal Papworth Hospital NHS Foundation Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust University Hospitals Birmingham NHS Foundation Trust

	Sheffield Teaching Hospitals NHS Foundation Trust (follow-up only)
Expenditure	>£50 million (adult and children, heart and lung)
Number of transplants	135
Outcomes collated	• 30-day risk-adjusted patient survival rates after first adult DBD (donor after brain death) heart transplant: - Manchester University: 93.7% - Royal Brompton & Harefield: 89.6% - Royal Papworth: 93.1% - University Hospitals Birmingham: 87.8% - The Newcastle Upon Tyne: 94.4% • 1-year risk-adjusted patient survival rates after first adult DBD heart transplant by centre: - Manchester University: 83% - Royal Brompton & Harefield: 81.1% - Royal Papworth: 86.7% - University Hospitals Birmingham: 77.1% - The Newcastle Upon Tyne: 86.7% • 5-year risk-adjusted patient survival rates form listing for first heart DBD transplants: - Manchester University: 65.3 - Royal Brompton & Harefield: 71.3% - Royal Papworth: 78.3% - University Hospitals Birmingham: 72.7% - The Newcastle Upon Tyne: 59.6%
Geographical equity access	Numbers too small to analyse

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

The High Consequence Infectious Disease Units (Airborne) provide safe and effective treatment of High Consequence Infectious Diseases (HCIDs) that are known or suspected to be transmissible from person to person via the airborne route.

NHS centres	Guy's and St Thomas' NHS Foundation Trust, adult and paediatric Liverpool University Hospitals NHS Foundation Trust, paediatric service provided by Alder Hey Children's Hospital NHS Foundation Trust Royal Free London NHS Foundation Trust, paediatric service provided by Imperial Hospitals NHS Foundation Trust Sheffield Teaching Hospitals NHS Foundation Trust, adult service only. This Trust was commissioned to provide this service in February 2020. The Newcastle upon Tyne Hospitals NHS Foundation Trust, adult and paediatric
Expenditure	<£1 million
Caseload	Not applicable
Outcomes collated	• Number of cases where HCID infection has spread from the specialist isolation unit: - Guy's and St Thomas': 0 - Liverpool University: 0 - Royal Free: 0 - Sheffield: 0 - The Newcastle Upon Tyne: 0 • Number of occasions where unit is unable to admit and start treatment of any patient with a confirmed diagnosis of airborne HCID within 6 hours (maximum) of notification: - Guy's and St Thomas': 0 - Liverpool University: 0

	- Royal Free London NHS Foundation Trust: 0 - Sheffield: 0 - The Newcastle Upon Tyne: 0
Geographical equity access	Numbers too small to analyse

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

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

NHS centres	Liverpool University Hospitals NHS Foundation Trust Royal Free London NHS Foundation Trust Sheffield Teaching Hospital NHS Foundation Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust
Expenditure	• >£1 Million but <£8 million
Number of admissions	0
Outcomes collated	• Outcomes are to be developed
Geographical equity access	Numbers too small to analyse

Insulin resistant diabetes (adults and children)

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

NHS centre	Cambridge University Hospitals NHS Foundation Trust
Expenditure	<£0.5 million
Active caseload	206
Outcomes collated	• % of patients with specific diagnosis: 86% • % of patients maintaining HbA1c below 75 mmol/mol: 63% Note: glycated haemoglobin (HbA1c) is measured primarily to identify the 3-month average plasma glucose concentration
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Islet transplantation service (adults)

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

NHS centres	King's College Hospital NHS Foundation Trust Manchester University NHS Foundation Trust North Bristol NHS Trust Oxford University Hospitals NHS Trust Royal Free London NHS Foundation Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust
Expenditure	<£1 million but <£5 million
Number of transplants	15
Outcomes collated	• Median number of severe hypoglycaemic events between registration and transplant: before transplant was 16.5 per year and at 1-year post transplant was no events per year. Of the patients for whom number of severe hypoglycaemic events at 1-year post-transplant was available, 84% experienced no events, 11% experienced one or two events and 3% experienced three or more events • For routine islet transplants between 1 April 2015 and 31 March 2019, median HbA1c dropped from 62 mmol/mol before transplant to 48 mmol/mol at 1-year post-transplant. Of those patients for whom HbA1c was reported at one year, 64% had an HbA1c of <53 mmol/mol Note: Glycated haemoglobin (HbA1c) is measured primarily to identify the 3-month average plasma glucose concentration. Note: NHS BT in conjunction with NHSE have a formal process for investigating transplant centres with significant variation in mortality rates
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Liver transplantation service, including live liver transplantation (adults)

This service provides assessment, transplantation and lifelong follow-up for patients requiring liver transplant surgery, including from living donors. The three main indications for liver transplantations are primary and secondary biliary cirrhosis, chronic hepatitis and fulminant hepatic failure.

NHS centres	Cambridge University Hospitals NHS Foundation Trust King's College Hospital NHS Foundation Trust Leeds Teaching Hospitals NHS Trust Royal Free London NHS Foundation Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust University Hospitals Birmingham NHS Foundation Trust
Expenditure	>£50 million (adults and children)
Number of transplants	777
Outcomes collated	• 1-year risk-adjusted patient survival for adult elective deceased donor first liver transplants 01/04/15 – 31/03/2019 - Cambridge University: 96.8% - King's College: 95.6%; - Leeds Teaching: 91.9% - Royal Free: 92.4% - Newcastle upon Tyne: 89.5% - Birmingham: 94.1% • 5-year risk-adjusted patient survival for adult elective deceased donor first liver transplants 01/04/11 – 31/03/2019: - Cambridge University: 87.6% - King's College: 84.4% - Leeds Teaching: 82% - Royal Free: 88.7% - Newcastle upon Tyne: 77.3%

	- Birmingham: 81% Note: The national survival rates after joining the transplant list for adult elective first liver only patients are 84.9% at 1 year, 72% at 5 years. Note: NHS BT in conjunction with NHSE have a formal process for investigating transplant centres with significant variation in mortality rates
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

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	>£50 million (adults and children)
Number of transplants	88
Outcomes collated	• 1-year unadjusted patient survival for paediatric elective deceased donor first liver transplants 01/04/15 – 31/03/19: - Birmingham Women's and Children: 91.9% - King's College: 97.2% - Leeds Teaching: 98.1% • 5-year unadjusted patient survival for paediatric elective deceased donor first liver transplants 01/04/11 – 31/03/15: - Birmingham Women's and Children: 93%

	- King's College: 92.1% - Leeds Teaching: 96.6% Note: NHS BT in conjunction with NHSE have a formal process for investigating transplant centres with significant variation in mortality rates
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

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 Royal Papworth Hospital NHS Foundation Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust University Hospitals Birmingham NHS Foundation Trust
Expenditure	>£50 million (adults and children, heart and lung)
Number of transplants	135
Outcomes collated	• 90-day risk-adjusted patient survival rate after first adult lung transplant: - Manchester University: 96.4% - Royal Brompton & Harefield: 90% - Royal Papworth: 91.6% - Newcastle upon Tyne: 89% - Birmingham: 87.2% • 1-year risk-adjusted patient survival rate after first adult lung transplant: - Manchester University: 87.4%

	- Royal Brompton & Harefield 84.2% - Royal Papworth: 80.4% - Newcastle upon Tyne: 83.2% - Birmingham: 74.3% • 5-year risk-adjusted patient survival rate from listing for first lung only transplants: - Manchester University: 51.6% - Royal Brompton & Harefield: 59.6% - Royal Papworth: 59.1% - Newcastle upon Tyne: 58.2% - Birmingham: 32% Note: NHS BT in conjunction with NHSE have a formal process for investigating transplant centres with significant variation in mortality rates
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions and have been calculated for the combined adult and paediatric patient groups

Lymphangioleiomyomatosis

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

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

NHS centre	Nottingham University Hospitals NHS Trust
Expenditure	<£0.5 million
Caseload	196

Outcomes collated	• % of patients having a pneumothorax: 3% • % of patients having a renal angiolipoma bleed: 0% • % of patients having an FEV_1 decline of >150 mL per annum: 13%
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Lysosomal storage disorders service (children and adults)

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

There are licensed disease-modifying treatments for nine 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.
- Batten's Disease, Ceroid Lipofuscinosis Neuronal 2 (CLN2)

NHS centres	Birmingham Women's and Children's Hospital NHS Foundation Trust Cambridge University Hospitals NHS Foundation Trust Great Ormond Street Hospital for Children NHS Foundation Trust

	Manchester University NHS Foundation Trust Royal Free London NHS Foundation Trust Salford Royal NHS Foundation Trust University College London Hospitals NHS Foundation Trust University Hospitals Birmingham NHS Foundation Trust
Expenditure	>£50 million
Active caseload	2,535
Outcomes collated	• Proportion of Fabry patients having a cardiac device implanted among patients treated for 3 years or more: - Birmingham Women's and Children's: 0% - Cambridge University: 2% - Great Ormond Street: 0% - Manchester University: 0% - Royal Free: 2% - Salford: 4% - University College London: 6% - University Hospitals Birmingham: 0% • Proportion of Fabry patients having a new stroke among patients treated for 3 years or more: - Birmingham Women's and Children's: 0% - Cambridge University: 0% - Great Ormond Street: 0% - Manchester University: 0% - Royal Free: 0% - Salford: 1% - University College London: 4% - University Hospitals Birmingham: 0% • Proportion of Gaucher patients having a hospital admission for bone crisis among patients treated for 3 years or more: - Birmingham Women's and Children's: 0%

	- Cambridge University: 0% - Great Ormond Street: 0% - Manchester University: 0% - Royal Free: 0% - Salford: 0% - University College London: 0% - University Hospitals Birmingham: 0% • Proportion of MPS patients having a new cranio cervical episode among patients treated for 3 years or more: - Birmingham Women's and Children's: 0% - Cambridge University: 50% - Great Ormond Street: 0% - Manchester University: 0% - Royal Free: 0% - Salford: 0% - University College London: 0% - University Hospitals Birmingham: 0% • Proportion of patients initiating renal replacement therapy among patients treated for 3 years or more: - Birmingham Women's and Children's: 0% - Cambridge University: 0% - Great Ormond Street: 0% - Manchester University: 0% - Royal Free: 2% - Salford: 4% - University College London: 2% - University Hospitals Birmingham: 0%
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

McArdle’s disease service

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

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

NHS centre	University College London Hospitals NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Caseload	221
Outcomes collated	• Median functional capacity – 12MWD: 830 • Percentage of patients requiring hospital assessment: 9.6% • Median quality of life (SF36) score: physical functioning: 39 Note: 12MWD is a 12-minute walking distance test used to estimate functional exercise capacity.
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Multiple sclerosis management service for children

Multiple Sclerosis (MS) is a condition of the central nervous system characterised by chronic brain inflammation which damages the myelin coating around nerve fibres, a process known as demyelination. There is currently no cure for MS, but treatments and specialist help can help to control disease activity, decrease disability from the condition and reduce ongoing symptoms. There is a particular need to focus on the early recognition of these serious neuroinflammatory disorders to reduce long-term morbidity and neuro-disability and for early intervention.

Patients with MS or ‘MS-like’ conditions are assessed and treated in specialist paediatric neurology centres in age-appropriate outpatient and inpatient settings by professionals working in multi-disciplinary teams with expertise in multiple sclerosis.

NHS centres	**North Hub Lead Centre (single centre with three units):** Alder Hey Children's NHS Foundation Trust Manchester University NHS Foundation Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust **Midland hubs:** Birmingham Women's and Children's Hospital NHS Foundation Trust Cambridge University Hospitals NHS Foundation Trust **London and the South hubs:** Great Ormond Street Hospital for Children NHS Foundation Trust Guy's and St Thomas' NHS Foundation Trust
Expenditure	>£0.5 million but <£1 million
Caseload	387
Outcomes collated	• Proportion of patients achieving a slowing in progression of disease using Annualised Relapse Rate (ARR): - Alder Hey Children's NHS FT: Data not submitted - Birmingham Women's and Children's: 80% - Cambridge University Hospitals NHS FT: 100% - Great Ormond Street: Data not submitted - Guy’s and St Thomas': 36% - Manchester: University NHS FT: Data not submitted - Newcastle Upon Tyne: Data not submitted • Proportion of patients achieving a slowing in progression of disease using No Evidence of Disease Activity (NEDA) - no evidence of relapse: - Alder Hey Children's NHS FT: 33%

- Birmingham Women's and Children's: 80%
- Cambridge University Hospitals NHS FT: 33%
- Great Ormond Street: Data not submitted
- Guy’s and St Thomas': 9%
- Manchester: University NHS FT: 0%
- Newcastle Upon Tyne: 67%

- Proportion of patients achieving a slowing in progression of disease using No Evidence of Disease Activity (NEDA) - no evidence of relapse and no MRI activity:
  - Alder Hey Children's NHS FT: 33%
  - Birmingham Women's and Children's: 31%
  - Cambridge University Hospitals NHS FT: 33%
  - Great Ormond Street: Data not submitted
  - Guy's and St Thomas': 55%
  - Manchester: University NHS FT: 0%
  - Newcastle Upon Tyne: 100%
- Proportion of patients achieving a slowing in progression of disease using No Evidence of Disease Activity (NEDA) - no evidence of relapse, no MRI activity and no evidence of disease progression:
  - Alder Hey Children's NHS FT: 33%
  - Birmingham Women's and Children's: 31%
  - Cambridge University Hospitals NHS FT: 33%
  - Great Ormond Street: Data not submitted
  - Guy's and St Thomas': 55%
  - Manchester: University NHS FT: 0%
  - Newcastle Upon Tyne: 67%
- Proportion of patients starting the first dose of disease modifying therapy within 4 weeks of the agreement of the treatment plan, as defined in the service specification:
  - Alder Hey Children's NHS FT: 100%
  - Birmingham Women's and Children's: 85%
  - Cambridge University Hospitals NHS FT: 0%
  - Great Ormond Street: 23%

	- Guy's and St Thomas': 20% - Manchester: University NHS FT: 75% - Newcastle Upon Tyne: 67% Note: This is a newly commissioned service and commissioners are working with providers to improve data reporting and key outcome measures.
Geographical equity access	• Numbers insufficient for robust analysis due to being a newly commissioned service.

Neurofibromatosis type 2 service (all ages)

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

The service includes:

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

NHS centres	Cambridge University Hospitals NHS Foundation Trust Guy's and St Thomas' NHS Foundation Trust Manchester University NHS Foundation Trust Oxford University Hospitals NHS Trust

Expenditure	>£5 million but <£10 million
Caseload	1,011
Outcomes collated	• Hearing preservation: Proportion of NF2 patients diagnosed since inception of service maintaining useful hearing (target >80%): - Cambridge University: 97% - Guy's and St Thomas': 94% - Manchester University: 93% - Oxford University: 93% • Survival: Proportion of patients who have died prematurely from an NF2 related complication (target <5%): - Cambridge University: 1% - Guy's and St Thomas': data supressed to maintain patient confidentiality0% - Manchester University: 1% - Oxford University: 0%
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Neuromyelitis optica service (adults and children)

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

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

NHS centres	Oxford University Hospitals NHS Trust

	The Walton Centre NHS Foundation Trust
Expenditure	>£1 million but <£5 million
First evaluations	153
Outcomes collated	• Median annualised relapse rate: - Oxford University: 8% - The Walton Centre: 0
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Ocular oncology service (adults)

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

There are a number of different treatment modalities:

- surgery
- radiotherapy
- phototherapy
- cryotherapy
- chemotherapy.

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

NHS centre	Liverpool University Hospitals NHS Foundation Trust Moorfields Eye Hospital NHS Foundation Trust

	Sheffield Teaching Hospitals NHS Foundation Trust
Expenditure	>£5 million but <£10 million
Positive assessment	718
Outcomes collated	• % primary enucleation among patients with melanoma: - Liverpool University: 17% - Moorfields: 24% - Sheffield Teaching: 26% • % secondary enucleation among patients with melanoma: - Liverpool University: 0% - Moorfields: 0% - Sheffield Teaching: 1% • % developing metastatic disease among patients with melanoma: - Liverpool University: 2% - Moorfields: 1% - Sheffield Teaching: 2%
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Open fetal surgery to treat foetuses with open spina bifida

Spina bifida is an in-utero condition where the spinal column and cord are not fully formed. Babies born with this condition are often unable to walk, incontinent of urine and faeces, may develop hydrocephalus due to incomplete closure of the spinal canal and often require postnatal neurosurgical interventions.

For a carefully selected group of women and their babies open fetal surgery (operating on the baby while it is still in the womb) can be used to successfully close the spinal defect and achieve good clinical outcomes for the baby.

The service is provided by two fetal surgery centres that provide assessment, open fetal surgery and supporting medical services. The service is delivered by an expert MDT in a shared care pathway with existing local maternity units / Regional Fetal Medicine Units (RFMUs) and regional neurosurgery centres.

NHS centre	University College London Hospital NHS Foundation Trust Universitair Ziekenhuis, Leuven, Belgium
Expenditure	Less than £0.5m
No. of operations	The number of operations is less than five, so the data has been suppressed to maintain patient confidentiality
Outcomes collated	Outcomes are to be developed
Geographical equity access	Numbers insufficient for robust analysis due to being a newly commissioned service

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	Liverpool University Hospitals NHS Foundation Trust Manchester University NHS Foundation Trust Sheffield Teaching Hospitals NHS Foundation Trust University College London Hospitals NHS Foundation Trust

Expenditure	>£1 million but <£5 million
cases reviewed	3,955
Outcomes collated	• % of simple cases reported within 7 working days: - Liverpool University: 88% - Manchester University: 89% - Sheffield Teaching: 93% - University College London: 91% • % of complex cases reported within 10 working days: - Liverpool University: 91% - Manchester University: 90% - Sheffield Teaching: 95% - University College London: 90% • % of all cases reported within 21 working days: - Liverpool University: 98% - Manchester University: 100% - Sheffield Teaching: 100% - University College London: 100%
Geographical equity access	Data not available or not comparable

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

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

During OOKP, patients are initially assessed by ophthalmic and maxillofacial consultants, involving examination of the eyes, teeth and mouth. OOKP is then a two-stage procedure that firstly involves the extraction of the patient’s own tooth

and bone, which are then fashioned into a ‘bolt’ and placed within the eye for supporting a synthetic optical cylinder.

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

NHS centre	Brighton and Sussex University Hospitals NHS Trust
Expenditure	<£0.5 million
Stage 2 surgery	Data suppressed to maintain patient confidentiality
Outcomes collated	• % patients with visual acuity 6/12 or better at 12 months post operation: data supressed to maintain patient confidentiality
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Paediatric intestinal pseudo-obstructive disorders service

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

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

The service treats about 20 children each year.

NHS centre	Great Ormond Street Hospital for Children NHS Foundation Trust
Expenditure	>£1 million but <£5 million

Number of new patient referrals	7
Outcomes collated	• Proportion of patients admitted for Phase 1 admission within 8 weeks: data supressed to maintain patient confidentiality • Proportion of definitive diagnosis made and results fed back via MDT within 4 weeks of Phase 1 admission discharge: data supressed to maintain patient confidentiality
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Pancreas transplantation service (adults)

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

NHS centres	Cambridge University Hospitals NHS Foundation Trust Guy's and St Thomas' NHS Foundation Trust Imperial College Healthcare NHS Trust Manchester University NHS Foundation Trust Oxford University Hospitals NHS Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust
Expenditure	>£5 million but <£10 million
Number of transplants	131
Outcomes collated	• 1-year risk-adjusted patient survival for first SPK transplant from deceased donors: - Cambridge University: 99% - Guy's and St Thomas': 98% - Imperial College: 100% - Manchester University: 100%

	- Oxford University: 97% - Newcastle upon Tyne: 100% • 5-year risk-adjusted patient survival for first SPK transplant from deceased donors: - Cambridge University: 98% - Guy's and St Thomas': 86% - Imperial College: 92% - Manchester University: 87% - Oxford University: 86% - Newcastle upon Tyne: 92%
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Paroxysmal nocturnal haemoglobinuria

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

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

Outreach clinics are held in locations outside of the centres.

NHS centres	Leeds Teaching Hospitals NHS Trust King's College Hospital NHS Foundation Trust
Expenditure	>£50 million
Caseload	867
Outcomes collated	• 5-year relative survival rate: - Leeds Teaching: Data not submitted

	- King's College: 87% • Median transfusions per patient in previous 12 months: - Leeds Teaching: 6 - King's College: 3 *All outcome measures are discussed at the annual clinical meeting including reasons for apparent variation
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Primary ciliary dyskinesia management service (children)

Primary ciliary dyskinesia (PCD) is a genetic condition in which the microscopic cells in the respiratory system called cilia do not function normally. Ciliary dysfunction prevents the clearance of mucous from the lungs, paranasal sinuses and ears. Recurring respiratory infections can lead to an irreversible scarring and obstruction in the bronchi (bronchiectasis) and severe lung damage. Cilia are also present in the ventricles of the brain and in the reproductive system so ciliary dysfunction can also affect other parts of the body.

Primary ciliary dyskinesia management services include services provided by Highly Specialist Primary Ciliary Dyskinesia Management centres including outreach when delivered as part of a provider network. This provision applies to adults. Patients will transition into the service from the paediatric HSS PCD service.

The figures below are for children only as the adult service only started in November 2019.

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

Expenditure	>£1 million but <£5 million
Caseload	1,100
Outcomes collated	• % of patients in the PCD management service offered an annual review appointment (consisting of the processes listed in the service specification: - Leeds Teaching: 90% - Royal Brompton & Harefield: 94% - Southampton: 100% - Leicester: 95% • % of patients seen by a physiotherapist at annual review: - Leeds Teaching: 100% - Royal Brompton & Harefield: 94% - Southampton: 100% - Leicester: 100% • % patients seen by a nurse specialist at annual review: - Leeds Teaching: 94% - Royal Brompton & Harefield: 94% - Southampton: 100% - Leicester: 84% • % of patients seen by an ENT specialist at annual review: - Leeds Teaching: 0% - Royal Brompton & Harefield: 76% - Southampton: 100% - Leicester: 96% *All outcome measures are discussed at the annual clinical meeting including reasons for apparent variation
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Primary malignant bone tumours service (adults and adolescents)

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

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

NHS centres	Oxford University Hospitals NHS Trust Royal National Orthopaedic Hospital NHS Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust The Robert Jones and Agnes Hunt Orthopaedic Hospital NHS Foundation Trust The Royal Orthopaedic Hospital NHS Foundation Trust
Expenditure	>£10 million but <£20 million
Number of confirmed cases	409
Outcomes collated	• % 3-year local recurrence among patients having limb salvage: - Oxford University: data not submitted - Robert Jones and Agnes Hunt: 7% - Royal Orthopaedic: 11% - Royal National Orthopaedic: 4% - Newcastle upon Tyne: 12% • % limb salvage: - Oxford University: 82% - Robert Jones and Agnes Hunt: 86% - Royal Orthopaedic: 73% - Royal National Orthopaedic: 93% - Newcastle upon Tyne: 81%

	• % 3-year prosthesis infection/loosening: - Oxford University: 13% - Robert Jones and Agnes Hunt: 0% - Royal Orthopaedic: 15% - Royal National Orthopaedic: 10% - Newcastle:24%
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Proton beam therapy service (adults and children)

Proton beam therapy (PBT) is a type of radiotherapy that uses a beam of high energy protons, (rather than high energy x-rays) to treat specific types of cancer. The physical properties of protons, results in almost no radiation dose being deposited in the normal tissue beyond the tumour.

PBT is a highly complex technology and the services are part of major cancer centres, including highly specialist surgery and cancer services. The PBT service improves cancer outcomes, reduce morbidity arising from treatment, and support the patient and family throughout their cancer journey and beyond.

Centres	The Christie NHS Foundation Trust University of Florida Health Proton Institute, Jacksonville, USA Westdeutsches Protonentherapiezentrum (WPE) Essen, Germany Paul Scherrer Institut, Villigen, Switzerland
Expenditure	Overseas providers: >£30 million but <£50 million The Christie NHS Foundation Trust: >£20 million but <£30 million
Number of patients approved for referral	Total patients overseas: 74 Total patients The Christie: 264 (Treatment completed 143 – Adult 51, Teen/Young Adult, 37, Paeds 55)

Outcomes collated	• Number of adults referred: 79 • Number of adults planned: 59 • Number of adults treated: 59 • Number of teenagers & young adults referred: 70 • Number of teenagers & young adults planned: 62 • Number of teenagers & young adults treated: 58 • Number of children referred: 125 • Number of children planned: 104 • Number of children treated: 100
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Pseudomyxoma peritonei service (adults)

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

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

Treatment options include:

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

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

NHS centre	Hampshire Hospitals NHS Foundation Trust

	The Christie NHS Foundation Trust
Expenditure	>£20 million but <£30 million
Major full cytoreduction	232
Outcomes collated	• 5-year patient survival – all operative cases: - Christie: 78% - Hampshire: 60% • 5-year patient survival – complete cytoreduction: - Christie: 91% - Hampshire: 68%
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Pulmonary hypertension service for children

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

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

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

NHS centre	Great Ormond Street Hospital for Children NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Caseload	539

Outcomes collated	• % of patient followed up in the year with at least one functional class measure (denominator) who achieved a functional class of 2 or better (numerator): 68% • Proportion/% of children receiving epoprostenol who required a line change due to infection: 18% • Proportion of children receiving epoprostenol who experienced a line-related bloodstream infection: 6%
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

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).

Through the network of adult pulmonary hypertension units, all patients with a diagnosis of CTEPH are referred for consideration of surgery.

NHS centre	Papworth Hospital NHS Foundation Trust [now Royal Papworth Hospital NHS Foundation Trust]
Expenditure	>£5 million but <£10 million
Surgical Operations	182
Outcomes collated	• 90-day patient survival: 96% • 3-year patient survival: 93% • In-hospital mortality: 3%
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Rare mitochondrial disorders service (adults and children)

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

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

The Highly Specialist Mitochondrial Disorders Centres provide:

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

The service diagnoses about 280 new patients each year.

NHS centres	Oxford University Hospitals NHS Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust University College London Hospitals NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Outpatient referrals	410
Outcomes collated	• % of patients given an alert card*: - Oxford University: 94% - Newcastle upon Tyne: 100% - University College London: 62%

	Note: * That the provider must give every patient with a rare disease an ‘alert card’, including information about: the patient’s rare disease; any particular aspects of the treatment of that rare disease that need to be taken into account in providing care to that patient; and details of how to contact an individual expert in that patient’s care
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Retinoblastoma service (children)

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

The treatment modalities are as follows:

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

NHS centres	Barts Health NHS Trust Birmingham Women’s and Children's Hospital NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Confirmed patients	51
Outcomes collated	• % 5-year survival: - Barts Health: 100% - Birmingham Women’s and Children’s: 100%

	• % primary enucleation: - Barts Health: 29% - Birmingham Women's and Children's: 33%
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Severe acute porphyria (adults and children)

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

The service comprises two elements:

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

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

NHS centres	King's College Hospital NHS Foundation Trust University Hospital of Wales
Expenditure	>£0.5 million but <£1 million
Active caseload	131
Outcomes collated	• % mortality rate: - King's College: 0% - Wales (Cardiff): 0%

	• % of patients having four or more hospital admissions (porphyria-related) in the previous 12 months - King's College: 7% - Wales (Cardiff): 5%
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions

Severe combined immune deficiency and related disorders service (children)

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

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

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

NHS centres	Great Ormond Street Hospital for Children NHS Foundation Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust
Expenditure	>£10 million but <£20 million
Number of transplants	62
Outcomes collated	• 2-year survival rate: - Great Ormond Street: 95% - Newcastle upon Tyne: 89%

Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Small bowel transplantation service (adults)

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

NHS centres	Cambridge University Hospitals NHS Foundation Trust Oxford University Hospitals NHS Trust
Expenditure	>£5 million but <£10 million (adults and children combined)
Number of transplants	12
Outcomes collated	• % unadjusted 90-day patient survival for first intestine transplants between 01/04/2010- and 31/03/2020: Cambridge University: 100% (not including liver) Cambridge University: 88.5% (including liver) Oxford University: 91.3% (not including liver) • % unadjusted 1-year patient survival for first intestine transplants between 01/04/2010 to 31/03/2020: Cambridge University: 87.3% (not including liver) Cambridge University: 73.9% (including liver) Oxford University: 88.8% (not including liver) • % unadjusted 5-year patient survival for first intestine transplants between 01/04/2010 and 31/03/2020: Cambridge University: 79.1% (not including liver) Cambridge University: 39.3% (including liver) Oxford University: 65.8% (not including liver)
Geographical equity access	Combined analysis conducted for adult and paediatric service. The expected numbers of patients are accessing the service from all NHSE regions

Small bowel transplantation service (children)

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

NHS centres	Birmingham Women's and Children's Hospital NHS Foundation Trust King's College Hospital NHS Foundation Trust
Expenditure	>£5 million but <£10 million (adults and children combined)
Number of transplants	The number of transplants is less than five, so the data has been suppressed to maintain patient confidentiality
Outcomes collated	• % unadjusted 90-day patient survival for first intestine transplants including liver between 01/04/2010 and 31/03/2020: - Birmingham Women's and Children: 84.6% - King's College: 100% • % unadjusted 1-year patient survival for first intestine transplants including liver between 01/04/2010 and 31/03/2020: - Birmingham Women's and Children: 76.9% - King's College: 90.9%
Geographical equity access	Combined analysis conducted for adult and paediatric service. The expected numbers of patients are accessing the service from all NHSE regions

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 Women's and Children's Hospital NHS Foundation Trust King's College Hospital NHS Foundation Trust Leeds Teaching Hospitals NHS Trust
Expenditure	>£10 million but <£20 million
Inpatient episodes	1,044
Outcomes collated	• % survival for patients diagnosed with extrahepatic biliary atresia (EHBA) at 16 years of age (% alive on 16th birthday with or without native liver): - Birmingham Women's and Children: 93% - King's College: 93% - Leeds: Data not submitted • % patients diagnosed with autoimmune liver disease (AILD) to be in biochemical remission on 16th birthday (biochemical remission as per laboratory references) - Birmingham Women's and Children: 51% - King's College: 100% - Leeds: Data not submitted
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Stickler syndrome diagnostic service (adults and children)

Stickler syndrome is an inherited disorder of connective tissue associated with cleft palate, deafness and arthropathy. It is the commonest inherited cause of rhegmatogenous retinal detachment in children (where fluid passes into the space between the retina and the retinal pigment layer). Although the systemic features

are widespread, the sight-threatening complications are generally the most serious, particularly the risk of giant retinal tear, which is frequently bilateral and, if untreated, can lead to blindness.

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

NHS centre	Cambridge University Hospitals NHS Foundation Trust
Expenditure	>£0.5 million but <£1 million
Index patients	70
Outcomes collated	• % of patients with a definitive diagnosis or diagnosis ruled out: 89%
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Total Pancreatectomy with Islet Autotransplant

Chronic pancreatitis (CP) is chronic inflammation of the pancreas characterised by an irreversible, permanent and progressive destruction of pancreatic tissue. It may be hereditary or acquired.

It is a disabling condition with symptoms including severe, persistent, intractable abdominal pain and diabetes.

Total pancreatectomy with islet auto transplant surgery involves removal of the pancreas followed by islet auto transplantation (a procedure where the patient's own islet cells are isolated and infused into their liver).

NHS centre	King's College Hospital NHS Foundation Trust Oxford University Hospitals NHS Trust The Newcastle Upon Tyne Hospitals NHS Foundation Trust; University Hospitals of Leicester NHS Trust

Expenditure	More than £1m but less than £5m
No. of procedures	The number of procedures is less than five, so the data has been suppressed to maintain patient confidentiality
Outcomes collated	No data available due to being a newly commissioned service
Geographical equity access	Numbers insufficient for robust analysis due to being a newly commissioned service

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

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

NHS centres	Alder Hey Children's Hospital Great Ormond Street Hospital for Children NHS Foundation Trust
Expenditure	<£0.5 million
Number of procedures	39
Outcomes collated	• Proportion of neonates alive and well: – Alder Hey Children's Hospital: 100% – Great Ormond Street: 75% • Proportion of neonates alive but impaired: – Alder Hey Children's Hospital: 0% – Great Ormond Street: 21%

	• Proportion of neonates who died – Alder Hey Children's Hospital: 0% – Great Ormond Street: 4% • Proportion of infants alive and well: – Alder Hey Children's Hospital: 100% – Great Ormond Street: No data • Proportion of infants alive but impaired: – Alder Hey Children's Hospital: 0% – Great Ormond Street: Data not submitted • Proportion of infants who died: – Alder Hey Children's Hospital: 0% – Great Ormond Street: Data not submitted • Proportion of older children alive and well: – Alder Hey Children's Hospital: Data not submitted – Great Ormond Street: 0% • Proportion of older children alive but impaired: – Alder Hey Children's Hospital: Data not submitted – Great Ormond Street: 100% • Proportion of older children who died: – Alder Hey Children's Hospital: Data not submitted – Great Ormond Street: 0% • Total number treated as neonatal patients: – Alder Hey Children's Hospital: 8 – Great Ormond Street: 28
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

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

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

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

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

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

NHS centres	Great Ormond Street Hospital for Children NHS Foundation Trust Manchester University NHS Foundation Trust Royal Papworth Hospital NHS Foundation Trust [now Royal Papworth Hospital NHS Foundation Trust Royal Brompton & Harefield NHS Foundation Trust The Newcastle upon Tyne Hospitals NHS Foundation Trust University Hospitals Birmingham NHS Foundation Trust
Expenditure	Figure included in heart and lung transplant
Number of procedures	Long-term VAD procedures: 82 Short-term VAD procedures: 87
Outcomes collated	• Bridging devices used in paediatrics: - 35% of patients received a transplant within 90 days of implantation and the 1-year patient survival rate from the point of implant was 76.2% • Adults given a long-term VAD as bridge to transplant who received a transplant within 3 years: 14%

	• 3-year survival rate in adults with long-term bridging devices: 60.1 which has improved over time (58.4% in 2018/19 and 46.6% in 2016/2017) • Unadjusted patient survival rates after first adult DBD (donor after brain death) heart transplant, by long-term support status: - 30 days: 80% - 90 days: 73.3% - one year: 69.5% Note: NHS BT in conjunction with NHSE have a formal process for investigating transplant centres with significant variation in mortality rates
Geographical equity access	The expected numbers of patients are accessing the service from all NHSE regions and have been calculated for the combined adult and paediatric patient groups

Wolfram syndrome service (adults and children)

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

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

NHS centres	Birmingham Women's and Children's Hospital NHS Foundation Trust University Hospitals Birmingham NHS Foundation Trust
Expenditure	<£0.5 million
Caseload	69
Outcomes collated	• % of children with HbA1c in target range: - Birmingham Women's and Children: data suppressed to

	maintain patient confidentiality • % of adults with a BMI <35: - Birmingham University: 90% Note: glycated haemoglobin (HbA1c) is measured primarily to identify the 3-month average plasma glucose concentration.
Geographical equity access	The number of patients accessing the service from NHSE regions is different to what is expected. The potential reasons for this are being explored with the service.

Appendix A: UK-wide commissioning arrangements for highly specialised services during 2019/20

Name of Service	NHS England commissioning arrangements on behalf of the devolved administrations
Alkaptonuria service (adults)	Fully commissioned on behalf of England & Scotland
Alström syndrome service (adults and children)	Fully commissioned on behalf of England & Scotland
Ataxia telangiectasia services for adults	Fully commissioned on behalf of England & Scotland
Ataxia telangiectasia services for children	Fully commissioned on behalf of England & Scotland
Atypical haemolytic uraemic syndrome (adults and children)	Fully commissioned on behalf of England only
Auditory brainstem implant for patients with congenital abnormality of the auditory nerves or cochleae	Fully commissioned on behalf of England only
Autologous intestinal reconstruction service for adults	Fully commissioned on behalf of England only
Bardet Biedl syndrome service (adults and children)	Fully commissioned on behalf of England & Scotland
Barth syndrome service (adults and children)	Fully commissioned on behalf of England & Scotland
Beckwith-Wiedemann syndrome with macroglossia service (children)	Fully commissioned on behalf of England & Scotland
Behçet's syndrome service (adults and adolescents)	Fully commissioned on behalf of England only
Bladder exstrophy service (children)	Fully commissioned on behalf of England & Scotland
Breast radiotherapy injury rehabilitation service (a discrete cohort of adult females)	Fully commissioned on behalf of England only
Cardiothoracic Transplantation Service (Paediatrics)	Fully commissioned on behalf of England, in-part for Scotland by arrangement, in full for NI
Choriocarcinoma service (adults and adolescents) Gestational trophoblastic disease	Fully commissioned on behalf of UK (Pre-1991)
Chronic pulmonary aspergillosis service (adults)	Fully commissioned on behalf of England & Scotland

Name of Service	NHS England commissioning arrangements on behalf of the devolved administrations
Complex childhood osteogenesis imperfecta service	Fully commissioned on behalf of England only
Complex Ehlers Danlos syndrome service (adults and children)	Fully commissioned on behalf of England & Scotland
Complex neurofibromatosis type I service (adults and children)	Fully commissioned on behalf of England only
Complex tracheal disease service (children)	Fully commissioned on behalf of England & Scotland
Congenital hyperinsulinism service (children)	Fully commissioned on behalf of England & Scotland
Craniofacial service (adults and children)	Fully commissioned on behalf of UK (Pre-1991)
Cryopyrin associated periodic fever syndromes (CAPS) also know as Autoinflammatory Diseases treated with IL blockers	Fully commissioned on behalf of England & Scotland
Diagnostic service for amyloidosis (all ages0	Fully commissioned on behalf of England & Scotland
Diagnostic service for primary ciliary dyskinesia (adults and children)	Fully commissioned on behalf of England & Scotland
Diagnostic service for rare neuromuscular disorders (adults and children)	Fully commissioned on behalf of England & Scotland
DNA Nucleotide Excision Repair Disorders Service	Fully commissioned on behalf of England only
Encapsulating peritoneal sclerosis treatment service (adults)	Fully commissioned on behalf of England only
Epidermolysis bullosa service (adults and children)	Fully commissioned on behalf of England & Scotland
Extra corporeal membrane oxygenation service for adults	Fully commissioned on behalf of England only
Extra corporeal membrane oxygenation service for neonates, infants and children with respiratory failure	Fully commissioned on behalf of England only
Ex-vivo partial nephrectomy service (adults)	Fully commissioned on behalf of England only
Gender identity development service for children and adolescents	Fully commissioned on behalf of England & Scotland
Hand and upper limb reconstruction using vascularised composite allotransplantation	Fully commissioned on behalf of England & Scotland
Heart Transplantation Service (adults)	Fully commissioned on behalf of England, in-part for Scotland by arrangement, in full for NI
High Consequence Infectious Diseases Special Isolation Unit (Airbourne) Adults	Fully commissioned on behalf of England, Scotland and NI
High Consequence Infectious Diseases, Special Isolation Unit (Airborne)Children aged 16 and under	Fully commissioned on behalf of England, Scotland and NI
Insulin Resistant Diabetes (Adults and Children)	Fully commissioned on behalf of England only
Islet transplantation service (adults)	Fully commissioned on behalf of England only
Live Liver transplantation (all ages)	Fully commissioned on behalf of England only

Name of Service	NHS England commissioning arrangements on behalf of the devolved administrations
Liver transplantation service - ADULTS	Fully commissioned on behalf of England, NI & Wales and by exception for Scotland
Liver transplantation service CHILDREN	Fully commissioned on behalf of UK (Pre-1991)
Lung Transplantation Service (Adults)	Fully commissioned on behalf of England, in-part for Scotland by arrangement, in full for NI
Lymphangioleiomyomatosis	Fully commissioned on behalf of England & Scotland
Lysosomal storage disorders service (Children & Adults)	Fully commissioned on behalf of England, in-part for Scotland (not drugs) & in-part for NI (not ERT drugs)
McArdle's disease service (children)	Fully commissioned on behalf of England & Scotland
Multiple Sclerosis Management Service for Children	Fully commissioned on behalf of England only
Neurofibromatosis type 2 service (All Ages)	Fully commissioned on behalf of England & Scotland
Neuromyelitis optica service (adults and children)	Fully commissioned on behalf of England & Scotland
Ocular oncology service (adults and adolescents)	Fully commissioned on behalf of England, from devolved administrations for Scotland
Open fetal surgery to treat fetuses with open spina bifida	Fully commissioned on behalf of UK (Pre-1991)
Ophthalmic pathology service (adults and children)	Fully commissioned on behalf of England, from devolved administrations for Scotland
Osteo-odonto-keratoprosthesis service for corneal blindness (adults)	Fully commissioned on behalf of England only
Paediatric intestinal pseudo-obstructive disorders service	Fully commissioned on behalf of England & Scotland
Pancreas transplantation service (adults)	Fully commissioned on behalf of England, from devolved administrations for Scotland
Paroxysmal nocturnal haemoglobinuria	Fully commissioned on behalf of England and in part for Scotland (service only not drugs)
Primary Ciliary Dyskinesia (adults) (Management)	Fully commissioned on behalf of England only
Primary ciliary dyskinesia management service (children)	Fully commissioned on behalf of England & Scotland
Primary Malignant Bone Tumours service (adults and adolescents)	Fully commissioned on behalf of England only
Proton Beam Therapy (PBT) (All Ages)	Fully commissioned on behalf of England, Scotland and NI
Proton beam therapy overseas service (adults and children)	Fully commissioned on behalf of England, Scotland and NI
Pseudomyxoma peritonei service (adults)	Fully commissioned on behalf of England & Scotland
Pulmonary hypertension service for children	Fully commissioned on behalf of England and in part for Scotland (service only not drugs)

Name of Service	NHS England commissioning arrangements on behalf of the devolved administrations
Pulmonary thromboendarterectomy service (adults and adolescents)	Fully commissioned on behalf of England & Scotland
Rare mitochondrial disorders service (adults and children)	Fully commissioned on behalf of England & Scotland
Retinoblastoma service (children)	Fully commissioned on behalf of UK (Pre-1991)
Severe acute porphyria	Fully commissioned on behalf of England & Scotland
Severe combined immune deficiency and related disorders service (children)	Fully commissioned on behalf of England & in-part for Scotland
Small bowel transplantation service - Adults	Fully commissioned on behalf of England only
Small bowel transplantation service - Children	Fully commissioned on behalf of England & Scotland
Specialist paediatric liver disease service	Fully commissioned on behalf of UK (Pre-1991)
Stickler syndrome diagnostic service (adults and children)	Fully commissioned on behalf of England & Scotland
Total Pancreatectomy with Islet Autotransplant	Fully commissioned on behalf of England only
Vein of Galen malformation service (adults and children)	Fully commissioned on behalf of England & Scotland
Ventricular Assist Devices (VADs) as a bridge to heart transplantation or myocardial recovery (All Ages)	Fully commissioned on behalf of England & in-part for Scotland
Wolfram syndrome service (adults and children)	Fully commissioned on behalf of England & Scotland