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Highly specialised services 2020/21

This report provides the following information about the provision and commissioning highly specialised services during 2020/21:

- a description of each service
- a list of the expert centres that deliver the service
- NHS England's expenditure on 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
- an update on new highly specialised services.

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

28 November 2023

Contents

1. Introduction

This report provides the following information about the provision and commissioning of highly specialised services during 2020/21:

- a description of each service
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Appendix B summarises NHS England's commissioning arrangements for highly specialised services across the devolved nations.

During the COVID-19 pandemic, highly specialised services adapted how they delivered their services to patients, to ensure these remained accessible but safe to patients. There was rapid adoption of a range of virtual technologies and subsequent learning about their optimal use.

Specialised services

NHS England is responsible for commissioning specialised services to meet a wide range of health and care needs. In 2020/21 it spent £20.5 billion on specialised services.

Four factors determine whether NHS England commissions a service as a prescribed specialised service (rather than by clinical commissioning groups [CCGs] as a non-specialised service):

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

Highly specialised services

A subset of specialised services are 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. For this reason they are most appropriately delivered and co-ordinated nationally through a few expert

centres. This model of delivery makes it easier to recruit appropriately qualified professionals and to ensure that they have the level of training needed to maintain their experience and expertise, thereby offering the best possible service to patients. 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.

However, planning highly specialised services on a national, rather than local, basis presents the Highly Specialised Commissioning Team (HSCT) with a challenge in ensuring equitable access to services, given that with only a small number of expert centres, a patient's nearest centre may be a long way away. It is also important to have a robust process for selecting and monitoring the centres that provide these services, given the high level of expertise they need to have.

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

- NHS England 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 (NHSBT) 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 advising on which services or technologies should be recommended 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 clinical quality of services and makes recommendations on any action required as a consequence of poor outcomes, as well as ensuring proper provision of commissioned services, 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 on the implementation of the UK Strategy for Rare Diseases.

Expenditure on highly specialised services

A special financial regime was put in place during 2020/21 – because of the COVID-19 pandemic – and individual service budgets were not set.

This report gives NHS England's expenditure on each service, in 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 a high level of clinical outcome monitoring is in place. The HSCT work closely with the services and the NHS England Quality Team to ensure data completeness.

The clinical outcome data for each centre providing a service is collated on an annual basis and this report presents the data for 2020/21, unless otherwise stated. Clinical outcomes data are presented and discussed at the annual audit meeting for the service. Any variation in outcomes is a stimulus for challenge and learning (or confirmation of good practice) and these conversations are followed up subsequent to the meeting and throughout the year. NHSBT in conjunction with NHS England has a formal process for investigating transplant centres with significant variation in mortality rates triggered through routine statistical monitoring. The HSCT also regularly review the organ-specific reports for the relevant highly specialised service and discuss relevant data with the providers at the annual audit meetings or in year as appropriate. All clinical outcomes for each centre are reviewed annually and unless indicated under each service heading, compare favourably with previous years, align with other centres and meet the standards expected. If required and appropriate, benchmarking against international outcome data can be undertaken.

Outcome information cannot be published for some services because the small number of patients involved risks identification of individual patients, or the dataset is too small to analyse. In these cases, the data is reviewed and held by the HSCT and discussed at the annual audit meetings. Where the number of patients is small, data can be aggregated over a number of years to create a meaningful dataset.

Geographical variation in 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 implied trade-off is that access to care may be difficult for those patients who need to travel long distances to an expert centre. Hence it is incumbent on the HSCT to monitor the geographical access to highly specialised services.

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

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

The observed and expected figures are unlikely to be identical 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 unlikely to be due to chance (SCV >0.2), genuine clusters of disease are possible. 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 seems to be rarer in the north east of England.

For those services where the SCV is above 0.2, the HSCT and the service review the information in greater detail to understand the possible causes. They will then explore options and take specific actions to mitigate any variation. These mitigations can include developing new referral pathways, awareness raising by participating in regional clinical teaching and developing new outreach clinics. In exceptional cases a revision to the service specification may be needed.

The HSCT analyse geographical variation for each service every 3 to 4 years. For most analyses, patients are mapped if they received the intervention (eg transplant) or have a confirmed diagnosis of the relevant condition (eg xeroderma pigmentosum).

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

2. New highly specialised services commissioned during 2020/21

Onasemnogene abeparvovec for spinal muscular atrophy type 1

In July 2021, NICE recommended the gene therapy onasemnogene abeparvovec (Zolgensma®) as an option for treating 5q spinal muscular atrophy (SMA) with a bi-allelic mutation in the SMN1 gene and a clinical diagnosis of type 1 SMA. The list price of the product is £1.795 million per dose and it was made available on the NHS following a landmark deal struck with the manufacturer, Novartis Gene Therapies, in March 2021. The first child was treated in May 2021.

Untreated SMA is the leading genetic cause of death for children and, until 2019, there were no treatment options available for individuals diagnosed with the disease. With this gene therapy, babies can potentially develop to sit, crawl and walk. Nusinersen (2019) and risdiplam (2021) have also been recommended by NICE.

The HSCT ran a process to select four expert centres to deliver the treatment:

- Evelina London Children's Hospital (part of Guy's and St Thomas' NHS Foundation Trust)
- Manchester University NHS Foundation Trust
- Sheffield Children's NHS Foundation Trust
- University Hospitals Bristol and Weston NHS Foundation Trust.

These centres were selected because they were able to demonstrate that they had both expertise in treating children with SMA and the technical expertise and facilities to deliver the gene therapy. Each of the four centres contributes to a national multidisciplinary team (MDT) that confirms patient eligibility for treatment. Overall patient numbers are in line with those anticipated and include incident patients and patients who have previously been treated with other drugs.

3. Rare disease collaborative networks

Rare disease collaborative networks (RDCNs) are an important part of NHS England's approach to supporting the NHS to better care for patients with a rare disease. RDCNs are made up of groups of providers (rare disease collaborative centres [RDCCs]) that have an interest in developing understanding of a particular rare disease and are committed to working together to progress research, increase knowledge and improve patient experience.

To date, 13 RDCNs have been established across a range of specialties and disease groups, and in providers from a wide spread of regions. While the RDCNs are organised by NHS England, RDCCs can be from across the UK; see Appendix A.

RDCNs help to bring together experts working in a particular rare disease area, to improve understanding of the disease, raise awareness, standardise pathways and establishing best practice.

RDCNs can be a forum to discuss complex cases and to promote national MDT working, thereby ensuring the best outcomes for patients.

Some RDCNs are working towards developing their own disease databases or registries, which will advance disease surveillance and research.

There may also be advantages for providers in being part of an RDCN. This recognises their role as leaders in the field, which is likely to boost the organisation's reputation both nationally and internationally and, potentially, public interest or research funding.

4. Services and providers of highly specialised services for 2020/21

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.

The service provides an inpatient-based assessment for patients with AKU and subsequent annual review. 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	Liverpool University Hospitals NHS Foundation Trust
Expenditure	>£0.5 million to <£1 million
Patient caseload	125
Outcomes collated	• The median score for the general health domain of SF-36 for English and Scottish patients treated with nitisinone over the course of the reporting year. A higher SF-36 score indicates a better general health state with the maximum score being 100: 45 • The median AKUSSI (alkaptonuria severity score index) score for English and Scottish patients treated with nitisinone over the course of the reporting year. The AKUSSI is a composite score that measures systemic features of the disease that indicate morbidity. A higher score indicates more severe disease with a maximum score of over 200 : 83

	• Stability in both these scores indicates the treatment is arresting disease progression and its wider impact on general health.
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England regions.

Alström syndrome service (adults and children)

Alström syndrome is a rare complex genetic syndrome that usually presents with blindness in childhood. Patients go on to develop insulin-resistant diabetes, obesity, fibrosing cardiomyopathy (where growth of abnormal tissue 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 assess 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 Women's and Children's Hospital NHS Foundation Trust University Hospitals Birmingham NHS Foundation Trust
Expenditure	<£0.5 million
Patient caseload	71
Outcomes collated	Glycated haemoglobin (HbA1c) is measured primarily to identify the 3-month average plasma glucose concentration and is used to monitor diabetes control. The thresholds for the adult and child HbA1c are set at different levels due to the progressive nature of extreme insulin resistance. This also links into the higher rates of obesity in the adult cohort. • Proportion of children with HbA1c <48 mmol/mol: - Birmingham Women's and Children's: 86% • Proportion of adults with HbA1c <75 mmol/mol: - University Hospitals Birmingham: 50%* • Median age at death of patients on active caseload: - University Hospitals Birmingham: 28

	• Proportion of patients with a body mass index (BMI) <35: - Birmingham Women's and Children's: 92% - University Hospitals Birmingham: 77.3% * Note: provider has confirmed that adults were not able to attend in-person clinics where this test would have been performed – so the numerator was lower for this year
Geographical equity access	The number of patients accessing the service from NHS England 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 and causes 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 patients with AT. 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 progress.

NHS centre	Royal Papworth Hospital NHS Foundation Trust
Expenditure	<£0.5 million
Patient caseload	92
Outcomes collated	• Median BMI: 20.8 interquartile range 18.4–25.9 kg/m^2 (an important measure because patients with the condition often do not achieve optimum BMIs) • Median age at death of patients on active caseload: 24
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England regions

Ataxia telangiectasia services for children

Ataxia telangiectasia (AT) is a rare, neurodegenerative and progressive condition that starts in early childhood and causes 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 2 days with a MDT of experts. Previously unrecognised morbidity is often picked up at these reviews. This can include musculoskeletal and orthopaedic problems, immunological compromise and infections. Some of these can be treated and some require ongoing monitoring. Patients can have treatment initiated in the AT clinic, through local services or both.

Following the review, a management plan for local care providers is agreed and communicated to allow the local healthcare professionals to implement the recommendations and monitor progress.

NHS centre	Nottingham University Hospitals NHS Trust
Expenditure	<£0.5 million
Patient caseload	144
Outcomes collated	• Proportion of patients with previously unrecognised treatable or untreatable morbidity: 76% • Proportion of patients for whom active intervention was undertaken in clinic or arranged locally: 92% • Median quality of life (QOL) score at transition (using PedsQL Version 4): Data not submitted as provider has switched to a new QOL measure. This will be captured in future reporting
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England 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). 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. aHUS is characterised by microangiopathic haemolytic anaemia, thrombocytopaenia (low platelets) and acute kidney injury. Prior to the introduction of the drug eculizumab, most patients would progress to end stage renal failure, recurrent disease and extra-renal manifestations.

The aim of the national service is to provide rapid diagnostic and management advice for patients with aHUS. It offers comprehensive diagnostic, clinical and pathological investigations and expert opinion, facilitating optimal patient management (including access to eculizumab) 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
Number of patients being actively managed on eculizumab	132
Outcomes collated	• Number of deaths in patients with a diagnosis of complement mediated aHUS: no patient in England died of aHUS in 2020/21
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England 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 5 years with no functional hearing as a result of congenital abnormalities affecting the auditory nerves or the cochleae. This 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	If the number of surgical operations is <5, the data has been suppressed to maintain patient confidentiality
Outcomes collated	The Categories of Auditory Performance (CAP) score is a profile made up of several performance categories relating to auditory perception. It is a tool for monitoring long-term progress in the development of listening skills. • Mean length of stay in hospital (days): - Guy's and St Thomas': 6 - Manchester University: data suppressed to maintain patient confidentiality • Proportion of patients with improved soundfield hearing at 24 months: - Guy's and St Thomas': 100%

	- Manchester University: data suppressed to maintain patient confidentiality • Proportion of patients with improved CAP score at 24 months: - Guy's and St Thomas': 50% - Manchester University: 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 that can be undertaken in adult patients with short bowel syndromes who are on parenteral nutrition. Patients have insufficient bowel to take in enough food by mouth for 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 no longer require, or have a reduced requirement for, HPN. This is an established procedure in children.

NHS centre	Northern Care Alliance (previously Salford Royal NHS Foundation Trust)
Expenditure	<£0.5 million
Patient caseload	No new cases in 2020/21
Outcomes collated	• Proportion of patients not needing parenteral nutrition at 24 months post operation: 100% • Proportion of patients alive 1-year post operation: 100%
Geographical equity access	Numbers too small to analyse

Bardet-Biedl syndrome service (adults and children)

Bardet-Biedl syndrome is a highly debilitating autosomal-recessive genetic disorder that causes early-onset blindness, renal failure, obesity, diabetes, Hirschsprung disease (a type of intestinal blockage), urological problems and neurological deficits. About 1 in 100,000 babies are born with Bardet-Biedl syndrome, ie 5 or 6 each year in England.

Both the adult and paediatric services run dedicated clinics that assess all patients in a multidisciplinary structure. Patients are assessed and reviewed by all the specialties appropriate to their needs during the clinic. Early, tailored intervention and management

provide significant benefits for this patient group. The clinical outcomes measure that benefit, are routine measurements and can be responded to in a timely manner.

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 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
Patient assessments	83
Outcomes collated	• Proportion of children with HbA1c <48 mmol/mol: - Birmingham Women's and Children's: 94% - Great Ormond Street Hospital: 100% • Proportion of adult patients with HbA1c <75 mmol/mol: - Guy's and St Thomas': 69% - University Hospitals Birmingham: 79% • Proportion of adult patients with a BMI <35: - Guy's and St Thomas': 59% - University Hospitals Birmingham: 44% Glycated haemoglobin (HbA1c) is measured primarily to identify the 3-month average plasma glucose concentration and is used to monitor diabetes control. The thresholds for the adult and child HbA1c are set at different levels due to the progressive nature of the condition. This also links into the rates of obesity in the adult cohort.
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England regions

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. About 30 people in England have Barth syndrome.

When undiagnosed or treated by non-specialists, patients typically experience frequent hospital admissions for a range of diagnostic tests and treatment of severe and potentially life-threatening infections. The frequency and severity of these infections are significantly reduced through protocol-driven prescription of granulocyte colony stimulating factor (G-CSF). 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 MDT 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 and Weston NHS Foundation Trust (previously University Hospitals Bristol NHS Foundation Trust)
Expenditure	>£0.5 million but <£1 million
Patient caseload	27
Outcomes collated	• Median age at diagnosis: data suppressed to maintain patient confidentiality • Median age at death: No deaths • Proportion of hospital admissions for bacterial infections for patients on G-CSF: 15% • Number of hospital admissions for bacterial infections for patients on G-CSF: 20
Geographical equity access	The number of patients accessing the service from NHS England 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, and is characterised by an increased risk of childhood cancer and certain congenital features. One congenital feature is macroglossia (significant enlargement of the tongue), which causes: drooling; feeding, speech, orthodontic and dental problems; and significant psychosocial consequences. About 1 in 15,000 babies are born with Beckwith-Wiedemann syndrome (about 15–20 babies each year), but only about half have macroglossia.

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

NHS centre	Great Ormond Street Hospital for Children NHS Foundation Trust
Expenditure	<£0.5 million
Patient caseload	239
Outcomes collated	• Proportion of patients achieving improvement at the 3–6 month postoperative assessment of resting tongue position: 100% • Proportion of patients achieving improvement at the 3–6 month postoperative assessment in the reduction or cessation of drooling: 100% • Proportion of patients achieving improvement at the 3–6 month postoperative assessment in the reduction or elimination of macroglossia-related errors: 100% • Proportion of patients achieving improvement at the 3–6 month postoperative assessment in the reduction or elimination of oral stage difficulties related to the macroglossia: 100% • Proportion of patients achieving improvement at the 3–6 month postoperative 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 NHS England 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. Symptoms typically come and go in attacks called ‘flares’, which can be debilitating. The aim of the service is to ensure that patients of all ages 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
Patient caseload	2,044

Outcomes collated	• Median number of flares per patient during the previous 12 months: - Barts Health: 0 - Liverpool University: 0 - Sandwell and West Birmingham: 0
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England 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. With expert management and appropriate surgical reconstruction, a child with bladder exstrophy can have a near normal lifestyle. The goals of exstrophy reconstruction are:

- anatomical 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 with bladder exstrophy, ie around 20 babies each year in England.

The service is provided by a MDT 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	18
Outcomes collated	• Proportion of patients who achieved closure without dehiscence (wound dehiscence is a surgical complication in which a wound ruptures along a surgical incision): - Great Ormond Street: 100% - Manchester University: 100%

	• Proportion of patients' continent (dry by day) at age 5 years: - Great Ormond Street: 66% - Manchester University: 57% Note: For Great Ormond Street, this is the percentage of children who turned 5 in the financial year 2020/21 and were continent (dry by day) with no augmentation/catheterisation to empty their bladder. For Manchester University, this is 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 their bladder.
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England 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 regimen 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. The scope of referrals has broadened in recent years to accept referrals for patient with severe and persistent pain due to the consequences of cancer treatment from any tumour site.

NHS centre	Royal National Hospital for Rheumatic Diseases – Royal United Hospitals Bath NHS Foundation Trust
Expenditure	<£0.5 million
Patient caseload	27
Outcomes collated	• Proportion of patients who achieved improvement in mood: 100% Note: Due to the broadening of referrals, the service is moving to the use of a new set of outcome measures that better reflect the patient's needs and the delivery of the service. These will be published in full in the next report.

Geographical equity access	The number of patients accessing the service from NHS England 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 with services for heart failure, cystic fibrosis/respiratory medicine and pulmonary hypertension, and 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 (NHSBT). 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	28
Outcomes collated	Outcome measures for this service are provided through NHSBT routine statistical monitoring. Reporting covers the period 1 April 2011 to 31 March 2021. • 30-day unadjusted patient survival rate after first paediatric heart-only transplant: - Great Ormond Street: 100% - Newcastle upon Tyne: 93% • 1-year patient survival rate after first paediatric heart-only transplant: - Great Ormond Street: 100% - Newcastle upon Tyne: 94.2% • 5-year patient survival after first paediatric heart-only transplant: - Great Ormond Street: 84.2%

	- Newcastle upon Tyne: 80.2% • 90-day patient survival rate after first paediatric lung-only transplant: - Great Ormond Street: 94% - Newcastle upon Tyne: Data suppressed to maintain patient confidentiality • 1-year unadjusted patient survival rate after first paediatric lung-only transplant: - Great Ormond Street: 89% - Newcastle upon Tyne: Data suppressed to maintain patient confidentiality • 5-year unadjusted patient survival after first paediatric lung-only transplant: - Great Ormond Street: 77% - Newcastle upon Tyne: Data suppressed to maintain patient confidentiality
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:

- Hydatidiform mole (also known as molar pregnancy): in this condition, the sperm and egg fuse but a healthy fetus does not develop. The placenta grows to an abnormal size, requiring surgical evacuation of the uterus.
- Choriocarcinoma: 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 can 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	495
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 NHS England 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* following a lung insult (typically sarcoidosis, atypical tuberculosis or recurrent pneumothoraces) and occurs in those with one or more innate genetic defects. CPA progresses without treatment.

This 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 the specification are offered clinically appropriate treatment options. The two main aims of therapy are infection control and prevention of radiological progression.

NHS centre	Manchester University NHS Foundation Trust
Expenditure	>£5 million but <£10 million
Patient caseload	418
Outcomes collated	The effect of treatment can be accurately assessed using CT thorax and apergillus IgG, an immunological marker of infection burden. A high proportion indicates successful intervention. • Proportion of patients showing stability or improvement in CT thorax and improvement in aspergillus IgG: 75%
Geographical equity access	The number of patients accessing the service from NHS England 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 significantly: one person can have just a few and another 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 children 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 and Weston NHS Foundation Trust (previously University Hospitals Bristol NHS Foundation Trust)
Expenditure	>£1 million but <£5 million
Patient caseload	315
Outcomes collated	• Median number of new non-vertebral fractures: - Birmingham Women’s and Children’s: 1 - Great Ormond Street: 0 - Sheffield Children’s: 0 - University Hospitals Bristol and Weston: 0 • Median number of new vertebral fractures: - Birmingham Women’s and Children’s: 0 - Great Ormond Street: 0 - Sheffield Children’s: 0 - University Hospitals Bristol and Weston: 0 • Proportion of patients with scoliosis and Cobb angle >45 degrees (the Cobb angle measures the degree of abnormal lateral spinal curvature): - Birmingham Women’s and Children’s: 8% - Great Ormond Street: 6% - Sheffield Children’s: 3% - University Hospitals Bristol: 0%
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England regions

Complex Ehlers-Danlos syndrome service (adults and children)

Ehlers-Danlos syndrome (EDS) is a group of inherited 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 type 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 straightforward 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
Number of patients with a definitive diagnosis	92
Outcomes collated	• Proportion of patients with a definitive diagnosis or diagnosis ruled out: - London North West University: 100% - Sheffield Children's: 95% • Proportion of patients with a genetic diagnosis: - London North West University: 44% - Sheffield Children's: 44%
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England 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, some of whom have a high risk of developing rare complications, which may affect most body systems, causing significant morbidity and mortality. Complex NF1 is defined by the presence of these complications, 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 MDT (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, eg cervical cord compression.

NHS centres	Guy's and St Thomas' NHS Foundation Trust Manchester University NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Outpatient attendances	1,205
Outcomes collated	• Total number of appropriate interventions facilitated: - Guy's and St Thomas': 34 - Manchester University: 30 • Total number of interventions avoided: - Guy's and St Thomas': 14 - Manchester University: 4 • Mean age at death in the previous 10 years of patients with NFI: - Guy's and St Thomas': 40 - Manchester University: 44 Interventions include neurosurgery, vascular surgery, chemotherapy, radiotherapy, orthopaedic surgery, craniofacial surgery, endocrine surgery, additional biopsies and imaging
Geographical equity access	The number of patients accessing the service from NHS England 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	38
Outcomes collated	• 1-year survival: 100%
Geographical equity access	The number of patients accessing the service from NHS England 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 characterised by excess insulin production, resulting in hypoglycaemia. The clinical presentation and progress of CHI vary across a spectrum from those with transient hypoglycaemia to those unresponsive to medical treatment and requiring pancreatectomy. In the absence of expert management, children may show developmental delay as a result of brain injury from prolonged or recurrent hyperinsulinaemic hypoglycaemia in infancy.

The service diagnoses patients (usually in the newborn 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 works with a surgical team to manage those children whose condition and response to medical management indicate 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
Patient caseload	1,297
Outcomes collated	• Deaths in patients with CHI as a consequence of CHI: - Great Ormond Street: 0 - NORCHI: 0 • Unplanned admissions due to CHI and admitted under the paediatric endocrine team, day cases excluded: - Great Ormond Street: 22 - NORCHI: 12

Geographical equity access	The number of patients accessing the service from NHS England 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	465
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: 0% • Proportion of patients with level 5 surgical complications: - Alder Hey Children's: 1% - 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 NHS England 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, 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	161
Outcomes collated	• Median 20-point CAPS activity score (a low CAPS score indicates symptom control): 3/20
Geographical equity access	The number of patients accessing the service from NHS England 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 and 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.

NHS centre	Royal Free London NHS Foundation Trust
Expenditure	>£5 million but <£10 million
First patient evaluations	1,106
Outcomes collated	• Proportion of patients with a definitive diagnosis or diagnosis ruled out: 93% • Proportion of patients with a genetic diagnosis: 12% Note: Amyloidosis can be genetic or acquired
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England regions

Diagnostic service for primary ciliary dyskinesia (adults and children)

Primary ciliary dyskinesia (PCD) is a genetic disorder of the air tubes of the lungs (the bronchi), which become infected and filled with pus due to abnormalities of the hair-like structure (cilia) of the cells lining the respiratory tract. This can lead to repeated infections and damage the lung, especially if 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 patients 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	44
Outcomes collated	• Paediatric outcomes collected by the paediatric PCD management service: • Adult outcomes will be collected by the adult PCD management service, a newly commissioned service that will report its first set of measures in 2022
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England regions

Diagnostic service for rare neuromuscular disorders (adults and children)

This service provides a diagnostic, advisory and clinical service for patients with four groups of very rare inherited neuromuscular disorders:

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

The aim of the service is to make a precise molecular or clinical diagnosis, and to assess fully the extent of their disease. Definitive diagnosis is made by identifying the primary gene defect. Each disease group involves multiple genes, and a disease-specific battery of techniques is used to decide which gene to search first for DNA mutations. These techniques may include detailed clinical assessments, specialist neurophysiological tests and immunological analyses of 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 patient referrals	1,291
Outcomes collated	• Proportion of patients with a genetic diagnosis: - Congenital muscular dystrophies – Great Ormond Street Hospital: 37% - Congenital myasthenic syndromes – Oxford University: 81% - Limb girdle muscular dystrophies – Newcastle upon Tyne: 54% - Muscle channelopathies – University College London: 61% Note: Centres diagnose and assess different conditions, so outcome measures are not comparable
Geographical equity access	The number of patients accessing the service from NHS England 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 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, there is potential to significantly improve health and quality of life through a comprehensive, expert patient focused 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.

Many with CS/TTD require photoprotection, have reduced mobility or are on supplemental feeds. These circumstances can impact their ability to maintain optimal vitamin D levels. Therefore, regular vitamin D monitoring is important. Since its establishment the service has been working to improve rates of measurement in partnership with local services.

NHS centres	Guy's and St Thomas' NHS Foundation Trust
Expenditure	>£1 million but <£5 million

Patient caseload	108
Outcomes collated	• Proportion of patients with a newly confirmed diagnosis of XP being offered support within 10 working days of referral: 100% • Proportion of patients living in a UV-safe home environment as defined by a safe reading on the UV meter: 85% • Proportion of patients with CS/TTD who have vitamin D levels measured: 32% • Proportion of patients with a confirmed CS/TTD diagnosis referred to the service who are offered support including home-visiting and bespoke care passport: 100%
Geographical equity access	Numbers insufficient for robust analysis due to being a newly commissioned service.

Encapsulating peritoneal sclerosis treatment service (adults)

Encapsulating peritoneal sclerosis (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 subacute 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 associated with poor outcomes if not recognised early and treated. By centralising treatment in specified national centres, experience has been consolidated, leading to patient outcomes that now mirror 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	4
Outcomes collated	• 1-year survival rate for patient's post operation for all cases (renal and non-renal): - Cambridge University Hospitals: 50% - Manchester University: 20%* • Proportion of patients TPN-free post operation: - Cambridge University Hospitals: 50% - Manchester University: 100%

	* The trust has reviewed this survival rate, and found a number of factors unrelated to EPS or the operation contributed to the mortality rate
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England regions

Epidermolysis bullosa service (adults and children)

Epidermolysis bullosa (EB) is 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 EB present.

The national EB service aims to provide diagnosis and assessment of infants, children, adolescents and adults with suspected or known EB, supporting (including long term) symptom relief and prevention of complications.

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
Patient caseload	516 severe patients 1194 mild patients

Outcomes collated	• Median quality of life score at transition (QoLEB): - Birmingham Women's and Children's: 29 - Guy's and St Thomas': 20 - University Hospitals Birmingham: 26 • Proportion of unplanned admissions among patients with recessive dystrophic EB: - Birmingham Women's and Children's and Heart of England combined: 26% - Great Ormond Street: 63%* - Guy's and St Thomas': 10% - University Hospitals Birmingham: 8% Note: Recessive dystrophic EB is the most severe type of EB * 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 NHS England 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)
Number of patients starting treatment	606
Outcomes collated	• Proportion of patients survived at discharge:

	- Guy's and St Thomas': 77% - Manchester University: 53% - Royal Brompton & Harefield: 74% - Royal Papworth: 69%* - Leicester: 59%% * 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 NHS England 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
Number of patients starting treatment	54
Outcomes collated	• Proportion of patients survived at discharge (neonatal): - Alder Hey Children's: 50%* - Birmingham Women's and Children's: 91% - Great Ormond Street: 89% - Newcastle upon Tyne: no patients - Leicester: 92% • Proportion of patients survived at discharge (children): - Alder Hey Children's: 67%* - Birmingham Women's and Children's: 86% - Great Ormond Street: 67%*

	- Newcastle upon Tyne: 50%* - Leicester: 100% * 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 NHS England regions

Ex-vivo partial nephrectomy service (adults)

The aim of the service is to provide a potential cancer cure for patients with complex renal tumours in solitary kidneys or bilateral disease who are not suitable for conventional treatments, as well avoid their need for 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	6
Outcomes collated	• Proportion of patients 1-year survival post operation: 100% • Proportion of patient's dialysis-free 1-year post-operation: 50%
Geographical equity access	Numbers too small to analyse

Hand and upper limb transplantation service (adults)

Hand and upper limb transplantation is possible with cadaveric donation. The surgery 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 patient referrals	The number of transplants is <5, so the data has been suppressed to maintain patient confidentiality

Outcomes collated	• Number of patients with improvement in Canadian Occupational Performance Measure (COPM) score of >1: Data suppressed to maintain patient confidentiality • Number of patients showing psychological improvement: Data suppressed to maintain patient confidentiality • Number of patients with survived transplanted limb at 1 year: Data suppressed to maintain patient confidentiality • Number of patients with survived transplanted limb at 5 years: Data suppressed to maintain patient confidentiality
Geographical equity access	Numbers too small to analyse

Heart transplantation service (adults)

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

NHS centres	Manchester University NHS Foundation Trust Guy's and St Thomas' NHS Foundation Trust (previously 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	109
Outcomes collated	Reporting covers the period 1 April 2011 to 31 March 2021 • 30-day risk-adjusted patient survival rates after first adult DBD (donor after brain death) heart transplant: - Guy's and St Thomas': 88.7% - Manchester University: 95.6% - Royal Papworth: 98.8% - University Hospitals Birmingham: 93.1% - Newcastle upon Tyne: 93.6% • 1-year risk-adjusted patient survival rates after first adult DBD heart transplant:

	- Guy's and St Thomas': 80.8% - Manchester University: 88.9% - Royal Papworth: 85.6% - University Hospitals Birmingham: 86.1% - Newcastle upon Tyne: 84.3% • 5-year risk-adjusted patient survival rates from listing for first DBD heart transplant: - Guy's and St Thomas': 69.8% - Manchester University: 66% - Royal Papworth: 79.1% - University Hospitals Birmingham: 63.3% - Newcastle upon Tyne: 66.4%
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
Patient caseload	Data suppressed to maintain patient confidentiality
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 Teaching: 0 - 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: 0 - Sheffield Teaching: 0 - 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 patient admissions	Data suppressed to maintain patient confidentiality
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 affect the liver and 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 patient caseload	222
Outcomes collated	• Proportion of patients with specific diagnosis: 81% • Proportion of patients with diabetes maintaining HbA1c below 75 mmol/mol: 74% Note: Glycated haemoglobin (HbA1c) is measured primarily to identify the 3-month average plasma glucose concentration. The service is aiming to keep as many patients as possible below the HbA1c threshold – indicating good diabetes control
Geographical equity access	The number of patients accessing the service from NHS England 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	9
Outcomes collated	• Median number of severe hypoglycaemic events between registration and transplant: 13 • Median number of severe hypoglycaemic events at 1 year post transplant: 0 • Median HbA1c before transplant (for routine transplants between 1 April 2016 and 31 March 2021): 64 mmol/mol • Median HbA1c 1 year post-transplant (for routine transplants between 1 April 2016 and 31 March 2021): 48 mmol/mol Note: Glycated haemoglobin (HbA1c) is measured primarily to identify the 3-month average plasma glucose concentration Some data is presented in aggregate over several years due to small patient numbers
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England 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	599
Outcomes collated	Reporting covers the period 1 April 2011 to 31 March 2021

	• 1-year risk-adjusted patient survival for adult elective deceased donor first liver transplants: - Cambridge University: 96% - King's College: 96.5% - Leeds Teaching: 90.9% - Royal Free: 94.3% - Newcastle upon Tyne: 89.3% - University Hospitals Birmingham: 93.5% • 5-year risk-adjusted patient survival for adult elective deceased donor first liver transplants: - Cambridge University: 87.5% - King's College: 84.6% - Leeds Teaching: 86.4% - Royal Free: 86.2% - Newcastle upon Tyne: 73.6% - University Hospitals Birmingham: 81.6% Note: NHSBT in conjunction with NHS England has 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 NHS England 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 Women’s and Children's Hospital NHS Foundation Trust King's College Hospital NHS Foundation Trust Leeds Teaching Hospitals NHS Trust
Expenditure	>£50 million (adults and children)
Number of transplants	92
Outcomes collated	Reporting covers the period 1 April 2011 to 31 March 2021

	• 1-year unadjusted patient survival for paediatric elective deceased donor first liver transplants: - Birmingham Women's and Children: 90% - King's College: 97.2% - Leeds Teaching: 98.1% • 5-year unadjusted patient survival for paediatric elective deceased donor first liver transplants: - Birmingham Women's and Children's: 93.6% - King's College: 92.5% - Leeds Teaching: 100% Note: NHSBT in conjunction with NHS England 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 NHS England 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 Guy's and St Thomas' NHS Foundation Trust (previously 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	78
Outcomes collated	Reporting covers the period 1 April 2011 to 31 March 2021 • 90-day patient survival rate after first adult lung transplant: - Guy's and St Thomas': 88.6% - Manchester University: 97.4% - Royal Papworth: 90.5% - Newcastle upon Tyne: 86.5% - Birmingham: 86%

	• 1-year patient survival rate after first adult lung transplant:
	  - Guy's and St Thomas': 81.2%
	  - Manchester University: 86.7%
	  - Royal Papworth: 80.9%
	  - Newcastle upon Tyne: 79.6%
	  - Birmingham: 76.4%
	• 5-year patient survival rate from listing for first lung-only transplant:
	  - Guy's and St Thomas': 62.8%
	  - Manchester University: 57.2%
	  - Royal Papworth: 60.7%
	  - Newcastle upon Tyne: 57.2%
	  - Birmingham: 31%
	Note: NHSBT in conjunction with NHS England has 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 NHS England 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
Patient caseload	225
Outcomes collated	• Proportion of patients having a pneumothorax: 0% • Proportion of patients having a renal angiolipoma bleed: 0% • Proportion of patients having an FEV_1 decline of >150 mL per annum: 1%

Geographical equity access	The expected numbers of patients are accessing the service from all NHS England regions

Lysosomal storage disorders service (children and adults)

Lysosomal storage disorders (LSDs) are rare genetic disorders characterised by specific lysosomal enzyme deficiencies. Abnormal depositions build up in different organs (including the brain, skeleton, heart, liver, kidneys and spleen) and lead to the functional deterioration of these body systems.

Some LSDs can be treated with enzyme replacement therapies (ERTs), substrate reduction therapy (SRT) or other disease modifying drugs. However, there is no treatment for many LSDs.

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 Northern Care Alliance (previously Salford Royal NHS Foundation Trust) Royal Free London NHS Foundation Trust University College London Hospitals NHS Foundation Trust University Hospitals Birmingham NHS Foundation Trust
Expenditure	>£50 million
Active patient caseload	2,623
Outcomes collated	• Proportion of patients with the LSD Fabry disease, having a cardiac device implanted 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% - Northern Care Alliance: 3% - Royal Free: 3% - University College London:0% - University Hospitals Birmingham: 3%

- Proportion of patients with the LSD Fabry disease 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%
  - Northern Care Alliance: 0%
  - Royal Free: 0%
  - University College London: 2%
  - University Hospitals Birmingham: 0%
- Proportion of patients with the LSD Gaucher disease 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%
  - Northern Care Alliance: 0%
  - Royal Free: 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: 0%
  - Great Ormond Street: 4%
  - Manchester University: 0%
  - Northern Care Alliance: 0%
  - Royal Free: 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%
  - Northern Care Alliance: 1%
  - Royal Free: 0%
  - University College London: 0%

	- University Hospitals Birmingham: 0%
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England regions

McArdle's disease service

McArdle's disease is caused by an inborn deficiency of muscle phosphorylase that results in 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 accurate diagnosis and outpatient management of the condition.

NHS centre	University College London Hospitals NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Patient caseload	328
Outcomes collated	• Median functional capacity – 12MWD: 732 m • Number of patients requiring hospital assessment: 36 • Median quality of life (SF-36) score: physical functioning: 40 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 NHS England regions is different to what is expected. The potential reasons for this are being explored with the service

Multiple sclerosis management service for children

The Multiple Sclerosis (MS) management Service for children, is commissioned to provide a service for children with: MS; suspected MS; equally rare 'MS-like' recurrent acquired demyelinating syndromes; or who have had a first demyelination episode. There is currently no cure for MS, but treatments and specialist input can help control disease activity, decrease disability from the condition and reduce ongoing symptoms. Early recognition of this serious neuroinflammatory disorders is important to reduce long-term morbidity and neuro-disability, and for early intervention.

Patients with MS or 'MS-like' conditions are assessed and treated in age-appropriate outpatient and inpatient settings in specialist paediatric neurology centres, by professionals working in MDTs with expertise in MS.

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
Patient caseload	401
Outcomes collated	• Proportion of patients achieving a slowing in progression of disease using annualised relapse rate (ARR): - Alder Hey Children's: 50% - Birmingham Women's and Children's: 94% - Cambridge University: 57% - Great Ormond Street: Data suppressed to maintain patient confidentiality - Guy's and St Thomas': 67% - Manchester University: 40% - 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: - Alder Hey Children's: 50% - Birmingham Women's and Children's: 94% - Cambridge University: 57% - Great Ormond Street: Data suppressed to maintain patient confidentiality - Guy's and St Thomas': 86% - Manchester University: 83% - 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 and no MRI activity: - Alder Hey Children's: 50% - Birmingham Women's and Children's: 58% - Cambridge University: 57% - Great Ormond Street: Data suppressed to maintain patient confidentiality - Guy's and St Thomas': 27% - Manchester University: 74% - 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: 50% - Birmingham Women's and Children's: 58% - Cambridge University: 57% - Great Ormond Street: Data suppressed to maintain patient confidentiality - Guy's and St Thomas': 0% - Manchester University: 74% - Newcastle upon Tyne: 100% • 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: 67% - Birmingham Women's and Children's: 40% - Cambridge University: 50% - Great Ormond Street: 100% - Guy's and St Thomas': 40% - Manchester University: 20% - Newcastle upon Tyne: Data suppressed to maintain patient confidentiality 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-schwannomatosis* service (all ages)

* gene name

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

The service includes:

- outpatients: MDT outpatients and satellite outpatients
- mutation testing for NF2-schwannomatosis
- auditory brainstem implants and auditory implants
- vestibular schwannomas surgery
- stereotactic radiosurgery
- Hearing Link's NF2-schwannomatosis 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
Patient caseload	1,027
Outcomes collated	• Hearing preservation: Proportion of NF2-schwannomatosis patients diagnosed since inception of service maintaining useful hearing (target >80%): - Cambridge University: 92% - Guy's and St Thomas': 91% - Manchester University: 93% - Oxford University: 96% • Survival: Proportion of patients who have died prematurely from an NF2-schwannomatosis related complication (target <5%): - Cambridge University: 2%

	- Guy's and St Thomas': 0% - Manchester University: 1% - Oxford University: 0%
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England 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).Further severe attacks often follow and usually result in permanent disability (visual loss, limb weakness, respiratory muscle weakness). The condition is associated with high mortality and morbidity rates. 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 patient evaluations	137
Outcomes collated	• Median annualised relapse rate: - Oxford University: 0% - Walton Centre: 0%
Geographical equity access	The number of patients accessing the service from NHS England 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 are confirmed as having eye cancer (about 700 each year).

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
New patient positive patient assessments	611
Outcomes collated	• Proportion of patients experiencing primary enucleation among patients with melanoma: - Liverpool University: 13% - Moorfields: 33% - Sheffield Teaching: 24% • Proportion of patients experiencing secondary enucleation among patients with melanoma: - Liverpool University: 1% - Moorfields: 1% - Sheffield Teaching: 1% • Proportion of patients developing metastatic disease among patients with melanoma: - Liverpool University: 2% - Moorfields: 1% - Sheffield Teaching: 4% Enucleation (removal of the eyeball) is only undertaken when it is not possible to safely conserve vision and/or control pain, eg if the tumour is very large or involves the optic nerve. Enucleation rates will depend on tumour type and site, stage at presentation, feasible treatment options and patient choice.
Geographical equity access	The number of patients accessing the service from NHS England regions is different to what is expected. The potential reasons for this are being explored with the service.

Open fetal surgery to treat spina bifida

Spina bifida is a congenital condition where the spinal column and cord do not fully form in utero. 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 unborn babies, open fetal surgery (operating on the baby while still in the womb) can 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 Hospitals NHS Foundation Trust Universitair Ziekenhuisen, Leuven, Belgium
Expenditure	<£0.5 million
Number of operations	26
Outcomes collated	• Proportion of in utero deaths following surgery during the last 12 months: - University College London: 0% - Leuven Belgium: 0% • Proportion of babies with postnatal cerebrospinal fluid leakage: - University College London: 0% - Leuven Belgium: 0% • Proportion of babies with improvement in Chiari II malformation: - University College London: 100% - Leuven Belgium: 100%
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. It 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).

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
Annual cases reviewed	5,047
Outcomes collated	• Proportion of simple cases reported within 7 calendar days: - Liverpool University: 92% - Manchester University: 93% - Sheffield Teaching: 93% - University College London: 86% • Proportion of complex cases reported within 10 calendar days: - Liverpool University: 94% - Manchester University: 96% - Sheffield Teaching: 93% - University College London: 90% • Proportion of all cases reported within 21 calendar days: - Liverpool University: 99% - Manchester University: 100% - Sheffield Teaching: 100% - University College London: 99%
Geographical equity access	Data not available or not comparable

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

Osteo-odonto-keratoprosthesis (OOKP) is a specialist surgical intervention that can restore meaningful vision to patients with 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 for whom no other treatments would restore sight.

Patients are initially assessed by ophthalmic and maxillofacial consultants; involving examination of the eyes, teeth and mouth. OOKP is then a two-stage procedure: (1) extraction of the patient's tooth and bone, which are fashioned into a 'bolt' and placed in the eye to support a synthetic optical cylinder, and (2) the procedure is performed about four months after the first stage. Each surgical procedure lasts about 6 hours and patients require lifelong follow-up.

NHS centre	Brighton and Sussex University Hospitals NHS Trust
Expenditure	<£0.5 million
Number of patients with stage 2 surgery	Data suppressed to maintain patient confidentiality
Outcomes collated	• Proportion of patients with visual acuity 6/12 or better at 12 months post operation: 50%
Geographical equity access	The number of patients accessing the service from NHS England 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 and treatment for infants and children under 5 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 that in the absence of access to a specialised service, patients can be subjected to multiple unnecessary investigations that do not lead to a definitive diagnosis.

The service treats about 20 children each year.

NHS centre	Great Ormond Street Hospital for Children NHS Foundation Trust
Expenditure	>£1 million but <£5 million
Number of new patient referrals	Data suppressed to maintain patient confidentiality
Outcomes collated	• Proportion of patients admitted for Phase 1 admission within 8 weeks: 43%

	• Proportion of definitive diagnosis made and results fed back via MDT within 4 weeks of Phase 1 admission discharge: 71%
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England regions

Pancreas transplantation service (adults)

This service provides assessment, transplantation and lifelong follow-up for patients with diabetes who require 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 The Newcastle upon Tyne Hospitals NHS Foundation Trust Oxford University Hospitals NHS Trust
Expenditure	>£5 million but <£10 million
Number of transplants	66
Outcomes collated	• 1-year risk-adjusted patient survival for first simultaneous pancreas and kidney or islets (SPK) transplant from deceased donors: - Cambridge University: 100% - Guy's and St Thomas': 97% - Imperial College: 100% - Manchester University: 100% - Newcastle upon Tyne: 97% - Oxford University: 96% • 5-year risk-adjusted patient survival for first SPK transplant from deceased donors: - Cambridge University: 87% - Guy's and St Thomas': 86% - Imperial College: 86% - Manchester University: 86% - Newcastle upon Tyne: 86% - Oxford University: 86%%

	1-year survival includes transplants performed between 1 April 2016 and 31 March 2020 5-year survival includes transplants performed between 1 April 2012 and 31 March 2016
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England 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.

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

Outreach clinics are held in locations outside the centres.

NHS centres	King's College Hospital NHS Foundation Trust Leeds Teaching Hospitals NHS Trust
Expenditure	>£50 million
Patient caseload	923
Outcomes collated	• 5-year relative survival rate: - King’s College: 86% - Leeds Teaching: 87% • Median transfusions per patient in previous 12 months: - King’s College: 8 - Leeds Teaching: 6
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England regions

Primary ciliary dyskinesia management service (children and adults)

Primary ciliary dyskinesia (PCD) is a genetic condition in which the respiratory system cilia, hair-like microscopic cells, do not effectively clear mucus from the lungs, paranasal sinuses and ears. Recurring respiratory infections can lead to 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.

Some PCD management services are provided by highly specialist PCD 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 highly speciality PCD service.

The adult service started in 2019/20, with this service set up in the same centres that provide the paediatric service, but mobilisation was impacted by the pandemic response.

NHS centres	Leeds Teaching Hospitals NHS Trust Royal Brompton & Harefield NHS Foundation Trust University Hospitals of Leicester NHS Trust University Hospital Southampton NHS Foundation Trust
Expenditure	Data to be included in the next report
Patient caseload	448 – paediatric patients
Outcomes collated	• Proportion of patients in the PCD management service offered an annual review appointment (consisting of the processes listed in the service specification): - Leeds Teaching: Children 100% - Royal Brompton & Harefield: Children 93% - Southampton: Children 100% - Leicester: Children 96% • Proportion of patients seen by a physiotherapist at annual review: - Leeds Teaching: Children 99% - Royal Brompton & Harefield: Children 100% - Southampton: Children 100% - Leicester: Children 87% • Proportion of patients seen by a nurse specialist at annual review: - Leeds Teaching: Children 87% - Royal Brompton & Harefield: Children 100% - Southampton: Children 100% - Leicester: Children 87%

	• Proportion of patients seen by an ENT specialist at annual review: - Leeds Teaching: Children 12%* - Royal Brompton & Harefield: No data submitted** - Southampton: Children 100% - Leicester: Children 82% * All outcome measures are discussed at the annual clinical meeting including reasons for apparent variation Only paediatric outcomes have been reported as the establishment of the adult service was significantly disrupted by COVID-19. Adult outcome measures will be presented in the next report * Due to COVID-related staff redeployment, sickness and restrictions, patients were seen by ENT in the community and this data is not available. Outpatient data for this centre will be presented in the next report
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England regions

Primary malignant bone tumours service (adults and adolescents)

This service provides diagnosis and surgery for primary malignant bone tumours (PMBTs), eg osteosarcoma, chondrosarcoma and Ewing's sarcoma. The aim is to avoid amputation if possible while ensuring complete removal of the cancer.

The service receives about 1,000 referrals of suspected PMBT each year.

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	446
Outcomes collated	• Proportion of 3-year local recurrence among patients having limb salvage: - Oxford University: 15%

	- Robert Jones and Agnes Hunt: 5% - Royal Orthopaedic: 13% - Royal National Orthopaedic: 7% - Newcastle upon Tyne: 0% • Proportion of limbs salvaged: - Oxford University: 97% - Robert Jones and Agnes Hunt: 88% - Royal Orthopaedic: 81% - Royal National Orthopaedic: 0% Data not submitted* - Newcastle upon Tyne: 54% • Proportion of patients with 3-year prosthesis infection/ loosening: - Oxford University: 3% - Robert Jones and Agnes Hunt: 13% - Royal Orthopaedic: 15.2% - Royal National Orthopaedic: 6% - Newcastle:0% * The provider was unable to review records for the appropriate patient cohort. NHS England is working with this provider and the service as a whole to review the outcome measures and ensure consistent data collection 3-year period refers to patients treated in the 3 years preceding the data collection (2018–2021)
Geographical equity access	The number of patients accessing the service from NHS England 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. There is almost no radiation exposure for 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, reduces morbidity arising from treatment, and supports the patient and family throughout their cancer diagnosis, treatment and follow-up.

The PBT service has contributed significantly to the development of the international evidence base through peer reviewed publications, and posters and presentations at international conferences.

Centres	The Christie NHS Foundation Trust Westdeutsches Protonentherapiezentrum (WPE) Essen, Germany
Expenditure	Christie: >£20 million but <£30 million Overseas providers: <£0.5 million
Number of patients referred and treated	Christie: 300 (treatment completed 234 – adult 89, teen/young adult 58, paeds 87) Overseas: 7
Details of referrals	• Number of adults referred:114 • Number of adults planned: 98 • Number of adults treated: 89 • Number of teenagers and young adults referred: 54 • Number of teenagers and young adults planned: 43 • Number of teenagers and young adults treated: 46 • Number of children referred: 132 • Number of children planned: 107 • Number of children treated: 109 NHS England is working with the PBT providers to develop a set of outcome measures for the service
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England regions

Pseudomyxoma peritonei service (adults)

Pseudomyxoma peritonei (PMP) is a rare, mucus-producing tumour, which spreads slowly and locally to compress the abdominal organs. PMP usually arises from a ruptured tumour of the appendix. It is of borderline malignancy in that it does not metastasise via bloodstream or lymphatic system in the early stages. However, without specialist cancer treatment, most patients die 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, which affects their ability to eat normally.

Treatment options include:

- cytoreduction with HIPEC (hyperthermic intraperitoneal chemotherapy)
- debulking of the tumour

- 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
Number of patients requiring major full cytoreduction	232
Outcomes collated	• Proportion of 5-year patient survival – all operative cases: - Christie: 70 - Hampshire: 72% • Proportion of 5-year patient survival – complete cytoreduction: - Christie: 79% - Hampshire: 80%
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England regions

Pulmonary hypertension service for children

Paediatric pulmonary hypertension (PH) is high blood pressure in the artery carrying blood from the heart to 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 MDT. Diagnostics include cardiac catheterisation, invasive radiology, echocardiography, non-invasive imaging (CT scanning, MRI), 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
Patient caseload	552

Outcomes collated	• Proportion of patients 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: 11% • Proportion of children receiving epoprostenol who experienced a line-related bloodstream infection: 0%
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England 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). Clots 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 chronic thromboembolic pulmonary hypertension (CTEPH) are referred for consideration of surgery.

NHS centre	Royal Papworth Hospital NHS Foundation Trust
Expenditure	>£5 million but <£10 million
Surgical operations	111
Outcomes collated	• 90-day patient survival: 97% • 3-year patient survival: 90% • In-hospital mortality: 4%
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England regions

Rare mitochondrial disorders service (adults and children)

Mitochondria are small organelles present in every cell in the body that have their own genetic complement, the mitochondrial genome. Their principal task is to provide energy for normal cell functioning and maintenance. Disruption of this energy supply can have devastating effects on 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 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
- multidisciplinary 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 diagnosis	476
Outcomes collated	• Proportion of patients given an alert card:* - Oxford University: 100% - Newcastle upon Tyne: 92% - University College London: 70% * 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 NHS England regions

Retinoblastoma service (children)

Retinoblastoma is a malignant tumour of the retina and usually presents in children under the age of 2. It is an aggressive eye cancer that can result in the loss of vision and, in extreme cases, death.

The treatment modalities are:

- 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	43
Outcomes collated	• Proportion of patient 5-year survival covering the period 2017 to 2021: - Barts Health: 96% - Birmingham Women's and Children's: 100% • Proportion of patients with primary enucleation: - Barts Health: 44% - Birmingham Women's and Children's: 46%
Geographical equity access	The number of patients accessing the service from NHS England 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 porphyria is 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 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 year with acute porphyria who meet the definition of 'severe' disease.

NHS centres	King's College Hospital NHS Foundation Trust Cardiff and Vale University Health Board (previously University Hospital of Wales)
Expenditure	>£0.5 million but <£1 million
Active patient caseload	127
Outcomes collated	• Mortality rate: - King’s College: 0% - Cardiff: 0% • Proportion of patients having 4 or more hospital admissions (porphyria-related) in the previous 12 months: - King’s College: 4% - Cardiff: 5%
Geographical equity access	The expected numbers of patients are accessing the service from all NHS England 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, T and B lymphocyte functions are both defective from birth.

Treatment is usually 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	52
Outcomes collated	• Proportion of patients alive at 2 years post treatment:

	- Great Ormond Street: 89% - Newcastle upon Tyne: 85%
Geographical equity access	The number of patients accessing the service from NHS England 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. The Cambridge service provides multi-visceral transplantation and the Oxford service intestinal only 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	5
Outcomes collated	Reporting covers the period 1 April 2011 to 31 March 2021 • Proportion of unadjusted 90-day patient survival for first intestine transplants: - Cambridge University: 100% (not including liver) - Cambridge University: 88.2% (including liver) - Oxford University: 90.9% (not including liver) • Proportion of unadjusted 1-year patient survival for first intestine transplants: - Cambridge University: 87.8% (not including liver) - Cambridge University: 70.3% (including liver) - Oxford University: 88.5% (not including liver) • Proportion of unadjusted 5-year patient survival for first intestine transplants: - Cambridge University: 79.8% (not including liver) - Cambridge University: 39.6% (including liver) - Oxford University: 69% (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 NHS England 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	5
Outcomes collated	Reporting covers the period 1 April 2011 to 31 March 2021 • Proportion of unadjusted 90-day patient survival for first intestine transplants: - Birmingham Women's and Children: 100% (not including liver) - Birmingham Women's and Children: 86.7 (including liver) - King's College: data suppressed to maintain patient confidentiality (not including liver) - King's College: 100% (including liver) • Proportion unadjusted 1-year patient survival for first intestine transplants: - Birmingham Women's and Children's: 90% (not including liver) - Birmingham Women's and Children's: 77% (including liver) - King's College: data suppressed to maintain patient confidentiality (not including liver) - King's College: 90% (including liver) Due to small numbers the data from NHSBT is presented for a number of years rather than 1 year alone. The small numbers of patients also mean that only unadjusted survival rates are presented, which do not account for differences in the case mix at each centre and transplant type
Geographical equity access	Combined analysis conducted for adult and paediatric service. The expected numbers of patients are accessing the service from all NHS England 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	881
Outcomes collated	• Proportion of survival for patients diagnosed with extrahepatic biliary atresia (EHBA) at 16 years of age (proportion alive on 16th birthday with or without native liver): - Birmingham Women's and Children's: 93% - King's College: 90% - Leeds Teaching: 88% • Proportion of 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's: 79% - King's College: 82% - Leeds Teaching: 97%
Geographical equity access	The number of patients accessing the service from NHS England 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 and their families each year.

NHS centre	Cambridge University Hospitals NHS Foundation Trust
Expenditure	>£0.5 million but <£1 million
Index patients	46
Outcomes collated	• Proportion of patients with a definitive diagnosis or diagnosis ruled out: 92%
Geographical equity access	The number of patients accessing the service from NHS England 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 can be hereditary or acquired.

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

Total pancreatectomy with islet autotransplant surgery involves removal of the pancreas followed by islet autotransplantation (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	>£1 million but <£5 million
Number of patient procedures	The number of procedures is <5, 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 of the blood vessels in the brain; the resulting excess blood flow can lead to cardiac problems.

VGMs usually occur in fetuses or newborn babies, although problems sometimes do not present until later in life.

Treatment for VGMs in children involves injecting acrylate or placing a coil in the blood vessels to restore arteriovenous equilibrium.

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

NHS centres	Alder Hey Children's NHS Foundation Trust Great Ormond Street Hospital for Children NHS Foundation Trust
Expenditure	<£0.5 million
Number of patient procedures	31
Outcomes collated	• Proportion of neonates alive but impaired: – Alder Hey Children's: 0% – Great Ormond Street: 10% • Proportion of neonates who died: – Alder Hey Children's: 0 – Great Ormond Street: 20% • Proportion of infants and children alive but impaired – Alder Hey Children's Hospital: 0 – Great Ormond Street: 0 • Proportion of infants and children who died: – Alder Hey Children's: 0 – Great Ormond Street: 0
Geographical equity access	The number of patients accessing the service from NHS England 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 in the body to support a 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, the main pumping chamber of the heart. They sometimes also need to be implanted in the right ventricle.

VAD implantation is only considered in adults 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 VADs.

NHS centres	Great Ormond Street Hospital for Children NHS Foundation Trust 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	Figure included in heart and lung transplant
Number of patient procedures	Long-term VAD procedures: 42 Short-term VAD procedures: 64
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 rates after long-term VAD implant in adults: 63.2%, which has improved over time (60.1% in 2019/20 and 58.4% in 2018/19) • 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% - 1 year: 69.5%

	Note: NHSBT in conjunction with NHS England has 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 NHS England 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 with diabetes, blindness, deafness and brain disease.

Both the adult and paediatric services run clinics that assess all patients in a multidisciplinary structure. Patients are assessed and reviewed by all the specialties 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
Patient caseload	89
Outcomes collated	• Proportion of children with HbA1c in target range: - Birmingham Women's and Children's: 16% • Proportion of adults with a BMI <35: - University Hospitals Birminghamy: 95% 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 NHS England regions is different to what is expected. The potential reasons for this are being explored with the service.

Appendix A: Rare disease collaborative networks

CDKL5

CDKL5 is a rare genetic neurodevelopmental condition that affects children. It is characterised by seizures and developmental challenges.

Designated providers

University Hospitals Bristol and Weston NHS Foundation Trust

National Refractory Coeliac Disease Network

Refractory coeliac disease is a complex autoimmune disorder that predominantly affects adults. It is characterised by persistence or recurrence of symptoms despite a gluten-free diet and is associated with an increased risk of lymphoma.

Designated providers

Lead: Sheffield Teaching Hospitals NHS Foundation Trust

Royal Brompton Hospital (Guy's and St Thomas' NHS Foundation Trust)

National Rare Cystic Lung Disease

Cystic lung disease is an umbrella term for a group of conditions that present with multiple lung cysts. Although the presentation of all these conditions can be similar, their management differs.

Designated providers

Lead: Royal Brompton Hospital (Guy's and St Thomas' NHS Foundation Trust)

Nottingham University Hospitals NHS Trust

Hereditary Haemorrhagic Telangiectasia (HHT)

HHT is a genetic disorder that affects the development of the blood vessels and presents at any age. It commonly first presents with bleeding but causes blood vessels to develop abnormally and can result in widespread complications.

Designated providers

Lead: Imperial College Healthcare NHS Trust

Bronglais District General Hospital, Aberystwyth

King's College Hospital NHS Foundation Trust

Congenital Thoracic Malformations Network

Congenital thoracic malformations are a broad group of abnormalities that result in underdevelopment of the chest or lungs. The abnormalities are often first identified before birth.

Designated providers

Royal Brompton Hospital (Guy's and St Thomas' NHS Foundation Trust)

Familial Pneumothorax

A genetic condition resulting in air accumulating within the chest but outside the lungs, which impacts on breathing. It can affect all ages.

Designated providers

Lead: Cambridge University Hospitals NHS Foundation Trust

Queen Elizabeth University Hospital, NHS Greater Glasgow & Clyde

Hereditary gastrointestinal polyposis syndromes

A genetic condition resulting in the formation of gastrointestinal polyps, often in children and adolescents, leading to a high risk of intestinal cancer.

Designated providers

Lead: St Mark's Hospital, London North West University Healthcare NHS Trust

University Hospitals Birmingham NHS Foundation Trust

Manchester University NHS Foundation Trust

Royal Infirmary of Edinburgh, NHS Lothian

UK Hyperoxaluria RDCN

Hyperoxalurias are a group of diseases (genetic and acquired) that impair the body's ability to remove oxalate. Accumulation of oxalate can result in complications including kidney stones, bone fractures, reduced growth and anaemia.

Designated providers

Lead: Royal Free London NHS Foundation Trust

Birmingham Women's and Children's NHS Foundation Trust

Great Ormond Street Hospital for Children NHS Foundation Trust

University Hospitals Birmingham NHS Foundation Trust

Juvenile Myasthenia Gravis network

Juvenile myasthenia gravis (JMG) is a rare disorder with an estimated incidence of 1.5 per million person years in the UK. JMG is defined as MG in patients 18 years and younger but 20% of cases are under 10 when diagnosed.

Designated providers

Oxford University Hospitals NHS Foundation Trust

Segmental overgrowth and vascular malformations

Segmental overgrowth and vascular malformations are a group of rare disorders characterised by asymmetric overgrowth of tissues.

Designated providers

Lead: Nottingham University Hospitals NHS Trust

United Hospitals of Derby and Burton NHS Foundation Trust,

St George's University Hospital NHS Foundation Trust

Birmingham Women's and Children's NHS Foundation Trust

Manchester University NHS Foundation Trust

Royal Free London NHS Foundation Trust

Paroxysmal motor disorders RDCN

Paroxysmal motor disorders (PMDs) include a number of very rare neurological diseases that affect the ability to move. Nearly all PMDs are rare genetic conditions.

Designated providers

St George's University Hospitals NHS Foundation Trust

Paediatric and primary lymphoedema

Patients with primary lymphoedema have developmental or functional anomalies of the lymphatic system. They may present with lymphoedema affecting one or multiple body segments or associated with problems affecting other organs or systems.

Designated providers

Lead: St George's University Hospitals NHS Foundation Trust

Nottingham University Hospitals NHS Trust

United Hospitals of Derby and Burton NHS Foundation Trust

Mosaic disorders

Mosaic disorders are a group of rare genetic disorders that present with extensive birthmarks; these affects a baby's development in utero.

Designated providers

Great Ormond Street Hospital for Children NHS Foundation Trust

Appendix B: UK-wide commissioning arrangements for highly specialised services during 2020/21

Service name	UK-wide commissioning arrangements
Alkaptonuria service (adults)	Fully commissioned on behalf of England and Scotland
Alström syndrome service (adults and children)	Fully commissioned on behalf of England and Scotland
Ataxia telangiectasia services for adults	Fully commissioned on behalf of England and Scotland
Ataxia telangiectasia services for children	Fully commissioned on behalf of England and Scotland
Atypical haemolytic uraemic syndrome (adults and children)	Fully commissioned on behalf of England and Scotland
Auditory brainstem implant for patients with congenital abnormality of the auditory nerves or cochleae	Fully commissioned on behalf of England only
Autologous intestinal reconstruction service for adults	Fully commissioned on behalf of England only
Bardet-Biedl syndrome service (adults and children)	Fully commissioned on behalf of England and Scotland
Barth syndrome service (adults and children)	Fully commissioned on behalf of England and Scotland
Beckwith-Wiedemann syndrome with macroglossia service (children)	Fully commissioned on behalf of England and Scotland
Behçet's syndrome service (adults and adolescents)	Fully commissioned on behalf of England only
Bladder exstrophy service (children)	Fully commissioned on behalf of England and Scotland
Breast radiotherapy injury rehabilitation service (a discrete cohort of adult females)	Fully commissioned on behalf of England only
Cardiothoracic transplantation service (Paediatrics)	Fully commissioned on behalf of England, in part for Scotland by arrangement, 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 and Scotland

Complex childhood osteogenesis imperfecta service	Fully commissioned on behalf of England only
Complex Ehlers-Danlos syndrome service (adults and children)	Fully commissioned on behalf of England and Scotland
Complex neurofibromatosis type I service (adults and children)	Fully commissioned on behalf of England only
Complex tracheal disease service (children)	Fully commissioned on behalf of England and Scotland
Congenital hyperinsulinism service (children)	Fully commissioned on behalf of England and Scotland
Craniofacial service (adults and children)	Fully commissioned on behalf of UK (pre-1991)
Cryopyrin associated periodic fever syndromes (CAPS) also known as autoinflammatory diseases treated with IL blockers	Fully commissioned on behalf of England and Scotland
Diagnostic service for amyloidosis (all ages)	Fully commissioned on behalf of England and Scotland
Diagnostic service for primary ciliary dyskinesia (adults and children)	Fully commissioned on behalf of England and Scotland
Diagnostic service for rare neuromuscular disorders (adults and children)	Fully commissioned on behalf of England and Scotland
DNA nucleotide excision repair disorders service	Fully commissioned on behalf of England and Scotland
Encapsulating peritoneal sclerosis treatment service (adults)	Fully commissioned on behalf of England only
Epidermolysis bullosa service (adults and children)	Fully commissioned on behalf of England and Scotland
Extracorporeal membrane oxygenation service for adults	Fully commissioned on behalf of England only
Extracorporeal 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
Hand and upper limb reconstruction using vascularised composite allotransplantation	Fully commissioned on behalf of England and 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 (airborne) (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
Liver transplantation service (adults)	Fully commissioned on behalf of England, NI and 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 and Scotland
Lysosomal storage disorders service (children and adults)	Fully commissioned on behalf of England, in part for Scotland (not drugs) and in part for NI (not ERT drugs)
McArdle's disease service (children)	Fully commissioned on behalf of England and Scotland
Multiple sclerosis management service for children	Fully commissioned on behalf of England only
Neurofibromatosis type 2-schwannomatosis* service (all ages) * gene name	Fully commissioned on behalf of England and Scotland
Neuromyelitis optica service (adults and children)	Fully commissioned on behalf of England and Scotland
Ocular oncology service (adults and adolescents)	Fully commissioned on behalf of England, from devolved administrations for Scotland

Open fetal surgery to treat 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
Intestinal pseudo-obstructive disorders service (children)	Fully commissioned on behalf of England and 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 and 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 and Scotland
Pulmonary hypertension service for children	Fully commissioned on behalf of England and in part for Scotland (service only, not drugs)
Pulmonary thromboendarterectomy service (adults and adolescents)	Fully commissioned on behalf of England and Scotland
Rare mitochondrial disorders service (adults and children)	Fully commissioned on behalf of England and Scotland
Retinoblastoma service (children)	Fully commissioned on behalf of UK (pre-1991)

Severe acute porphyria	Fully commissioned on behalf of England and Scotland
Severe combined immune deficiency and related disorders service (children)	Fully commissioned on behalf of England and in part for Scotland
Small bowel transplantation service (adults)	Fully commissioned on behalf of England only
Small bowel transplantation service (children)	Fully commissioned on behalf of England and Scotland
Specialist paediatric liver disease service	Fully commissioned on behalf of UK (pre-1991)
Stickler syndrome diagnostic service (adults and children)	Fully commissioned on behalf of England and Scotland
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 and Scotland
Ventricular assist devices (VADs) as a bridge to heart transplantation or myocardial recovery (all ages)	Fully commissioned on behalf of England and in part for Scotland
Wolfram syndrome service (adults and children)	Fully commissioned on behalf of England and Scotland

NHS England
Wellington House
133-155 Waterloo Road
London
SE1 8UG

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