NHS England

HIGHLY SPECIALISED SERVICES 2017

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Description	This Highlight Report provides detailed information on selected services and gives summary information on all the services in the Highly Specialised services portfolio. This includes: which NHS providers deliver the services; the funding, activity, clinical outcomes and geographical equity access and the NHS England commissioning arrangements on behalf of the devolved nations.
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Contact Details for further information	Fiona Marley Head of Highly Specialised Commissioning Specialised Commissioning NHS England Skipton House, 80 London Road SW1 6LH fiona.marley@nhs.net

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Highly Specialised Services 2017

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Contents

1 Introduction

1. The purpose of this document

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

- A description of the service
- A list of the expert centres that deliver the service
- NHS England's total expenditure on the service
- A measure of the activity that the service undertakes (Patients numbers fewer than 10 are not included because of the risk of identifying individual patients).
- In some cases, clinical outcomes from the service
- Information about geographical equity of access to the service

In a small number of cases, some additional information is provided about the service.

Appendix A details those highly specialised services that were commissioned for the first time in 2016/17.

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

2. Equality Statement

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

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

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

3. Specialised services

NHS England is responsible for commissioning £15.6 billion (2016/17 budget) of specialised services to meet a wide range of health and care needs.

Four factors determine whether a service is commissioned by NHS England as a prescribed specialised service (rather than by Clinical Commissioning Groups (CCGs) as a non-specialised service). The four factors are:

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

4. Highly specialised services

Within specialised services is a subset of services classified as ‘highly specialised’.

Highly specialised services are provided to a smaller number of patients compared to specialised services; usually no more than 500 patients per year.

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

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

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

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

5. Rare Diseases Advisory Group (RDAG)

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

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

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

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

6. Expenditure figures

The expenditure figures for each service reflect NHS England's expenditure on that service in 2016/17. Expenditure is set out into the following categories:

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

7. Clinical outcomes for Highly Specialised Services

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

The data for each unit providing a service are presented at the annual meeting of the service, and provide a stimulus for challenge and learning (or confirmation of good practice). Ideally the data would also be used for national and international

benchmarking, but obtaining comparable data from different units is often challenging, and practically impossible for international comparisons. The solid organ transplant services are an exception to this rule.

Even where comparative data and benchmarks are not available, clinicians report that they find it helpful to have an annual focus on clinical outcomes. It is accepted that the measures are sometimes crude, and simplify the complexity of caring for multi-system disease in heterogeneous patient populations. The data that the HSCT collect will continue to be refined over time.

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

8. Geographical equity of access to Highly Specialised Services

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

The best measure is a metric known as the standardised coefficient of variation (SCV). An SCV above 20 indicates variation greater than expected by chance. Where the SCV is below 20, variation can be considered random. For each service, patients have been mapped according to their home postcode, and rates per million population (child, adult or all-age as appropriate) calculated for each Region or Area.

For most analyses, patients are mapped if they received the intervention (e.g. transplant) or have a confirmed diagnosis of the relevant condition (e.g. Xeroderma pigmentosum).

Where variation is observed which is likely not due to chance (SCV greater than 20), it is possible that there are genuine clusters of disease. For example many genetic

disorders are commoner among highly consanguineous populations which are themselves unequally distributed in England. Conversely, there may be a genuine lack of patients with a particular condition – for example severe osteogenesis imperfecta is unlikely to be missed and there seems to be a genuine lack of patients in the North East of England.

For those services where the SCV is above 20, the HSCT is reviewing the information in greater detail to understand the possible causes. They will then explore options and take specific actions to reduce the inequalities.

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

2 Services and Providers of Highly Specialised Services for 2016/17

Alkaptonuria service (adults)	
Alkaptonuria (AKU) is a rare inherited disorder that causes considerable morbidity in the peak of adulthood due to severe premature destruction of the joints and spine. Disability, often severe, is the norm for those over 30 years of age. There are around 50 people in England with AKU.	
NHS Centre	The Royal Liverpool and Broadgreen University Hospitals NHS Trust
Expenditure	Between £0.5m and £1m
Caseload	50
Outcomes collated	In 2015/6, 9 new patients were seen and 39 were on follow up. Of 21 patients who had been seen for more than a year, improvements were seen in all domains of the SF36 quality of life scale except 'energy / fatigue'. Scores on the AKU Severity score index (AKUSSI) show improvement at one year compared to pre-clinic values.
Geographical equity access	There is some evidence of geographical inequity so further analysis will be undertaken along with ongoing discussions with the service.

Alström syndrome service (adults and children)	
Alström is a rare genetic syndrome that usually presents with blindness in childhood. Patients go on to develop insulin-resistant diabetes, fibrosing cardiomyopathy (where abnormal tissue grows in the heart and stops it working effectively) and renal failure. They may also become deaf. There are thought to be fewer than 100 people affected by Alström syndrome in England.	
NHS Centres	Birmingham Children's Hospital NHS Foundation Trust; University Hospitals Birmingham NHS Foundation Trust
Expenditure	Less than £0.5m
Caseload	74
Geographical equity access	Numbers too small to analyse

Ataxia telangiectasia services for adults	
Ataxia telangiectasia (AT) is a rare, neurodegenerative and progressive condition that starts in early childhood causing severe disability and premature death. It affects many parts of the body and a wheelchair is often needed by the age of 10. The average life expectancy is 25 years. During the adult stage of the condition, there is increased susceptibility to leukaemias, lymphoma, pneumonia, chronic lung disease and neurological decline. Fewer than 100 adults in England have AT.	
NHS Centres	Papworth Hospital NHS Foundation Trust
Expenditure	Less than £0.5m
Caseload	69
Outcomes collated for 2016/17	Median age at death: 35 (this is higher than the median cited in medical publications as 20 years) Median BMI: 22.5 interquartile range 18.4-25.9 kg/m2 (this is an important measure because the nature of the condition often means that patients do not achieve optimum BMIs).
Geographical equity access	No evidence of geographical inequity

Ataxia telangiectasia services for children	
Ataxia telangiectasia (AT) is a rare, neurodegenerative and progressive condition that starts in early childhood causing severe disability and premature death. It affects many parts of the body and a wheelchair is often needed by the age of 10. The average life expectancy is 25 years. About 150 people in England have AT.	
NHS Centres	Nottingham University Hospitals NHS Trust
Expenditure	Less than £0.5m
Caseload	131
Outcomes collated for 2016/17	Quality of life is measured by the PEDS QL instrument. The mean scores were 80 (teen report) and 33.7 (parent report). On their own, these scores are difficult to interpret but trends over time can be monitored.
Geographical equity access	No evidence of geographical inequity

Atypical haemolytic uraemic syndrome (adults and children)	
Atypical haemolytic uraemic syndrome (aHUS) can occur at any age. Onset in childhood appears slightly more frequently than in adulthood (around 60% and 40% of all cases respectively). Most children (70%) who develop aHUS will experience the disease for the first time before the age of 2 years. The true incidence and prevalence of aHUS in England is uncertain because some patients remain undiagnosed. Worldwide, the prevalence of aHUS ranges from 2.7–5.5 per million population, with an incidence of about 0.40 per million population.	
NHS Centres	The Newcastle Upon Tyne Hospitals NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Caseload	108
Outcomes collated for 2016/17	No patient in England died of aHUS in 2016/17
Geographical equity access	New service

Autologous intestinal reconstruction service for adults	
Autologous intestinal reconstruction in adults (AuGIR) is a new surgical procedure in adult patients with short bowel syndromes who are on parenteral nutrition. Patients have insufficient bowel to take in enough food by mouth to provide adequate nutrition. The operation lengthens the bowel so that food can be taken normally. This is an established procedure in children.	
NHS Centre	Salford Royal NHS Foundation Trust
Expenditure	Less than £0.5m
Outcomes collated for 2016/17	Data suppressed to maintain patient confidentiality
Geographical equity access	Numbers too small to analyse

Bardet Biedl syndrome service (adults and children)	
Bardet-Biedl syndrome is a highly debilitating autosomal-recessive genetic disorder that causes early-onset blindness, renal failure, obesity, diabetes, Hirschsprung disease, urological problems and neurological deficits. About 1 in 100,000 babies are born each year with Bardet-Biedl syndrome, i.e. five or six each year in England.	
NHS Centres	Birmingham Children's Hospital NHS Foundation Trust; Great Ormond Street Hospital for Children NHS Foundation Trust; Guy's and St Thomas' NHS Foundation Trust; University Hospitals Birmingham NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Assessments	216
Geographical equity access	Data not available or not comparable

Barth syndrome service (male adults and children)	
Barth syndrome is an x-linked disorder of lipid metabolism presenting as cardiac/skeletal myopathy, neutropenia and growth retardation with a high infant mortality rate. Patients with Barth Syndrome present with frequent cardiac problems and, in two-thirds of patients, neutropenia (reduced white blood cell count leading to susceptibility to infection). When undiagnosed or treated by non-specialists, patients typically experience frequent hospital admissions for a range of diagnostic tests and treatment of severe infections. Infections are significantly reduced through protocol-driven prescription of granulocyte colony stimulating factor (G-CSF). About 30 people in England have Barth syndrome.	
NHS Centre	University Hospitals Bristol NHS Foundation Trust
Expenditure	More than £0.5m but less than £1m
Caseload	30
Outcomes collated for 2016/17	Numbers too small to analyse
Geographical equity access	Numbers too small to analyse

Beckwith-Wiedemann syndrome with macroglossia service (children)	
Beckwith-Wiedemann syndrome is a disorder present at birth, characterised by an increased risk of childhood cancer and certain congenital features. One of the congenital features is macroglossia (significant enlargement of the tongue), which causes: drooling; feeding, speech, orthodontic and dental problems; and devastating psychosocial consequences. About 1 in 15,000 babies are born each year with Beckwith-Wiedemann syndrome but only about half have macroglossia, i.e. about 15-20 babies each year.	
NHS Centres	Great Ormond Street Hospital for Children NHS Foundation Trust
Expenditure	Less than £0.5m
Caseload	175
Outcomes collated for 2016/17	Data suppressed to maintain patient confidentiality as these are surgical interventions and the numbers very small.
Geographical equity access	Numbers too small to analyse

Behçet's syndrome service (adults and adolescents)	
Behçet's syndrome is a chronic, inflammatory, multi-systemic vasculitic disorder with a wide spectrum of clinical presentations that may include blindness, severe ulceration and cardiovascular problems. There are around 1,500 people in England that have Behçet's syndrome.	
NHS Centres	Aintree University Hospital NHS Foundation Trust; Barts Health NHS Trust; Sandwell & West Birmingham Hospitals NHS Trust
Expenditure	More than £1m but less than £5m
Caseload	1,300
Outcomes collated for 2016/17	35% of 751 patients experienced decreased flare activity. This decrease may be due to natural fluctuation in the disease but is also the result of effective new therapies now available.
Geographical equity access	Evidence of geographical inequity is being investigated but is likely due to data quality issues

Bladder exstrophy service (children)	
The service provides diagnostic, management advice and treatment for children with bladder exstrophy, primary epispadias, cloacal exstrophy and all variants. Expert management and appropriate surgical reconstruction can provide a child suffering from bladder exstrophy with a near normal lifestyle. The goals of exstrophy reconstruction are: • Anatomic reconstruction of the bladder/urethra, bony pelvis, abdominal wall and external genitalia • Creation of urinary continence with preservation of renal function • Healthy psychological adjustment and adaptation to the condition throughout life • Support during adolescence Between 1 in 30,000 and 1 in 50,000 babies are born each year with bladder exstrophy, i.e. around 20 babies each year in England.	
NHS Centres	Central Manchester University Hospitals NHS Foundation Trust; Great Ormond Street Hospital for Children NHS Foundation Trust
Expenditure	More than £1m but less than £5m
New babies	20
Geographical equity access	Data not available or not comparable

Breast radiotherapy injury rehabilitation service (a discrete cohort of adult females)	
This service is for a discrete cohort of women who have severe, chronic and complex conditions arising from radiation-induced injuries. The women received a treatment regime for breast cancer in the 1970s and 1980s that is now known to be associated with a particular risk of damage to the nerves of the brachial plexus.	
NHS Centre	Royal National Hospital for Rheumatic Diseases - Royal United Hospitals Bath NHS Foundation Trust
Expenditure	More than £0.5m but less than £1m
Caseload	77
Outcomes collated for 2016/17	Average 19% reduction in perception of pain; GP visits reduced from 1.7 in the three months before to 1.4 in the three months after attending service; visits to other doctors for pain reduced from 2.9 to 0.5.
Geographical equity access	Data not available or not comparable

Cardiothoracic Transplantation service (paediatrics)	
The heart and lung transplant service provides: assessment of patients who are eligible for a heart transplant; the transplant operation; and lifelong follow up.	
NHS Centres	Great Ormond Street Hospital for Children NHS Foundation Trust; The Newcastle Upon Tyne Hospitals NHS Foundation Trust
Expenditure	More than £50m (adults and children, heart and lung)
Number of transplants	35
Outcomes collated for 2016/17	30 day survival heart unadjusted 95.9% 90 day survival lung unadjusted 92% Unadjusted 1 year survival heart transplant 92.5% Unadjusted 1 year survival lung transplant 92% Unadjusted 5 year survival heart transplant 82.6% Unadjusted 5 year survival lung transplant 73.9%
Geographical equity access	Data not available or not comparable

Choriocarcinoma service (adults and adolescents)	
This service diagnoses and treats women with the different types of gestational trophoblastic disease including the following: • Hydatidiform mole (also known as molar pregnancy): in this condition, the sperm and egg cells join together but a healthy foetus does not develop. The placenta grows to an abnormal size, requiring surgical evacuation of the uterus. • Choriocarcinoma, which is an aggressive and malignant cancer that may spread from the uterus to other organs in the body, such as the lungs or brain. Each year about 10 women in England develop choriocarcinoma. • Placental site trophoblastic tumour, a rare variant of choriocarcinoma. This cancer is able to spread through the body via the lymphatic system.	
NHS Centres	Imperial College Healthcare NHS Trust; Sheffield Teaching Hospitals NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Inpatient Episodes	790
Outcomes collated for 2016/17	100% cure rate for low risk patients; 94% cure rate for high risk patients.
Geographical equity access	Data required from cancer register

Chronic pulmonary aspergillosis service (adults)	
Chronic pulmonary aspergillosis is a chronic, progressive infection of the lung with the fungus *Aspergillus fumigatus* that follows a lung insult (typically sarcoidosis, atypical TB or recurrent pneumothoraces) and occurs in those with one or more innate genetic defects.	
NHS Centre	University Hospital of South Manchester NHS Foundation Trust
Expenditure	More than £5 million but less than £10m
Caseload	425
Outcomes collated for 2016/17	Of 67 patients whose treatment was initiated in 2016/17, follow up data is available so far for 42. Seven of these patients showed a 12 point improvement in total score on the St George's Respiratory Questionnaire. This is a substantial improvement in quality of life in a condition which untreated shows no improvement at all.
Geographical equity access	Strong evidence of geographical inequity which is long-standing and commissioning solutions are being explored.

Complex childhood osteogenesis imperfecta service	
Osteogenesis imperfecta (OI) is a genetic condition characterised by bones that break easily, often from little or no apparent cause. The condition can vary quite drastically from one person to another: a person can have just a few or as many as several hundred fractures in a lifetime. About 300 children in England have severe or complex OI.	
NHS Centres	Birmingham Children's Hospital NHS Foundation Trust; Great Ormond Street Hospital for Children NHS Foundation Trust; Sheffield Children's NHS Foundation Trust; University Hospitals Bristol NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Caseload	308
Outcomes collated for 2016/17	37% of patients under follow up have non-vertebral fractures, and 60% have vertebral fractures. Scoliosis: 9% have Cobb angle >45.
Geographical equity access	No evidence of geographical inequity

Complex Ehlers Danlos syndrome service (adults and children)	
Ehlers-Danlos syndrome (EDS) is a group of heritable disorders of connective tissue. The major clinical features are hyperextensible skin, hypermobile joints and tissue fragility. In severe cases, patients can have life-threatening complications such as aortic dissection, where the layers of the wall of the aorta separate. There are several types of EDS each with their own specific management. The service diagnoses about 200 new patients each year with the rare forms of EDS.	
NHS Centres	London North West Healthcare NHS Trust; Sheffield Children's NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Definitive diagnosis	EDS confirmed in 189 patients and ruled out in 117
Geographical equity access	Data not available or not comparable

Complex neurofibromatosis type I service (adults and children)	
Neurofibromatosis type 1 (NF1) is an inherited genetic disorder, characterised by formation of neurofibromas (tumours involving nerve tissue) in the skin, subcutaneous tissue, cranial nerves and spinal root nerve tissue. About 1 in 25,000 of the population has NF1. With *complex* NF1 there is a high risk of developing rare complications, which may affect most of the body systems. Complex in NF1 is defined by the presence of other conditions that have the possibility of significant morbidity and mortality and which require integrated management by an expert team.	
NHS Centres	Central Manchester University Hospitals NHS Foundation Trust; Guy's and St Thomas' NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Outpatient Attendances	762
Outcomes collated for 2016/17	Guy’s and St Thomas’ NHS Foundation Trust saw 45 new and 388 follow-up complications, which require their integrated approach to management. The commonest problems were symptomatic and disfiguring plexiform neurofibroma, and brain glioma.
Geographical equity access	There is evidence of geographical inequity and the establishment of a new shared care clinic should help to resolve this issue.

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.	
NHS Centre	Great Ormond Street Hospital for Children NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Inpatient Episodes	25
Geographical equity access	Numbers too small to analyse

Congenital hyperinsulinism service (children)	
Congenital hyperinsulinism (CHI) is a condition characterised by excess insulin production from the pancreas, resulting in hypoglycaemia. The clinical presentation and progress of CHI lies on a spectrum, varying between those with transient hypoglycaemia to those unresponsive to medical treatment and requiring pancreatectomy. In the absence of expert management, children who have prolonged or recurrent hyperinsulinaemic hypoglycaemia in infancy can suffer harm to their brains and may be developmentally delayed.	
NHS Centres	Central Manchester University Hospitals NHS Foundation Trust; Great Ormond Street Hospital for Children NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Caseload	919
Geographical equity access	Data not available or not comparable

Craniofacial service (adults and children)	
This service provides assessment, surgical and non-surgical treatment and follow-up of patients with severe congenital deformities of the skull and face.	
NHS Centres	Alder Hey Children's NHS Foundation Trust; Birmingham Children's Hospital NHS Foundation Trust; Great Ormond Street Hospital for Children NHS Foundation Trust; Oxford University Hospitals NHS Trust
Expenditure	More than £5 million but less than £10m
Assessments	153
Outcomes collated for 2016/17	18 patients; 0 grade 3 or 4 complications; this measure confirms that treatment in an expert centre results in a low number of complications
Geographical equity access	Data not available or not comparable

Cryopyrin associated periodic syndrome service (adults)	
Cryopyrin associated periodic syndrome (CAPS), is a very rare lifelong inflammatory disease that interferes with growth and development that causes serious morbidity and is often fatal.	
NHS Centre	Royal Free London NHS Foundation Trust
Expenditure	More than £5 million but less than £10m
Patients on high cost drugs	120
Outcomes collated for 2016/17	No new cases of amyloidosis since programme started; no progression to renal dialysis among treated patients; CAPS activity score normal in >90% of patients
Geographical equity access	Data not available or not comparable

Cryopyrin associated periodic syndrome ('CAPS') is a very rare disease caused by widespread activation of inflammatory processes throughout the body. It results in rashes, fevers, joint pains and a very low quality of life.

The service, based at the Royal Free Hospital in London, cares for adults and children with CAPS (the latter in partnership with Great Ormond Street Hospital). The drug canakinumab is highly effective in treating CAPS but it is also one of the world's most expensive drugs. The alternative drug anakinra works for some patients but has unpleasant side effects so that many cannot tolerate it. The service ensures that canakinumab is used only in those patients who need it, and has developed several schemes such as vial sharing to reduce wastage and make cost-effective use of the product.

Effective treatment has resulted in major, sustained improvements in quality of life for patients in the last year: the CAPS activity score has returned to normal in over 90% of patients no new patient has developed the complication of amyloidosis while on treatment; and no patient who already had amyloid (deposits that can build up and cause damage to body systems) when started on treatment has gone on to develop kidney failure.

Diagnostic service for amyloidosis (adults)	
Amyloidosis is a condition in which abnormal protein deposits accumulate in many different organs.	
NHS Centre	Royal Free London NHS Foundation Trust
Expenditure	More than £5m but less than £10m
First evaluations	1,203
Outcomes collated for 2016/17	Amyloid type confirmed in 1005 patients. Amyloidosis ruled out in 482.
Geographical equity access	Data not available or not comparable

Diagnostic service for primary ciliary dyskinesia (adults and children)	
Primary ciliary dyskinesia is a genetic disorder of the air tubes of the lungs (the bronchi), which become infected and filled with pus due to abnormalities of the hair-like structure (cilia) of the cells lining the respiratory tract. This can lead to repeated infections and damage the lung, especially if the diagnosis is delayed. Around 100 children are diagnosed with PCD each year in England.	
NHS Centres	Royal Brompton & Harefield NHS Foundation Trust; University Hospital Southampton NHS Foundation Trust; University Hospitals of Leicester NHS Trust
Expenditure	More than £1m but less than £5m (for management and diagnostic elements)
Number of positive samples	71
Geographical equity access	Data not available or not comparable

Diagnostic service for rare neuromuscular disorders (adults and children)	
The aim of the service is to make a precise molecular or clinical diagnosis in patients with four rare neuromuscular conditions and to assess fully the extent of their disease: • Limb girdle muscular dystrophies • Congenital muscular dystrophies • Congenital myasthenic syndromes • Muscle channelopathies (also known as periodic paralysis)	
NHS Centres	Great Ormond Street Hospital for Children NHS Foundation Trust; Oxford University Hospitals NHS Trust; The Newcastle Upon Tyne Hospitals NHS Foundation Trust; University College London Hospital NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Number of referrals	1,573
Geographical equity access	No evidence of geographical inequity

The rare neuromuscular service provides a firm diagnosis for patients with rare neuromuscular disease. The service is based in London (two centres), Newcastle and Oxford. Each centre has expertise in a particular group of neuromuscular diseases: London (Great Ormond Street Hospital) and Newcastle focus on rare muscular dystrophies, in which the substance of the muscle is affected by mutations in key proteins; London (National Hospital for Neurosurgery and Nervous Disorders, Queen Square) focuses on disorders of the cell membrane channels which are crucial for triggering muscle contraction (channelopathies); and Oxford focuses on disorders of the junction between nerve and muscle where contraction is signalled (mutations of the acetylcholine receptor).

Based on long experience, the centres are able to make a firm clinical diagnosis from meticulous attention to the patient's account of symptoms and clinical examination; and in 70 – 80% a detailed molecular diagnosis of the causative mutation is possible. In the remainder the mutation is unknown or undiscovered.

The centres are all research active in the discovery of new genes and new mechanisms of disease; the channelopathy centre was also able to show that an old drug, previously used for treat heart arrhythmias but considered obsolete in cardiology, was effective in one form of channelopathy. Such repurposing of old drugs is hugely cost-effective compared to developing a completely new drug from scratch, but it depends on a detailed molecular knowledge of the disease process.

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 sub-acute or acute bowel obstruction. As a chronic fibrosing process, it leads to abdominal pain, nausea, vomiting, weight loss, fever, malnutrition, anaemia, ascites and finally surgical peritonitis and mortality.	
NHS Centres	Cambridge University Hospitals NHS Foundation Trust; Central Manchester University Hospitals NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Primary surgical procedures	15
Outcomes collated for 2016/17	94%of patients were alive post operation. 100% of survivors no longer needed total parenteral nutrition
Geographical equity access	No evidence of geographical inequity

Epidermolysis bullosa service (adults and children)	
Epidermolysis bullosa (EB) is the name given to a group of rare inherited disorders that cause lifelong blistering and ulceration of the skin and often the mucous membranes. Blistering is almost always apparent at or soon after birth, but the severity of the condition varies greatly, depending on the type of Epidermolysis bullosa present.	
NHS Centres	Birmingham Children's Hospital NHS Foundation Trust; Great Ormond Street Hospital for Children NHS Foundation Trust; Guy's and St Thomas' NHS Foundation Trust; Heart Of England NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Caseload	1050 (caseload based on three centres)
Outcomes collated for 2016/17	A quality of life scores is recorded for patients transitioning to the adult service but numbers are very small and it will be several years for any trend to become apparent.
Geographical equity access	Data not available or not comparable

Extra corporeal membrane oxygenation service for adults	
Extra corporeal membrane oxygenation (ECMO) supports adults with severe potentially reversible acute respiratory failure by oxygenating the blood through an artificial lung machine.	
NHS Centres	Guy's and St Thomas' NHS Foundation Trust; Papworth Hospital NHS Foundation Trust; Royal Brompton & Harefield NHS Foundation Trust; University Hospital of South Manchester NHS Foundation Trust; University Hospitals of Leicester NHS Trust
Expenditure	More than £20m but less than £30m (adults and children)
Starting treatment	209
Outcomes collated for 2016/17	82% survival at discharge 201617
Geographical equity access	There is some evidence of geographical inequity in the service. The issues are understood and the inequity should be resolved in 2017/18.

Extra corporeal membrane oxygenation service for neonates, infants and children with respiratory failure	
Extra corporeal membrane oxygenation (ECMO) supports critically ill babies and children who have severe potentially reversible acute respiratory failure by oxygenating the blood through an artificial lung machine.	
NHS Centres	Great Ormond Street Hospital for Children NHS Foundation Trust; The Newcastle Upon Tyne Hospitals NHS Foundation Trust; University Hospitals of Leicester NHS Trust
Expenditure	More than £20m but less than £30m (adults and children)
Starting treatment	84
Outcomes collated for 2016/17	81% survival at discharge 2016/17 (child) 87% survival at discharge 2016/17 (neonate)
Geographical equity access	No evidence of geographical inequity

Ex-vivo partial nephrectomy service (adults)	
Ex-vivo partial nephrectomy can be used to treat cancers in patients with a single kidney, and offers the possibility of cancer cure and avoiding a life of dialysis.	
NHS Centre	Oxford University Hospitals NHS Trust
Expenditure	Less than £0.5m
Patients Accepted into service	12
Outcomes collated for 2016/17	Data suppressed to maintain patient confidentiality
Geographical equity access	Numbers too small to analyse

Gender identity development service for children and adolescents	
The gender identity development service is a Tier 4 specialist multidisciplinary mental health service that provides support and therapeutic input for children and adolescents who have social and psychological difficulties with the development of their gender identity. Depending on need, the service works with paediatric endocrinology clinics to prescribe and administer hormone therapy and early intervention from 12 years onwards.	
NHS Centres	The Tavistock and Portman NHS Foundation Trust; Leeds Teaching Hospitals NHS Trust
Expenditure	More than £1m but less than £5m
Number of referrals	1,782
Outcomes collated for 2016/17	8% reduction (whose opinions were recorded at both timepoint 0 and timepoint 2) in self harm or thinking of self harm between the first appointment and 12 months. Children's global assessment scale recorded as generally positive changes in young people's functioning observed
Geographical equity access	Data not available or not comparable

Hand and upper limb transplantation service (adults)	
Hand and upper limb transplantation services include services provided by Highly Specialist Hand and Upper Limb Transplantation Centres. This applies to provision in adults.	
NHS Centres	Leeds Teaching Hospitals NHS Trust
Expenditure	Less than £0.5m
Outpatient assessments	34
Outcomes collated for 2016/17	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 patients who are eligible for a heart transplant; the transplant operation; and lifelong follow up.	
NHS Centres	Papworth Hospital NHS Foundation Trust; Royal Brompton & Harefield NHS Foundation Trust; The Newcastle Upon Tyne Hospitals NHS Foundation Trust; University Hospital of South Manchester NHS Foundation Trust; University Hospitals Birmingham NHS Foundation Trust; Sheffield Teaching Hospitals NHS Foundation Trust (Follow-up only).
Expenditure	More than £50m (adult and children, heart and lung)
Number of transplants	141
Outcomes collated for 2016/17	30 day survival unadjusted 89.9% Unadjusted 1 year survival 82.4% 5 year survival from listing 65.1%
Geographical equity access	Some evidence of geographical inequity which are thought to be of a temporary nature

Insulin Resistant Diabetes (adults and children)	
Insulin-resistant diabetes occurs because of either a genetic condition or because the individual has developed antibodies to insulin. In addition to the usual complications of diabetes (renal failure, stroke, etc.) the condition can also affect the liver and can result in pancreatitis.	
NHS Centre	Cambridge University Hospitals NHS Foundation Trust
Expenditure	Less than £0.5m
Active caseload	174
Outcomes collated for 2016/17	A genetic diagnosis of severe insulin resistance was achieved for 97/173 patients. HbA1c was reduced in 63 / 92 patients with elevated HbA1c at baseline (mean reduction from 81mmol/mol to 71mmol/mol). HbA1c levels did not reduce significantly in 29/ 92 patients.
Geographical equity access	Strong evidence of geographical inequity the reasons of which are not fully understood so will be explored.

Islet transplantation service (adults)	
Islet transplantation is of proven benefit for a very small group of eligible patients with Type 1 diabetes who suffer from recurrent episodes of severe hypoglycaemia. Successful transplantation can abolish episodes of hypoglycaemia unawareness and improve the quality of life of recipients, whilst also improving overall metabolic control. Patients who are already immunosuppressed for a kidney transplant may also benefit from islet transplantation through the improved metabolic control afforded by an islet after kidney transplant.	
NHS Centres	Central Manchester University Hospitals NHS Foundation Trust; King's College Hospital NHS Foundation Trust; North Bristol NHS Trust; Oxford University Hospitals NHS Trust; Royal Free London NHS Foundation Trust; The Newcastle Upon Tyne Hospitals NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Number of Transplants	10
Outcomes collated for 2016/17	Median number of severe hypoglycaemic events is zero at 12 months post-transplant (compared to 9 in the 12 months pre-transplant); HbA1c dropped from 8.0% to 6.6%

Geographical equity access	Numbers too small to analyse

Liver and live liver transplantation service (adults)	
This service provides assessment, transplantation and lifelong follow up for patients requiring liver transplant surgery, including from living donors. The three main conditions for liver transplantations are primary and secondary biliary cirrhosis, chronic hepatitis and fulminant hepatic failure. There are about 900 liver transplants in the UK each year.	
NHS Centres	Cambridge University Hospitals NHS Foundation Trust; King's College Hospital NHS Foundation Trust; Leeds Teaching Hospitals NHS Trust; Royal Free London NHS Foundation Trust; The Newcastle Upon Tyne Hospitals NHS Foundation Trust; University Hospitals Birmingham NHS Foundation Trust
Expenditure	More than £50m (adults and children)
Number of transplants	757
Outcomes collated for 2016/17	The national rates of patient survival after joining the transplant list for adult elective first liver only patients is 83% at one, 70% at five and 57% at ten years post- registration.
Geographical equity access	No evidence of geographical inequity

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 (ALF).There are about 100 paediatric liver transplants in England each year.	
NHS Centres	Birmingham Children's Hospital NHS Foundation Trust; King's College Hospital NHS Foundation Trust; Leeds Teaching Hospitals NHS Trust
Expenditure	More than £50m (adults and children)
Number of transplants	105
Outcomes collated for 2016/17	One year unadjusted patient survival for paediatric elective deceased donor first liver transplants: 97.1% Five year unadjusted patient survival for paediatric elective deceased donor first liver transplants: 91.5%
Geographical equity access	No evidence of geographical inequity

Lung Transplantation service (adults)	
The lung transplant service provides: assessment of patients who are eligible for a lung transplant; the transplant operation; and lifelong follow up.	
NHS Centres	Royal Brompton & Harefield NHS Foundation Trust; Papworth Hospital NHS Foundation Trust; The Newcastle Upon Tyne Hospitals NHS Foundation Trust; University Hospital of South Manchester NHS Foundation Trust; University Hospitals Birmingham NHS Foundation Trust
Expenditure	More than £50m (adult and children, heart and lung)
Number of transplants	140
Outcomes collated for 2016/17	90 day survival unadjusted 90% Unadjusted 1 year survival 76.7% 5 year survival from listing 47.2%
Geographical equity access	No evidence of geographical inequity

Lymphangioleiomyomatosis	
Lymphangioleiomyomatosis (LAM) is a rare, progressive disease characterised by lung cysts, kidney tumours and lymphatic abnormalities. LAM occurs in a sporadic form, which affects only females, usually of childbearing age; LAM also occurs in patients who have tuberous sclerosis complex (TSC), a genetic condition that causes non-malignant tumours to grow in the brain and on other vital organs. About 160 women in England have LAM.	
NHS Centre	Nottingham University Hospitals NHS Trust
Expenditure	Less than £0.5m
Caseload	164
Outcomes collated for 2016/17	5 % patients had a pneumothorax; this is a low number for patients with this condition. 0% patients had a renal angiolipoma bleed; this is a very low number for this group of patients as LAM patients are at increased risk of bleed from their angiolipomas. 17 % patients had an FEV1 decline of more than 150 ml per annum; 83% had decline rates of less than 150 ml per annum
Geographical equity access	Evidence of geographical inequity. This is caused by the expert centre's ability to detect very mild forms of the disease which would be missed elsewhere.

Lysosomal storage disorders service (children & adults)	
Lysosomal storage disorders (LSDs) are a group of rare genetic storage disorders, characterised by specific lysosomal enzyme deficiencies. Some LSDs can be treated using enzyme replacement therapies (ERTs), substrate reduction therapy (SRT) or other disease modifying drugs. There are licensed disease-modifying treatments for eight LSDs: • Gaucher's disease • Anderson-Fabry's disease • Mucopolysaccharidosis type I (MPSI, which occurs as Hurler's syndrome, Hurler-Scheie syndrome and Scheie syndrome) • Mucopolysaccharidosis Type IVa (Morquio syndrome) • Mucopolysaccharidosis type VI (MPSVI or Maroteaux Lamy syndrome) • Pompe's disease • Mucopolysaccharidosis type II (MPSII) • Niemann Pick type C	
NHS Centres	Birmingham Children's Hospital NHS Foundation Trust; Cambridge University Hospitals NHS Foundation Trust; Central Manchester University Hospitals NHS Foundation Trust; Great Ormond Street Hospital for Children NHS Foundation Trust; Royal Free London NHS Foundation Trust; Salford Royal NHS Foundation Trust; University College London Hospital NHS Foundation Trust; University Hospitals Birmingham NHS Foundation Trust
Expenditure	More than £50m
Active Caseload	1972 (Of these, the total number of patients receiving enzyme replacement therapy and substrate reduction therapy drugs is 935)
Outcomes collated for 2016/17	Markers agreed: Fabry – initiation of renal replacement therapy in patients who had been on enzyme replacement (or migalastat) for three years or more Gaucher – hospital admission for a bone crisis MPS (any) – acute cranio cervical episode
Geographical equity access	New data definition proposed

McArdle's disease service (children)	
McArdle's disease is a condition caused by an inborn deficiency of muscle phosphorylase resulting in an abnormal accumulation of glycogen in muscle tissue, characterised by exercise intolerance, muscular pain, fatigability and muscle cramping. Rhabdomyolysis leading to renal failure is a particularly severe complication of McArdle's disease. There are fewer than 200 people in England with McArdle's disease.	
NHS Centre	University College London Hospital NHS Foundation Trust
Expenditure	Less than £0.5m
Caseload	191
Outcomes collated for 2016/17	Rhabdomyolysis had occurred in 13/20 (65%) of new patients, and in 6/159 (3%) who were under follow up by the service. In 16 patients assessed over 4 clinic visits, there was a 4 point improvement in physical, and 7 point improvement in mental component scores on the SF36 measure.
Geographical equity access	No evidence of geographical inequity

Neurofibromatosis type 2 service (All Ages)	
Neurofibromatosis type 2 (NF2) is a genetic disorder characterised by the growth of non-cancerous tumours in the central nervous system. NF2 patients develop bilateral vestibular schwannomas (abnormal tissue growth originating in the cells of the sheath around the nerve), meningiomas (a type of benign brain tumour) and spinal tumours; usually causing deafness, balance problems, compression of the brain stem and premature death. There are about 900 people in England who have NF2.	
NHS Centres	Cambridge University Hospitals NHS Foundation Trust; Central Manchester University Hospitals NHS Foundation Trust; Guy's and St Thomas' NHS Foundation Trust; Oxford University Hospitals NHS Trust
Expenditure	More than £5 million but less than £10m
Caseload	903
Outcomes collated	The rate of facial palsy following vestibular schwannoma surgery across all four centres for the period 2011 – 2015 was 10%
Geographical equity access	No evidence of geographical inequity

Neuromyelitis optica service (adults and children)	
Neuromyelitis optica (NMO) (also known as Devic's disease) is a rare inflammatory demyelinating disorder of the central nervous system that typically presents as severe optic neuritis and longitudinally extensive myelitis often followed by further severe attacks, which usually result in permanent disability (visual loss, limb weakness, respiratory muscle weakness). There are high mortality and morbidity rates associated with the condition. About 1,000 people in England are living with NMO.	
NHS Centres	Oxford University Hospitals NHS Trust; The Walton Centre NHS Foundation Trust
Expenditure	More than £1m but less than £5m
First Evaluations	180
Outcomes collated for 2016/17	Annualised relapse rate: pre 0.37; post 0.22
Geographical equity access	Data not available or not comparable

Neuromyelitis optica (NMO) is a disease which affects the nervous system. The disease comes and goes but in severe forms can lead to blindness and paralysis, with death from respiratory failure. NMO has many similarities with multiple sclerosis but the distinction is crucial because treatments for the two diseases are almost exactly opposite – immune stimulation for multiple sclerosis and immune suppression for NMO.

The service, based in Liverpool and Oxford, provides accurate diagnosis based on a laboratory test for the aquaporin antibody, found in NMO patients, and advice on the use of the drug rituximab, which is effective but unfamiliar to many neurologists. The effectiveness of this drug is shown by a reduction in the frequency of attacks from an average of 0.4 per patient per year to 0.2.

Although the drug is effective, the disease progresses in many patients. Specialist nurses and the service provide advice on living with the disease through phone and email advice, and a portfolio of leaflets. There are close links to the patient organisation.

Ocular oncology service (adults)	
The ocular oncology service provides diagnosis and treatment of adults with suspected malignant tumours of the eye. Of the patients referred to the service one third (about 700 each year) are confirmed as having eye cancer.	
NHS Centres	Moorfields Eye Hospital NHS Foundation Trust; The Royal Liverpool and Broadgreen University Hospitals NHS Trust; Sheffield Teaching Hospitals NHS Foundation Trust
Expenditure	More than £5m but less than £10m
Positive assessment	673
Outcomes collated for 2016/17	Primary enucleation rate: 25%; unplanned retreatment for ocular melanoma: 44 patients among >3000 survivors
Geographical equity access	Data required from cancer register

The commonest eye cancer is melanoma. The aim of treatment is to cure the cancer without having to remove the eye. This is possible for the majority of patients, around 75%. Most patients are treated by sewing a small radioactive plaque over the tumour to destroy it, or by referral to the low energy[1] proton therapy facility at Clatterbridge. The highly specialised service is limited to surgical treatment; if the cancer spreads to other organs such as the liver, chemotherapy is required and the patient is treated in a regional cancer centre.

[1] This is different to the proton beam therapy service described elsewhere in this document.

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.	
NHS Centres	Central Manchester University Hospitals NHS Foundation Trust; The Royal Liverpool and Broadgreen University Hospitals NHS Trust; Sheffield Teaching Hospitals NHS Foundation Trust; University College London Hospital NHS Foundation Trust,
Expenditure	More than £1m but less than £5m
Total cases received	4,254
Outcomes collated for 2016/17	The service processed 8214 specimens in 2016 (calendar year). 95% of specimens were reported within 10 days.
Geographical equity access	Data not available or not comparable

Osteo-odonto-keratoprosthesis service for corneal blindness (adults)	
Osteo-odonto-keratoprosthesis (OOKP) surgery is a specialist surgical intervention that can restore meaningful vision to patients suffering from end stage corneal blindness, and for whom conventional corneal surgery is not possible for reasons such as severe ‘dry eyes’ that causes heavy scarring of the cornea. OOKP is only contemplated in patients where no other treatments would restore sight.	
NHS Centre	Brighton and Sussex University Hospitals NHS Trust
Expenditure	Less than £0.5m
Stage 2 surgery	Data suppressed to maintain patient confidentiality
Outcomes collated for 2016/17	Visual acuity 6/9 or better in 3 of 4 patients; 4th patient acuity 6/24
Geographical equity access	Numbers too small to analyse

Paediatric intestinal pseudo-obstructive disorders service	
Chronic intestinal pseudo-obstruction is an intestinal motility disorder. Impaired intestinal motor activity causes recurrent symptoms of intestinal obstruction in the absence of mechanical occlusion. The service provides expert, multi-disciplinary diagnostic services for infants and children under five with congenital and acquired variations of the condition.	
NHS Centre	Great Ormond Street Hospital for Children NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Number of new patients	20
Outcomes collated for 2016/17	Definitive diagnosis achieved in 6/10 patients aged under five years
Geographical equity access	Data not available or not comparable

Pancreas transplantation service (adults)	
This service provides assessment, transplantation and lifelong follow up for diabetic patients requiring pancreas transplant surgery.	
NHS Centres	Cambridge University Hospitals NHS Foundation Trust; Central Manchester University Hospitals NHS Foundation Trust; Guy's and St Thomas' NHS Foundation Trust; Imperial College Healthcare NHS Trust; Oxford University Hospitals NHS Trust; The Newcastle Upon Tyne Hospitals NHS Foundation Trust
Expenditure	More than £5 million but less than £10m
Number of transplants (SPK,PAK,PO)	154
Outcomes collated for 2016/17	One- and five-year patient survival after first simultaneous pancreas and kidney graft was 97% and 89%.
Geographical equity access	No evidence of geographical inequity

The pancreas transplant service is based at six centres in Newcastle, Manchester, Oxford, Cambridge and London (two centres). The aim of pancreas transplant is to improve long term survival in diabetic patients who are having a kidney transplant: the pancreas contains the insulin-producing islet cells which do not work properly in diabetes. Transplant is hugely beneficial to quality of life for patients with kidney failure, but the drugs needed to prevent rejection have some serious adverse effects; hence pancreas transplant is only justified in two circumstances: (a) when the patient needs a transplant anyway because of kidney failure; or (b) in a small number of cases, when the diabetes is so difficult to control as to be life-threatening. Pancreas transplant is a lengthy and technically demanding operation, so patients must be fit for surgery. Unfortunately, in patients whose diabetes has caused kidney failure the heart is often also affected; hence only about 150 patients per year are suitable for pancreas transplant. It is the only transplant service in which organ supply is not the main limitation.

The allocation of donated organs is organised by NHS Blood and Transplant using a points system which effectively creates a single national waiting list; a donated pancreas is allocated to the patient who most needs it.

Paroxysmal nocturnal haemoglobinuria	
Paroxysmal nocturnal haemoglobinuria (PNH) is a rare disease in which red blood cells break down earlier than normal. Symptoms include abdominal pain, back pain, blood clots, dark urine, easy bruising or bleeding, headache and shortness of breath. About 650 people in England suffer from PNH.	
NHS Centres	King's College Hospital NHS Foundation Trust; Leeds Teaching Hospitals NHS Trust
Expenditure	More than £50 million
Caseload	237 patients receiving eculizumab for PNH 386 patients are not receiving eculizumab
Outcomes collated for 2016/17	Mortality on eculizumab closely approximates mortality in the general population (matched for age and sex), i.e. 100% relative survival, compared to 65% survival at 5 years in untreated patients. Mean annual transfusion requirement is 11.5 units before eculizumab and 0.34 after.
Geographical equity access	No evidence of geographical inequity

Primary ciliary dyskinesia management service (children)	
Primary ciliary dyskinesia (PCD) is a genetic disorder of the air tubes of the lungs (the bronchi), which become infected and filled with pus due to abnormalities of the hair-like structure (cilia) of the cells lining the respiratory tract. This can lead to repeated infections and damage the lung, especially if diagnosis is delayed. About 1 in 100,000 of the population has PCD, which equates to about 560 patients in England.	
NHS Centres	Leeds Teaching Hospitals NHS Trust; Royal Brompton & Harefield NHS Foundation Trust; University Hospital Southampton NHS Foundation Trust; University Hospitals of Leicester NHS Trust
Expenditure	More than £1m but less than £5m (for management and diagnostic elements)
Caseload	501
Outcomes collated for 2016/17	Not available for 2016/17
Geographical equity access	Data not available or not comparable

Primary malignant bone tumours service (adults and adolescents)	
This service provides diagnosis and surgery for primary malignant bone cancers. Examples of conditions include osteosarcoma, chondrosarcoma and Ewing's sarcoma. The key aim is to avoid amputation if possible while ensuring complete removal of the cancer.	
NHS Centres	Oxford University Hospitals NHS Trust; Royal National Orthopaedic Hospital NHS Trust; The Newcastle Upon Tyne Hospitals NHS Foundation Trust; The Robert Jones and Agnes Hunt Orthopaedic Hospital NHS Foundation Trust; The Royal Orthopaedic Hospital NHS Foundation Trust
Expenditure	More than £10 million but less than £20m
Number of confirmed cases	552
Geographical equity access	Data not available or not comparable

Proton beam therapy overseas service (adults and children)	
Proton Beam Therapy (PBT) provides radiation by delivering a beam of proton particles, rather than X-Rays. The physical properties of protons results in almost no radiation dose being deposited in the normal tissue beyond the tumour. This is in contrast to X-rays where there is dose extension beyond the tumour.	
NHS Centres	Three overseas providers commissioned by NHS England
Expenditure	More than £20 million but less than £30m
Patients received proton beam therapy	189
Geographical equity access	No evidence of geographical inequity

In May 2016, Quality Assurance visits were undertaken to the Florida Health Proton Therapy Institute and ProCure Proton Therapy Institute (Oklahoma) by a team including clinical oncologists and NHS managers. Both centres were found to be providing a high quality service and feedback from NHS patients at both centres was positive.

NHS England published its clinical commissioning policy for proton beam therapy for cancer of the prostate. Following a careful review of the evidence, NHS England concluded there was not enough evidence to make the treatment available at this time.

Development of the NHS PBT service continues and the first NHS centre, at The Christie Hospital, Manchester is on course to open in 2018. Complications to the excavation of the London site, means the centre at University College London Hospitals will now open in 2020.

Pseudomyxoma peritonei service (adults)	
Pseudomyxoma peritonei (PMP) is a rare, mucus-producing tumour, which spreads to compress the abdominal organs. PMP usually arises from a ruptured tumour of the appendix. The condition is of borderline malignancy in that it does not metastasise by the blood stream or through lymphatic spread in the early stages. The tumour spreads locally within the peritoneal cavity and eventually compresses the abdominal organs. The disease is slow growing and is considered a relatively benign condition. However, without specialist cancer treatment, the majority of patients die either from complications of repeated surgery or from compression of the small bowel with resulting malnutrition. The symptoms of PMP are varied with most patients complaining of gradual abdominal swelling over a period of time affecting their ability to eat normally. About 100 new patients present each year in England with PMP.	
NHS Centres	Hampshire Hospitals NHS Foundation Trust; The Christie NHS Foundation Trust
Expenditure	More than £10 million but less than £20m
Major full cytoreduction	172
Outcomes collated for 2016/17	5 year survival among those who have complete cytoreduction exceeds 80%
Geographical equity access	Data not available or not comparable

Pulmonary hypertension service for children	
Paediatric pulmonary hypertension is a high pressure in the circulation of blood through the lungs, leading to progressive heart failure. The prognosis has improved with recently developed drugs. Some patients also need a lung (or heart and lung) transplant. The service cares for about 500 children with PH.	
NHS Centre	Great Ormond Street Hospital for Children NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Caseload	515
Geographical equity access	No evidence of geographical inequity

Pulmonary thromboendarterectomy service (adults and adolescents)	
Pulmonary thromboendarterectomy (PTE) is complex surgery to remove blood clots and related material from the pulmonary artery of people with chronic pulmonary thrombo-embolic disease (repeated episodes of blood clots travelling to the lung) that may cause life-threatening pulmonary hypertension (raised pressure in the artery that carries blood to the lung).	
NHS Centre	Papworth Hospital NHS Foundation Trust
Expenditure	More than £5m but less than £10m
Surgical operations	172
Outcomes collated for 2016/17	Only 7 in-hospital deaths from 192 total operations, including some for tumour and international patients (3.6%). 155 (>80%) discharged directly to home.
Geographical equity access	No evidence of geographical inequity

Chronic thromboembolic pulmonary hypertension is a rare condition in which, over time (chronic) blood clots (thrombi) form and are carried into (embolise) the arteries of the lung (pulmonary arteries), causing high blood pressure (hypertension) in these arteries. In a small proportion of patients, it is possible to remove the clot (thrombectomy). The operation is technically very demanding, and careful patient selection is required.

The service, which is based at Papworth hospital, treats about 170 patients each year and is now one of the most experienced centres in the world. Patients are typically aged about 60, but patients as young as 20 and as old as 88 were treated successfully last year. Ninety percent of patients are alive at three years after the operation, compared to 70% in comparable patients in whom the operation was not possible.

Rare mitochondrial disorders service (adults and children)	
Mitochondria are small organelles, present in every cell in the body – whose function is to process the cell's energy. They contain their own genetic complement, the mitochondrial genome and their principal task is to provide the energy necessary for normal cell functioning and maintenance. Disruption of this energy supply can have devastating effects for the cell, organ and individual. One important consequence of mitochondrial involvement in all cell types is that mitochondrial disease can affect virtually any organ and present with a plethora of symptoms and signs to a variety of specialties. These genuinely multi-system diseases are associated with significant morbidity and mortality.	
NHS Centres	Oxford University Hospitals NHS Trust; The Newcastle Upon Tyne Hospitals NHS Foundation Trust; University College London NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Outpatient referrals	368
Outcomes collated for 2016/17	100% of patients were given an alert card 100% of patients were given a definitive clinical or genetic diagnosis.
Geographical equity access	Data not available or not comparable

Failure of mitochondria can produce symptoms in almost any body system, including fits and brain damage, heart failure, liver failure and paralyses. This wide range of signs and symptoms can make diagnosis of the rarer forms of mitochondrial disease very difficult.

The rare mitochondrial disease service, based in London, Newcastle and Oxford, provides an accurate molecular diagnosis and advice on managing the condition, though care remains with the local hospital.

The mitochondria carry their own DNA, but some structures in the mitochondrion are coded by DNA in the main nucleus of the cell. Some mitochondrial disease are caused by mutations in the mitochondrial DNA; others by mutations in the main DNA of the nucleus. The distinction is important for genetic counselling and more recently for treatment.

Reconstructive surgery service for adolescents with congenital malformation of the female genital tract	
This service helps young women by providing assessment, inpatient care (including dilation therapy or surgical reconstruction), expert psychology input, outpatient support and follow up care after reconstructive surgery of female genital tract. Conditions include: • Congenital absence of the vagina and/or associated gynaecological structures • Gynaecological outflow tract obstruction with primary amenorrhoea with cyclical abdominal pain and pelvis mass • Occluded hemi-vagina • Virilisation • Abnormal anatomy requiring surgical reconstruction	
NHS Centre	Imperial College Healthcare NHS Trust
Expenditure	Less than £0.5m
Number of procedures	Data suppressed to maintain patient confidentiality
Outcomes collated for 2016/17	Data suppressed to maintain patient confidentiality
Geographical equity access	Numbers too small to analyse

Retinoblastoma service (children)	
Retinoblastoma is a malignant tumour of the retina and usually presents in children under the age of two. It is an aggressive eye cancer which can result in the loss of vision and in extreme cases, death. About 50 children are diagnosed with retinoblastoma each year.	
NHS Centres	Barts Health NHS Trust; Birmingham Children's Hospital NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Confirmed patients	46
Outcomes collated for 2016/17	At the Royal London, from 2012 to16, there were 75 unilateral and 45 bilateral patients. The service aims to treat the tumour while preserving the eye. Enucleation (removal of the eye) was required for 61% of unilateral and 33% of bilateral. 2.5% of patients died.
Geographical equity access	Data required from cancer register

Retinoblastoma is genetic which means that every cell in both eyes carries the cancer-causing mutation; so the cancer may affect both eyes, and it may recur. It may run in families. Long-term follow-up and surveillance is an important part of care.

The service, based in Birmingham and London, treats retinoblastoma with a combination of surgery and chemotherapy, together with genetic analysis. The aim of treatment is to cure the cancer but preserve the eye whenever possible. Great surgical expertise is required to remove a tumour from the eye of a baby, as well as considerable judgement as to whether the operation is feasible. The oncology is also highly specialised, with chemotherapy often delivered directly into the artery which supplies the eye so as to avoid toxicity elsewhere in the body. In general the outlook for patients is good and death from retinoblastoma is very rare.

Severe acute porphyria (adults and children)	
Acute porphyrias are a rare, inherited disorder, typically presenting in young adults. Acute attacks can be life-threatening. The condition can result in permanent disability and even death due to progressive motor neuropathy.	
NHS Centres	King's College Hospital NHS Foundation Trust; University Hospital of Wales
Expenditure	More than £1m but less than £5m
Active caseload	137
Outcomes collated for 2016/17	Of 126 patients in the service, 25 receive regular haem arginate. There have been 12 admissions, one for 8 weeks. Five patients had more than 4 attacks.
Geographical equity access	Numbers too small to analyse

Severe combined immune deficiency and related disorders service (children)	
Severe combined immunodeficiency disorders (SCID) is the term used to cover the most serious types of primary immunodeficiency where various components of the body's defence system are defective, leaving the child prone to unusual and/or frequent infections. In all forms of SCID, both T and B lymphocyte functions, the body's defence mechanisms, are defective from birth.	
NHS Centres	Great Ormond Street Hospital for Children NHS Foundation Trust; The Newcastle Upon Tyne Hospitals NHS Foundation Trust
Expenditure	More than £10 million but less than £20m
Number of transplants	56
Outcomes collated for 2016/17	12-month survival post HSCT(or gene therapy) in 88.4% of patients
Geographical equity access	No evidence of geographical inequity

Small bowel transplantation service (adults)	
NHS Centres	Cambridge University Hospitals NHS Foundation Trust; Oxford University Hospitals NHS Trust
Expenditure	More than £1m but less than £5m
Number of transplants	Data suppressed to maintain patient confidentiality
Outcomes collated for 2016/17	Data suppressed to maintain patient confidentiality
Geographical equity access	Numbers too small to analyse

Small bowel transplantation service (children)	
This service provides assessment, transplantation and lifelong follow up of paediatric patients requiring small bowel transplantation.	
NHS Centres	Birmingham Children's Hospital NHS Foundation Trust; King's College Hospital NHS Foundation Trust
Expenditure	More than £1m but less than £5m
Number of transplants	Data suppressed to maintain patient confidentiality
Outcomes collated for 2016/17	Data suppressed to maintain patient confidentiality
Geographical equity access	Numbers too small to analyse

Specialist paediatric liver disease service	
This service provides a diagnostic, assessment and treatment service for paediatric liver disease. The major conditions covered by the service are: • Acute liver failure • Biliary atresia • Chronic liver disease • Hepatitis A, B and C • Metabolic liver disease • Neonatal hepatitis	
NHS Centres	Birmingham Children's Hospital NHS Foundation Trust; King's College Hospital NHS Foundation Trust; Leeds Teaching Hospitals NHS Trust
Expenditure	More than £10m but less than £20m
Inpatient episodes	1,075
Outcomes collated for 2016/17	Data suppressed to maintain patient confidentiality on the basis that the service covers a large number of diagnoses but there are only a small number of patients who have each of the conditions
Geographical equity access	Numbers too small to analyse (by individual condition)

Stickler syndrome diagnostic service (adults and children)	
Stickler syndrome is an inherited disorder of connective tissue associated with cleft palate, deafness and arthropathy. It is the commonest inherited cause of rhegmatogenous retinal detachment in children (where fluid passes into the space between the retina and the retinal pigment layer). Although the systemic features are widespread, the sight-threatening complications are generally the most serious, particularly the risk of giant retinal tear, which is frequently bilateral and, if untreated, can lead to blindness.	
NHS Centre	Cambridge University Hospitals NHS Foundation Trust
Expenditure	More than £0.5m but less than £1m
Index patients	73
Outcomes collated for 2016/17	Definitive diagnosis of Stickler syndrome was confirmed in 55% of patients referred.
Geographical equity access	No evidence of geographical inequity

Vein of Galen malformation service (adults and children)	
Vein of Galen Malformations (VGMs) are extremely rare abnormalities in the blood vessels in the brain. They usually occur in new-born children and often result in cardiac problems, although sometimes these problems do not occur until later in life.	
NHS Centres	Great Ormond Street Hospital for Children NHS Foundation Trust; NHS Greater Glasgow & Clyde (until July 2016) Alder Hey Children's Hospital (from March 2017)
Expenditure	Less than £0.5m
Number of procedures	29
Outcomes collated for 2016/17	Data suppressed to maintain patient confidentiality
Geographical equity access	Numbers too small to analyse

Ventricular Assist Devices (VADs) as a bridge to heart transplantation or myocardial recovery (all Ages)	
Ventricular assist devices (VADs) can be attached externally or implanted within the body to support the adult's failing heart until a donor heart becomes available for transplantation, a technique known as ‘bridge to transplant’. VADs work by supporting the pumping action of the left ventricle, which is the main pumping chamber of the heart. They sometimes also need to be implanted in the right ventricle. The implantation of a VAD is only considered in patients with advanced heart failure who are listed for a transplant and who are deemed to be deteriorating so rapidly that they would not survive long enough to receive a heart via the urgent allocation scheme. Occasionally, a VAD enables the heart to recover sufficiently for the device to be removed (‘bridge to recovery’). A small but increasing number of children requiring a heart transplant are supported with ventricular assist devices (VADs), mechanical devices that circulate blood outside the body to supports the failing heart. This is known as ‘bridge to transplant’ and supports the heart until a donor heart becomes available for transplantation. In 2015/16 there were 137 heart transplants in adults. There were 28 heart transplants in children.	
NHS Centres	Great Ormond Street Hospital for Children NHS Foundation Trust; Papworth Hospital NHS Foundation Trust; Royal Brompton & Harefield NHS Foundation Trust; The Newcastle Upon Tyne Hospitals NHS Foundation Trust; University Hospital of South Manchester NHS Foundation Trust; University Hospitals Birmingham NHS Foundation Trust
Expenditure	Figure included in heart and lung transplant
Number of procedures	152
Outcomes collated for 2016/17	5 year outcomes for patients given a long term VAD between 2006 and 2016 were: 48% died before transplant;30% were transplanted;14% were alive on a VAD and still waiting;8% were explanted (i.e. recovered)
Geographical equity access	Numbers too small to analyse

Wolfram syndrome service (adults and children)	
Wolfram syndrome is a very rare inherited disorder. It is a progressive neurodegenerative disorder with a debilitating and life threatening association of diabetes, blindness, deafness and brain disease.	
NHS Centres	Birmingham Children's Hospital NHS Foundation Trust; University Hospitals Birmingham NHS Foundation Trust
Expenditure	Less than £0.5m
Caseload	87
Geographical equity access	Numbers too small to analyse

Xeroderma pigmentosum service (adults and children)	
Xeroderma pigmentosum is a life-threatening inherited disorder affecting skin, eyes and nervous system. A defect in the process of repairing ultraviolet-induced DNA damage results in: severe sunburn-type reactions to daylight; skin cancers in exposed skin from early childhood; eye disease; and progressive neurological degeneration in 20-30% of patients. There are thought to be about 100 people with the condition in the UK.	
NHS Centres	Guy's and St Thomas' NHS Foundation Trust
Expenditure	More than £0.5m but less than £1m
Caseload	110
Outcomes collated for 2016/17	95% of children (77% of adults) have window film at home; 76% (97%) wear sunglasses when outside; the number of skin cancers diagnosed has increased due to earlier and more accurate diagnosis.
Geographical equity access	No evidence of geographical inequity

When sunlight strikes the skin, its ultraviolet rays cause damage to the DNA in skin cells. Normally there is a mechanism to repair this damage, but in people with Xeroderma pigmentosum (XP) this mechanism is defective. In consequence the skin is prone to multiple cancers from childhood onward, and the ultraviolet radiation also damages the eyes. XP is also associated with cognitive problems and learning difficulties.

Although the cancers can be removed surgically if detected early enough, the sheer number and scale of the problem can eventually become overwhelming. Hence protecting the skin from sun damage is crucial, by clothes, sun cream and protective film on the windows of home, school and work.

The XP service based at St Thomas' hospital in London provides accurate diagnosis and lifetime care for patients, offering advice on prevention (and equipment such as UV meters), surveillance of the skin to detect cancers early and care for the eye and cognitive problems. Specialist nurses play a crucial role in visiting homes and schools to offer practical advice. The result is that over 94% of children have window protection at home and at school, and 88% in the family car. Unsurprisingly the percentages are lower in adults

The service is also research active and has won a large research grant to investigate in detail why it is that patients often fail to comply with life-saving recommendations from the care team.

The UK National XP service is the only service worldwide that offers formal and specialist long term follow up of XP patients. Recent work by the clinical team has led to the deep (detailed description) of patients by the services across the eight complementation groups, resulting in a number of very important observations. This has led to the design of disease severity scores for photo protection, ophthalmology, neurology and sunburn. Documentation of these scores will enable monitoring of patient under long-term follow-up and will allow more accurate assessment of disease progression across the different complementation groups. This work has been published in the Proceedings of the National Academy of Sciences https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4780618/

International cooperation and collaboration is important to this rare condition. The service has worked to establish the European XP Society (EXPS) with members from the UK, Germany, Spain, Italy and France. The first meeting was held at the EADV in Copenhagen in late 2015. Colleagues with an interest in XP from South Africa and Saudi Arabia also attended and joined the Society. It was agreed that a set of clinical standards and management guidelines should be created, using the EXPS as the vehicle, in order to improve patient care worldwide. The UK team will lead on this and use future annual EXPS meeting as the forum for discussion.

The service has ongoing close links to colleagues at the National Institute of Health in the US, about various aspects of XP and related disorders. The last International Symposium, for clinicians and scientists, looking after patients with DNA repair disorders was held in Kobe, Japan in 2014. The UK XP team hopes to arrange the next meeting in 2018 in London.

Appendix A: New Highly Specialised Services commissioned during 2016/17

1. Hand and upper limb reconstruction

The Hand and Upper Limb Transplant service was commissioned as a national service in 2016/17. The service provides hand and upper limb transplantation to reconstruct an absent hand or upper limb, lost as result of trauma or infection. Hand and upper limb transplantation would, ordinarily, only be offered to those for whom current reconstructive techniques are unsuitable or for those in whom prosthesis have been considered unsatisfactory. The overall aim of the service is to improve functional capacity and quality of life for the patient. The service completes detailed psychological, surgical, immunological and medical screening as well as occupational therapy assessment prior to patients being placed on the waiting list where they await a suitably matched organ donor.

The service, which is provided by a highly specialised clinical team at Leeds Teaching Hospitals NHS Trust, works closely with colleagues at NHS Blood and Transplant, who are the organisation responsible for organ donation.

It is anticipated that the number of patients seeking this procedure, who are clinically suitable and have a matched organ donor, may be in the region of five per year.

2. Atypical haemolytic uraemic syndrome (aHUS)

Atypical haemolytic uraemic syndrome (aHUS) is a chronic, rare, progressive condition that causes severe inflammation of blood vessels and the formation of blood clots in small blood vessels throughout the body, a process known as systemic thrombotic microangiopathy. aHUS can occur at any age. Onset occurs in childhood slightly more frequently than in adulthood. Most children who develop aHUS will experience the disease for the first time before the age of two years. The true incidence and prevalence of aHUS in England is uncertain because some patients

remain undiagnosed. Worldwide, the prevalence of aHUS ranges from 2.7–5.5 per million population, with an incidence of about 0.40 per million population.

The national aHUS service was commissioned by NHS England in 2016/17 and provides a national diagnostic and management advice for patients with aHUS. The service offers comprehensive diagnostic clinical and pathological investigations and expert opinion, facilitating optimal patient management on a shared-care basis with referring clinicians and other specialist services.

The service also has an important role in oversight of the use of the drug eculizumab for patients with a confirmed diagnosis of aHUS by initiating prescriptions directly for local patients, or by authorising the use of eculizumab in patients at remote centres under shared care arrangements. The service monitors the ongoing use of eculizumab and the surveillance of patients in whom the drug has been withdrawn because disease activity has reduced. No patient has died of aHUS in England in 2016/17.

3. Mitochondrial donation service

NHS England is making up to £8m available over five years to fund the treatment costs of a world-leading five year evaluation of mitochondrial donation – a form of IVF in which the future baby's mitochondrial DNA comes from a donor egg, to avoid passing on inherited mitochondrial diseases.

Mitochondrial diseases can be devastating, causing blindness, blocked heart, muscle wastage and weakness, learning disabilities, deafness and diabetes. An estimated 2,473 women of child bearing age are at risk of transmitting mitochondrial DNA disease and the lifetime treatment cost for a patient with serious mitochondrial disease is around £1.3m.

Wellcome is funding a five year research programme at Newcastle University and The Newcastle upon Tyne Hospitals NHS Foundation Trust to study the long term follow up of any children born following mitochondrial donation, and NHS England will fund the treatment costs. A total of 125 patients will be enrolled, with an estimated 25

progressing to treatment annually. The first patient is expected to be treated in autumn 2017.

Appendix B: UK-wide Commissioning Arrangements of Highly Specialised Services during 2016/17

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

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

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

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

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