2013/14 NHS STANDARD CONTRACT
FOR MEDICAL GENETICS (ALL AGES)

PARTICULARS, SCHEDULE 2- THE SERVICES –A. SERVICE SPECIFICATIONS

Service Specification No.	E01/S/a
Service	Medical Genetics (All Ages)
Commissioner Lead	
Provider Lead	
Period	12 months
Date of Review	

1. Population Needs

Medical Genetics can be divided into two main service areas, covering both children and adults: Clinical Genetics and Laboratory Genetics (consisting of molecular and cytogenetic tests and, in some specified centres, specialised biochemistry tests or other specialist tests). In summary, in Clinical Genetics Departments, clinical staff will see patients affected by, or at risk of, a genetic condition and will provide diagnostic and genetic counselling services. The Laboratory Genetics services will provide either or both molecular and cytogenetic testing, together with advice and support to referrers and interpretation of results.

Under this service specification, the laboratory services will only provide the genetic tests for patients referred by the Clinical Genetics service. (They will have other contractual arrangements for other referrals which they receive).

1.1 National/local context and evidence base

Diseases with a genetic component are estimated to affect at least 5-6% of the population. Often they are rare diseases, those defined as affecting fewer than 5 in 10,000 of the general population. Over 5,000 rare diseases have been identified and around 80% of rare diseases are genetic. These diseases can be hard to diagnose and patients can face delays in diagnosis. They also affect several generations of the same family and clinical genetics services look beyond individual patients to include relatives who are also at risk.

Genetic disorders can affect any body organ or system and include:

- **Chromosomal anomalies**, including deletions/duplication or balanced
- /complex rearrangements
- **Single gene disorders**, e.g. muscular dystrophies, dysmorphic syndromes, inherited cardiac conditions, skeletal and connective tissue disorders and neurological conditions, across all ages
- **Familial cancer syndromes**
- **Congenital anomalies**, including non-genetic and teratogenic anomalies
- **Learning disability** with or without autism/congenital anomalies
- **Common adult disorders with a single gene aetiology**

There are 23 Regional Genetic Centres (RGCs) in the UK, including 17 in England, all with strong links to genetics laboratories, mainstream medical specialties and their clinical networks. The services are consultant led and regionally-based, serving populations ranging from ~1 to ~5 million and are delivered in a 'hub and spoke' model.

In terms of laboratory services, there are many advances in genetics and genomic medicine, with much national and international research focussed on this field. There is the potential for this to speed diagnosis and change pathways of care; the translation of this research into practice is challenging.

Evidence base:

- Department of Health 2003 'Our Inheritance, Our Future – Realising the Potential of Genetics in the NHS'
- Annual Report of the Chief Medical Officer 2009, pp.38-45 Department of Health - www.dh.gov.uk
- House of Lords – House of Lords Scientific and Technology Committee: Genomic Medicine. London 2009 http://www.publications.parliament.uk/pa/ld200809/ldsctech/107/107i.pdf
- NICE guidelines: Familial Breast Cancer, 2006. NICE clinical guideline 41 (partial update) NICE – www.nice.org.uk
- 'Improving Lives, optimising resources – a vision for the UK Rare Disease Strategy', 2011-Rare Disease UK – www.raredisease.org.uk
- 'Building on our inheritance' Department of Health - Human Genome Strategy Group, January 2012
- Outcome measurement in Clinical Genetic Services: A Systematic Review of Validated Measures was undertaken by the North West Genetics Knowledge Park (Nowgen) and published by the International Society for Pharmacoeconomics and Outcomes Research (ISPOR) in 2007. Supplementary material can be found at: http://www.ispor.org/valueinhealth_index.asp.
- Professional roles in the multidisciplinary team in Genetics, 2006, and Genetic Testing of Children 2010, British Society for Human Genetics - www.bshg.org.uk
- Roles of the Clinical Geneticist, 2011 and Clinical standards for a Genetics unit,

2005 Clinical Genetics Society – www.cgs.org.uk

- Standards for Medical Laboratories, November 2010 Clinical Pathology Accreditation – www.cpa-uk.co.uk

Best Practice Guidelines - Clinical Molecular Genetics Society – www.cmgs.org, as listed below:

Disease Guidelines:			
Title	**Copyright**	**Publication**	**Status**
Cystic Fibrosis	2009	Clinical Molecular Genetics Society (CMGS) e-publication	Approved
Duchenne and Becker muscular dystrophies	2010	Neuromuscular disorders	Endorsed by CMGS
FAP (familial adenomatous polyposis) & MAP (MYH associated polyposis)	2011	CMGS e-publication	Approved
Fragile X syndrome	2005, CMGS	CMGS e-publication	Approved
Haemochromatosis	2006	BMC Med Genet 2006 Nov 29;7:81	Approved
Haemophilia A	2011, CMGS	CMGS e-publication	Approved
Haemophilia B	2011, CMGS	CMGS e-publication	Approved
Mitochondri disease Newcastle referral form Oxford referral form	2008, CMGS	CMGS e-publication	Approved
Huntington disease	2010	CMGS e-publication	Draft

Prader-Willi and Angelman syndromes	2010	Biomedical Central Medical Genetics	Approved
Von Willebrand disease	2008, CMGS/UK Haemophilia Centre Doctors' Organisation (UKHCDO)	CMGS e-publication	Approved

Additional Disease-specific information:

Title	Copyright	Publication	Status
HNPCC (hereditary non-polyposis colorectal cancer) - Immunohistochemical analysis interpretation	2008, UK National External Quality Assessment Services (UKNEQAS) immunocytochemistry (ICC) & in situ hybridisation (ISH)	UKNEQAS	Approved

General Guidelines:

Title	Copyright	Publication	Form
Denaturing High Performance Liquid Chromatography (DHPLC)	2004, CMGS	CMGS e-publication	Approved
Internal Quality	2004, CMGS	CMGS e-publication	Approved
Internal Quality – European Molecular Genetics	2002, EMQN	EMQN e-publication	Approved
Quality Network (EMQN)	2007, CMGS	CMGS e-publication	Approved
Maternal cell contamination (MCC)	2012, Quantitative Fluorescent Polymerase Chain Reaction (QFPCR) CMGS/ Association for Clinical Cytogenetics (ACC)	CMGS e-publication	Approved
Reporting Sequencing	2011, CMGS	CMGS e-publication	Approved

Unclassified variants, (Web Form 2)	2009, CMGS	CMGS e-publication	Approved
Resources, Evaluation Form 1,	2007, CMGS	CMGS e-publication	Approved publication

Association for Clinical Cytogenetics (ACC) – www.cytogenetics.org.uk

- Professional guidelines for clinical cytogenetics, 2007
  and related guidelines below:

ACC Best Practice Guidelines

- Acute myeloid leukaemia and myelodysplasia (Mar 2012) NEW
- Quantitative Fluorescent Polymerase Chain Reaction (QF-PCR) for the Diagnosis of Aneuploidy (Jan 2012)
- Constitutional Postnatal Chromosomal Microarra (Dec 2011)
- Acute lymphoblastic leukaemia (Jul 2011)
- Chronic myeloid leukaemia and other myeloproliferative neoplasms (Jul 2011)
- Solid Tissue (Nov 2010)
- Prenatal diagnosis (Dec 2009)
- Clinical Cytogenetics (Mar 2007)
- Haemato-Oncology (Mar 2007)
- Postnatal Samples (Mar 2007)
- European Molecular Genetics Quality Network - www.emqn.org
- **Duchenne and Becker Muscular Dystrophies**
  Download (198 Kb)
  v 2010, e-publication, Journal (NMD, Volume 20, Issue 6, Pages 422-427 (June 2010) - Approved.
- **Fragile-X Syndrome**
  Download (507 Kb)
  v 2006, e-publication - Approved.
- **Hereditary Breast and Ovarian Cancer**
  Download (176 Kb)
  v 2008, e-publication - Approved.
- **Hereditary Sensory and Motor Neuropathies**
  Download (238 Kb)
  v 2006,e-publication - Approved.
- **Maturity Onset Diabetes of the Young**
  Download (179 Kb)
  v 2008, e-publication, Journal (Diabetologia, Volume 51, Number 4 / April, 2008) - Approved.
- **Myotonic Dystrophy**
  Download (395 Kb)
  v 2010, e-publication, Journal - Approved.

- **Prader-Willi / Angelman Syndromes**
  v 2010, e-publication, Journal (BMC Medical Genetics 2010, 11:7) - Approved.
- **Spinocerebellar Ataxia's (2010)**
  Download (164 Kb)
  v 2010, e-publication, Journal (EJHG Feb 2010) - Approved.
- Genetic Counsellor Registration Board – www.gcrb.org.uk
- The UK Genetic Testing Network (UK GTN) provides information relevant to genetic diagnosis for rare conditions. It sets quality standards for genetic testing and provides a Directory of Genetic Tests that are of proven clinical validity and utility, which have been recommended for NHS service – www.ukgtn.nhs.uk

2. Scope

2.1 Aims and objectives of service

The aim of Medical Genetic services (clinical and laboratory) is to provide a patient centred, specialised service focussed on the provision of diagnosis and advice to promote improved clinical management and quality of life for those affected by or at risk of a genetic condition or congenital abnormality.

- Clinical Genetics provides services for any individual or family affected by, or at risk of, a genetic disorder or congenital abnormality. Individuals and families are helped to understand their condition, its implications, and their options with regard to reproduction, screening, prevention and management.
- In clinical genetics, the fundamental unit of responsibility is the 'family' and includes not only the affected individual who presents for diagnosis and treatment, but also relatives who are identified as being at risk. For example, whilst an individual who presents with ill health needs to be diagnosed and treated within a traditional NHS model, the awareness of family and the relationships within it ensures that the 'at risk but well' relatives can be managed appropriately, offering the opportunity for predictive and carrier testing, screening, early intervention and prenatal/preconception genetic counselling.
- The principal objective of the clinical genetics service is to provide integrated clinical and laboratory genetic services that are equitable, safe, efficient, appropriate, accessible and acceptable to all sectors of the community and of a demonstrably high quality.
- Medical Genetics services will provide the following:
    - Diagnosis or exclusion of inherited disease and genetic disorders affecting all ages
    - Targeted investigations, using both specialised genetic and conventional non-genetic tests, for the purposes of diagnosis, risk assessment and management
    - Diagnosis and early management of inherited diseases to reduce the morbidity and/or mortality for many of these clinical conditions in those affected; and/or provide relatives with risk information and possible interventions to reduce their risk of poor clinical outcome

- Communication of the natural history, complications and appropriate management of inherited disease, to the patient, relatives and relevant professionals
- Genetic counselling and attention to psychosocial aspects of inherited conditions
- Genetic counselling and effective communication of appropriate genetic information in the clinic and by a variety of means (e.g. letters, telephone, information leaflets)
- Predictive genetic testing of at-risk relatives for conditions where a familial mutation or cause has been identified, using agreed protocols where available
- Explanation of the reproductive options available, when appropriate, to women/couples who might wish to receive an early diagnosis or reduce their risk of having an affected child
- Where appropriate, follow-up, support and coordination of health surveillance / screening for specific genetic conditions. Identification of genetic risk to the wider family and, where appropriate, the offer of genetic services to extended family members
- Formal training programmes and ongoing professional development for clinical geneticists, genetic counsellors and healthcare scientists/practitioners
- Training and education for other healthcare professionals
- Laboratory investigations in support of clinical genetics services including discussion and interpretation of complex results and data of uncertain significance. Laboratories liaise with other genetics laboratories in local and national networks
- Participation in local and national clinical networks, e.g. oncology, neurology, fetal medicine, cardiology; national dysmorphology, Cancer Genetics Group meetings, UK Huntington Disease Consortium and Genethics meeting to inform best practice
- Provision of expertise and information for other secondary and primary care staff and other health professionals, including interpretation of laboratory reports conveying complex genetic results and data, both pathogenic mutation and those of uncertain significance
- A service that meets the needs of patients and their families, as monitored through validated patient satisfaction surveys

2.2 Service description/care pathway

Clinical Genetics

Clinical Genetics Departments will provide diagnostic and genetic counselling services, and in some multi-system disorders, co-ordination of management and follow up for individuals and families with, or at risk of, conditions which have, or may have, a genetic basis. Individuals and families should be helped to understand their condition, its implications, and their options with regard to reproduction, screening,

preventions and management. The provision of advice on clinical management of patients with a genetic condition, and their wider family, is a key component of the role of Clinical Geneticists.

All patients seen by a Clinical Geneticist/Genetic Counsellor will fall within the NHS England Clinical Genetics contract. Clinical Genetic services will be predominantly outpatient based.

Each service will include:

- Diagnosis or exclusion of genetic disorders and congenital malformations
- Investigation and genetic risk assessment
- Provision of information
- Predictive genetic testing
- Discussion of reproductive options
- Initiation and coordination of health surveillance and screening for genetic conditions
- Co-ordination of interventional management in specialist or multi- disciplinary clinics
- Management of the extended family
- Maintenance of genetic family disease specific records
- Liaison with genetic laboratories
- Participation in local and national genetic networks
- Education and training of genetic and other healthcare professionals
- Acting as an expert resource to all health professionals
- Audit of clinical services
- Research – clinical, biomedical, psychosocial and service related

(Developed from 'Roles of the Clinical Geneticist' 2011, Clinical Genetics Society)

Service outputs include:

- Clinical and genetic diagnosis, explanation and information about the disease, syndrome or condition, determination and communication of genetic/recurrence risk, identification of screening and/or intervention options and appropriate counselling support
- Service planning for integrating molecular testing in pathology and clinical genetics into mainstream specialities
- The provision of expert information and educational resources to healthcare disciplines
- Contribution to research through clinical and laboratory projects and recruitment to national studies and therapeutic trials
- Liaison with colleagues: Clinical Geneticists and genetic counsellors support colleagues across the medical specialties in order to ensure that the potential benefits and limitations of genetic testing are understood, made available to patients in all areas of medicine (referred to as 'mainstreaming'), and ethically applied. The clinical genetics services often provide an important 'gate-keeping'

function with respect to optimising the utility of genetic tests. Examples of effective working arrangements include clinical networks, multidisciplinary teams, joint clinics and GPs with a Special Interest working within Regional Genetic Centres. The liaison function includes ward consults and ward rounds

For some multi-system disorders with a genetic basis, Clinical Geneticists will provide on-going co-ordination of their management and follow up, linking with other specialties as appropriate. The majority of patients will either be discharged on receipt of diagnosis /advice or discharged back to the care of their referring clinician for ongoing management of their condition.

A very limited number of Clinical Genetic services will provide a specialised Pre-implantation Genetic Diagnosis service in conjunction with specialised fertility services. A commissioning policy for these services is in place.

Services provided by specialised genetic laboratories (molecular, cytogenetic and, in some regional centres, specialised biochemistry)

Clinical Genetics services are aligned with specialist genetic laboratories within Regional Genetic Centres (RGC). The specialist laboratories are usually managed either by being part of an integrated service with clinical genetics, or within pathology services; although it is possible for the linked Clinical Genetics and Laboratory Genetics services to be in different organisations, each of which will have a separate contract for their part of the service.

Genetics laboratories must hold Clinical Pathology Accreditation (CPA) and should provide accurate test results in accordance with nationally agreed target reporting times appropriate for the nature of the sample and referral reason.

- Molecular genetics: 3 days, 10 days, 20 days, 40 days, 80 days (all working days) using a RAG rating where Red = <80%, Amber = 80- 89.9% and Green = ≥90% reports issued within these times.
- Cytogenetics: 3 days, 10 days, 14 days, 21 days, 28 days (all calendar days with exception of 3 days which is measured in working days)using a RAG rating where Red =<80%, Amber= 80-89.9% and green =≥90% reports issued within these times
- Specialised biochemistry: no nationally agreed reporting times currently exist.

Only tests requested by Clinical Geneticists and Genetic Counsellors will form part of the NHS England Genetics contract. Genetic tests requested by clinicians from other specialties should be charged to the requesting clinician/ the organisation for which they work.

In NHS England contracts, services provided by specialised genetic laboratories for nationally defined specialised services may include:

- Diagnostic testing for children with learning difficulty, dysmorphism, developmental anomalies or symptoms indicative of a constitutional or acquired

chromosomal anomalies or singe gene disorder.

- Diagnostic testing in children or adults showing signs and symptoms that may be indicative of a genetic cause.
- Germ line mutation identification in both common (e.g. breast, bowel) and rare cancers and some common adult disorders, for example cardiac, endocrine, renal and neurological conditions where there is a significant family history in addition to symptoms in the index case. This is with the intention of offering confirmatory testing and pre-symptomatic testing in relatives of index cases.
- Pre-symptomatic testing in adults at high risk of some late onset inherited conditions including Huntington disease and familial cancers.
- Specialised services for children at high risk of a condition where management intervention is required in childhood, e.g. retinoblastoma or Multiple Endocrine Neoplasia (MEN) and related cancer syndromes.
- Definitive prenatal testing for pregnant women in whom screening identifies an increased risk of constitutional chromosomal imbalances and abnormalities e.g. Downs syndrome and related aneuploidies.
- Prenatal testing for single gene disorders e.g. cystic fibrosis and Duchenne muscular dystrophy.
- Testing for chromosomal imbalances and other genetic abnormalities indicated by reproductive history e.g. cases of recurrent miscarriages and male infertility.
- Carrier testing in adults at risk of an adverse reproductive outcome from balanced chromosomal conditions and some single gene conditions e.g. congenital adrenal hyperplasia.
- Testing to confirm an abnormal result from a population screening programme (funding of confirmatory tests is included as part of the diagnosis/treatment of the index patient) Services should ensure that there are links in place with population screening programmes.
- Long term banking of cells, preserved material and DNA (under the terms of the Human Tissue Act) to facilitate the long term commitment of medical genetic services to validate, provide and quality assure diagnostic and counselling services to families and future generations at risk of inherited disorders and to contribute to medical research under ethical committee approval.
- Cytogenetic or molecular tests on single cells biopsied from an early embryo as part of pre-implantation genetic diagnosis.
- Molecular Pathology tests to inform genetic testing (e.g. Immunohistochemistry and microsatellite instability).

(The details for each service relevant to each area listed above will be identified in the individual contracts. Other clinicians may also request these tests, but only referrals from Clinical Geneticists/ appropriately registered Genetic Counsellors apply for this specification.)

Laboratories will also:

- Advise and support referrers on appropriate tests to carry out
- Provide interpretation of results
- Determine the most clinically and cost effective means of carrying out testing (e.g. panels/sequential tests)

- Facilitate the integration of molecular testing in pathology and genetic testing in mainstream specialities
- Contribute clinically significant genetic variant information to appropriate
- data repositories
- Participate in local and national genetic networks
- Educate and train genetic and other healthcare professionals
- Act as an expert resource to all health professionals
- Carry out audits of laboratory services
- Carry out research - clinical, biomedical, and service related.

Genetic laboratories provide certain genetics tests, including, but not limited to those approved by the UKGTN. Tests not approved by the UKGTN are excluded from this specification, with the exception of rare tests accessed from abroad which are not available in the UK. The UKGTN Directory includes validated molecular and cytogenetic tests (including agreed 'grandfather' tests developed prior to the establishment of the UKGTN).

It is not anticipated that every laboratory will provide every test and laboratories must have appropriate systems in place for transporting specimens to other laboratories (or advising referrers of referral routes for specialised tests) and receiving results back safely and relaying these to the requesting clinician.

It is recognised that genetic laboratories will also provide other tests, outside of the NHS England contracts for example:

- During development of relevant tests prior to approval by the UK GTN
- From Clinical Commissioning Groups (CCGs) and non-clinical genetics referrers both within and outside the NHS

Requests from clinical geneticists for tests not (yet) approved by the UK GTN, may also be funded, although not from resources provided through NHS England and its contract. For example, where the science has moved on faster than the approval processes/ commissioning round:

- It is recognised that advances in genomic medicine and the supporting technologies are leading to the identification of new genetic conditions and new methods of diagnosis. Tests developed in the laboratories using new technologies need to be validated for clinical diagnostic use (e.g. panel tests that use Next Generation Sequencing (NGS).
- All laboratories should participate in the UK GTN audits of laboratory activity.

Paediatrics

When treating children, both the clinical and laboratory services will additionally follow the standards and criteria outlined in the Specification for Children's Services (attached as Annex 1 to this specification).

Service Model

The patient contact will be primarily outpatient-based, with clinics provided across a wide area geographically. NHS England expects to commission all activity within Clinical Genetics services from the Clinical Genetics Centres, including outreach clinics (rather than purchasing the latter from the trusts where they take place).

Background information should be gathered prior to a patient being seen and, indeed, this can help to determine whether an appointment is required. Background information may include a detailed family history, confirmation of diagnoses (e.g. from cancer registry), review of medical records, psychosocial circumstances and, in some cases, preliminary genetic tests e.g. array-based comparative genomic hybridization (aCGH), fragile (X).

A proportion of the referrals should be managed by Genetic Counsellors under varying levels of supervision depending upon competency, experience and professional registration.

Services will provide both general and specialist clinics, e.g. paediatric, cancer, neurogenetics, prenatal and cardiac as well as some combined clinics e.g. ophthalmology, skeletal dysplasia, cardiac, dermatology, fetal medicine. (In some circumstances it is beneficial for patients thought to have a genetic condition to attend a single multidisciplinary clinic where medical/surgical management can be discussed and clinical genetics expertise is available. (The funding for the joint clinic will come via whichever service the clinic is recorded under.)

Outpatient appointments should be of sufficient length to ensure adequate time to provide information and counselling and to enable the patient/relatives to discuss issues fully.

Clinical genetic services requesting tests from laboratories should monitor requests for compliance with referral criteria stipulated by the UKGTN (included in the UKGTN NHS Directory of Genetic Testing, as this promotes efficient use of resources).

Inpatient work is usually urgent and includes ward consultations and prenatal advice in fetal medicine units. All Regional Genetics centres will offer an on- call service for urgent advice (e.g. discussion about an abnormal prenatal result), which is available to clinicians and patients across their region. They should also offer ward consults (including neonatal and intensive care units) and rapid access clinics for urgent prenatal queries in their host trusts and where possible, or volume requires, in linked district general hospitals (DGHs) too, depending on location.

Familial Cancer Genetics Services

There will be a lead consultant for familial cancer genetic services in each Clinical Genetics Centre, whose role will include the development and co- ordination of management policies, referral guidelines and education programmes by linking with the genetics centres and units of the cancer network and primary care staff as appropriate.

The Regional Clinical Genetics Centres will provide cancer genetic outpatient services at the centre and in peripheral clinics in a similar fashion to clinical genetics services. Cancer genetic services will:

- Be led by a consultant specialising in cancer genetics
- Provide a service for a broad range of familial cancers including, but not limited to, breast/ovarian, colorectal, neurofibromatosis type 1, Von Hippel-Lindau syndrome and multiple endocrine neoplasia
- Provide written patient information including specific disease/test leaflets
- Provide advice about surveillance/preventative surgery to patients and their health professionals
- Provide referral guidelines for clinicians
- Advise clinicians in primary and secondary care about specific families, indicating whether or not genetic or other investigations and interventions are required
- Establish an agreed process with commissioners to discuss the prioritisation and implementation of significant new developments.
- Family based record systems

Each Genetics Centre should maintain a computerised family-based record system, incorporating disease-specific records.

Service user/carer information

- Patients/families must be provided with accurate, up-to-date information on genetic risks, testing and/or screening, and advised about reproductive choices that are available, with information discussed during face-to-face or telephone consultations summarised in writing afterwards.
- Clinicians should usually write to patients directly to explain complex terminology/concepts, or copy clinic letters to them and ensure effective patient involvement in all decision making.
- Consent for genetic testing and retention of deoxyribonucleic acid (DNA) samples must be undertaken in accordance with Department of Health guidance and the Mental Capacity Act, 2005.
- Education is provided at clinic, through post-clinic letters and in patient information leaflets (in different languages).
- Professional interpreting services, including signing interpreters, should be used if necessary.
- Patients and families should be directed to relevant lay support groups (including the Genetic Alliance UK and UNIQUE (Rare Chromosome Disorder Support Group).
- Services should recognise the role of the carer, which is vital for many patients, especially those with learning disability and neuromuscular disorders. Advocacy and support should be offered to carers appropriately.

Genetic Testing

The service model for Genetic testing is included within section 2.2.2 above

General Paediatric care

When treating children, the service will additionally follow the standards and criteria outlined in the Specification for Children's' Services (attached as Annex 1 to this Specification)

2.3 Population covered

The service outlined in this specification is for patients ordinarily resident in England*; or otherwise the commissioning responsibility of the NHS in England (as defined in

Who Pays?: Establishing the responsible commissioner and other Department of Health guidance relating to patients entitled to NHS care or exempt from charges).

* Note: for the purposes of commissioning health services, this EXCLUDES patients who, whilst resident in England, are registered with a GP practice in Wales, but INCLUDES patients resident in Wales who are registered with a GP practice in England.

Specifically, the service will cover any patient (adult or child) referred to a Clinical Geneticist / appropriately registered Genetic Counsellor who meets agreed referral pathways / standards and any subsequent genetic laboratory tests ordered by the Clinical Geneticist / Genetic Counsellor.

NHS England, by agreeing funding for genetic tests ordered by Clinical Geneticists/ Genetic Counsellors, will be agreeing this on a consistent national basis, reducing variation from previous commissioning arrangements. The UKGTN undertakes an annual review of genetic tests to monitor equity of access as requested by the Department of Health. This work is planned to inform the E-Atlas under development to provide national benchmarks.

Clinical Genetic Services will ensure that there is a reasonable spread and frequency of clinics across their region to ensure equity of geographic access, thus offering outreach clinics as well as clinics on their main hospital site.

2.4 Any acceptance and exclusion criteria

Referral processes and sources

Clinical Genetics referrals:

- Referrals will be accepted from primary, secondary and tertiary care.

- Referrals originate from all areas of medicine, most commonly general practice, paediatrics, obstetrics/antenatal screening services, oncology, neurology, surgery, cardiology, dermatology, nephrology, and audiology.
- Referrals may be urgent, e.g. neonatal or ward consultations, or for urgent prenatal advice and may be received from other Regional Genetics Centres (RGCs) if families are widely scattered. Additional family members for whom the discussion is of potential relevance may also attend appointments.
- On an exceptional basis, self referrals will be accepted, if there is clear evidence of a familial condition and a General Practitioner has been approached but declined to refer. Any such cases will be audited to ensure these criteria have been met – and are not expected to exceed 1% of referrals.

Criteria for a referral

- Detailed referral guidelines for clinical referrals and referral criteria for joint/multidisciplinary clinics should be provided on RGC websites.
- Clinical Genetics services should have robust referral criteria in place to ensure that they only see patients for whom they are likely to be able to offer advice.
- Criteria for consultant to consultant referrals to Clinical Genetics Services are under development.
- The Clinical Genetic Services will have a process in place to determine how referrals will be managed. These will describe who is the most appropriate person to manage a case (genetic counsellor or clinical geneticist) and will explain which patients need to be seen in a clinic.
- Genetic counsellors should have appropriate professional registration.

Laboratory service referrals

- Only requests from Clinical Geneticists/ appropriately registered Genetic Counsellors working for Regional Genetic Centres will fall within the NHS England contract. (Laboratories will require separate contracts with other provider organisations or CCGs for tests ordered by other clinicians.)

Criteria for a referral

- Clinical Genetics Services will refer patients for tests which they believe will contribute to the diagnosis or management of the patient's condition. Referrals should take account of the UK GTN testing criteria, where these are available.

Referral handling

- Laboratories will have systems in place for managing test requests, which allow them to be processed in a clinically appropriate, cost effective and timely way, in accordance with the testing turnaround times (see section 2.2.3.6.2). Laboratories will determine the most effective testing approach to be used, if necessary in discussion with the referrer.
- Not every laboratory will provide every test and laboratories must have appropriate systems in place for transporting specimens to other laboratories (or advising referrers of referral routes for specialised tests) and receiving results back safely and relaying these to the requesting clinician.

The following exclusions apply:

- Patients requiring specialist procedures should be referred to the appropriate specialists
- Genetic tests requested from non clinical genetics staff are excluded.
- Genetic testing within population screening programmes are subject to a separate contract.
- Companion diagnostics (including genetic tests) for drug treatment are the responsibility of the treating specialty.
- The lead Multidisciplinary Team (MDT) speciality has responsibility for ordering and paying for diagnostic tests.
- It should be noted that there are commissioned patient care pathways for a number of inherited disorders, which have a defined management specification and are delivered outside the clinical genetics service and are thus not part of this specification. These include genetic diagnoses for the symptomatic patient and their family members where agreed locally for example:
- Haemophilia and Sickle cell disease
- Familial hypercholesterolaemia
- There are some Clinical Genetic services not located in RGCs that are not commissioned as a specialised service as they have a single specialty interest (e.g. cancer genetic services at the Royal Marsden NHS Foundation Trust). These are excluded from this specification.
- Where the UK GTN's testing criteria are available these should be adhered to in the ordering of tests.
- Tests not approved by the UKGTN are excluded from this specification, with the exception of rare tests accessed from abroad which are not available in the UK. The UKGTN Directory includes validated molecular and cytogenetic tests (including agreed 'grandfather' tests developed prior to the UKGTN.)

The following commissioning policies support this specification (currently under development):

- Access to Pre-Implantation Genetic Diagnosis Services.
- Criteria for Consultant to Consultant Referrals to Clinical Genetics Services.

2.5 Interdependencies with other services

Co-located services

Genetics laboratory services should be provided on the same site as at least some other pathology services to allow access to other specialist expertise and technology where required. (Where there are other specialist stand – alone laboratories, links should be made with these as appropriate). Although most clinical genetics services are co-located with genetics laboratories, there is not a requirement for this to be the case.

Interdependent services

Laboratories: most clinical genetics services are co-located with genetics laboratories, although this is not a requirement. Whichever model is in place, there will be regular joint meetings to enable discussion about service developments, challenging/complex cases and the interpretation of novel findings. The clinicians may collaborate with their local scientists to aid the interpretation of genetic test results from external laboratories.

Haematology: Hereditary blood diseases (haemoglobinopathies/haemophilias) are normally managed by clinical haematologists who may require support or advice from geneticists.

Clinicians from medical or surgical specialties: some clinicians with a specialised area of expertise (but not trained as clinical geneticists) undertake the clinical genetic aspects of a condition, or group of conditions, and offer a national or supra-regional service, e.g. familial hypercholesterolaemia and mitochondrial disorders. Increasing numbers of specialties will integrate genetic testing within their established patient pathways but will still require input from Geneticists for rare cases.

Pre-Implantation Genetic Diagnosis (PGD) involves both specialist clinical genetic services and speciality infertility services. PGD requires input from laboratory and genetic counselling services as well as close collaboration with specialised infertility services for medical management, gamete and embryo work.

Pathology Services: The application of common technologies has led many genetic laboratories to develop links with local pathology services to exploit synergies in technological approaches for the provision of pathology services across the medical disciplines.

Outpatient clinics and infrastructure: Clinical Genetics Services will be expected to provide clinics on other hospital sites and therefore will need to have appropriate clinical and organisational links to support this.

Related services

Clinical Networks: Clinical Genetics Services will be expected to link with appropriate networks as required, e.g. Cancer, Cardiac.

Primary Care: Clinical Geneticists will be expected to have appropriate systems in place to link with primary care over the management of individual patients' condition and to provide education/ general advice as required.

Other Medical Genetics Services: Both Clinical Genetics and Laboratory Genetics will be expected to have appropriate links with the other Regional Genetic Services.

3. Applicable Service Standards

3.1 Applicable national standards e.g. NICE, Royal College

Core standards are:

- Familial Breast Cancer, 2006. NICE clinical guideline 41 (partial update) www.nice.org.uk
- Standards for medical laboratories, 2010 Clinical Pathology Accreditation – www.cpa-uk.co.uk
- National waiting times and access criteria as outlined in the National Operating Framework
- The UK GTN provides information relevant to genetic diagnosis for rare conditions on quality and standards for genetic testing and a Directory of Genetic Testing that lists tests that have been recommended for NHS service and approved for clinical validity and utility – www.ukgtn.nhs.uk
- Recommended standards are set out in the Best practice Guidelines of the organisation listed below (and detailed in section 1 Evidence Base)
- Clinical Molecular Genetic Society – www.cmgs.org
- Association for Clinical Cytogenetics – www.cytogenetics.org.uk

Required testing reporting times:

Accurate test results provided in accordance with the agreed reporting times. Reporting times vary depending on the nature and urgency of the referral (potential diagnosis, sample type, scale of testing, prenatal tests, etc). Typical target report times in current professional standards guidance are:

- Molecular genetics: 3 days, 10 days, 20 days, 40 days, 80 days (all working days) using a RAG rating where Red = <80%, Amber = 80- 89.9% and Green = ≥90% reports issued within these times.
- Cytogenetics: 3 days, 10 days, 14 days, 21 days, 28 days (all calendar days with exception of 3 days which is measured in working days) using a RAG rating where Red = <80%, Amber = 80-89.9% and Green = ≥90% reports issued within these times

Target	Time	Definition
3 days	3 working days	Polymerase chain reaction (PCR)-based tests where the result is needed urgently for prenatal diagnosis
2 weeks	10 working days	Southern blot tests where the result is needed urgently for prenatal diagnosis
4 weeks	20 working days	PCR-based tests where

		the familial mutation is known, specific mutation tests, or gene tracking by microsatellite analysis Non-urgent PCR-based tests where the familial mutation is known, specific mutation tests, or gene tracking by microsatellite analysis
8 weeks	40 working days	Mutation screening or tests that require Southern blot analysis

Family Based Record Systems

Each Genetics Centre will maintain a computerised family-based record system, incorporating disease-specific records.

4. Key Service Outcomes

Section 2.1 4 gives a list of the services expected from Genetic Services and a number of 'proxy' outcomes.

The information section of the contract provides the basic individual patient data and activity reporting required as part of the basic billing and contract monitoring arrangements (e.g. new/follow up outpatients and types of test/source of referral).

Proxy outcome measures that will be monitored and for which providers will be required to submit information are (in addition to those listed in the Genetics Dashboard):

- New to follow up ratios
- Numbers of new and follow up patients seen
- Proportion of appointments held at RGC compared with proportion in outreach clinics
- Reason for referral broken down into main disease categories (this will aid future service planning)
- Percentage of ethnic origin data recorded
- Number of genetic tests carried out by MOLU and CYTU (GENU) (methods of counting genetic tests)
- Numbers and type of tests sent to other laboratories

Genetic Dashboard measures (subject to some review for 2013/14)

The summary measures are listed below. Details of numerators and denominators and other guidance can be found on the Genetics Dashboard)

- Pick up rate for genetic testing - Proportion of tests that return a positive result for affected patients that have the test to determine a diagnosis and are seen in clinical genetics
- Multi Disciplinary Clinics (MDC) - Proportion of clinical genetic clinics that are part of a MDC/multi-disciplinary team (MDT)
- Clinical audits- Proportion of nationally approved clinical audits completed and action plans put in place (the number and type of audits need to be agreed)
- Laboratory reporting times - Proportion of Cytogenetics reports meeting turn round times as agreed by the professional organisations (CMGS/ACC)
- Laboratory reporting times - Proportion of Molecular reports meeting turn round times as agreed by the professional organisations (CMGS/ACC)
- Educational sessions provided by clinical genetics to other specialties to support genetics in mainstream medicine - number of educational sessions provided by clinical genetics to other specialties
- Poor patient experience - number of written complaints about the genetics department
- Good patient experience - number of letters/emails from patients, carers or non-genetics consultants registering thanks to the genetics department
- Patients waiting excessively for pre-natal (PN) genetic test results where the patient is seen in the clinical genetics department - proportion of patients receiving test result within 5 working days after the clinic receives the laboratory report for PN genetic test results.
- Do Not Attends (DNAs) as defined in the Data Dictionary - proportion of appointments that are not attended
- Patients consulted without a referral
- Patients consulted by a genetic counsellor - number of patients consulted by a genetic counsellor during period and number of appointments provided by a genetic counsellor during period
- Serious untoward incidents regarding patient care - number of serious untoward Incidents involving patient care
- Serious untoward incidents regarding lab tests - number of serious untoward Incidents involving laboratory tests
- External Quality Assurance (EQA) - scores from EQA schemes the laboratory participates in
- Activity audits (laboratory) - proportion of nationally agreed audits participated in by the genetics laboratory (the number and type of audits need to be agreed)
- Non-adherence to the UKGTN Testing Criteria (laboratory) as per UKGTN website - proportion of test requests from clinical genetics that did not comply to the UKGTN Testing Criteria where those criteria apply.

ANNEX 1 TO SERVICE SPECIFICATION:

PROVISION OF SERVICES TO CHILDREN

Aims and objectives of service

This specification annex applies to all children's services and outlines generic standards and outcomes that would be fundamental to all services.

The generic aspects of care:

The Care of Children in Hospital (Health Service Circular (HSC) 1998/238) requires that:

- Children are admitted to hospital only if the care they require cannot be as well provided at home, in a day clinic or on a day basis in hospital.
- Children requiring admission to hospital are provided with a high standard of medical, nursing and therapeutic care to facilitate speedy recovery and minimize complications and mortality.
- Families with children have easy access to hospital facilities for children without needing to travel significantly further than to other similar amenities.
- Children are discharged from hospital as soon as socially and clinically appropriate and full support provided for subsequent home or day care.
- Good child health care is shared with parents/carers and they are closely involved in the care of their children at all times unless, exceptionally, this is not in the best interest of the child; Accommodation is provided for them to remain with their children overnight if they so wish.

Service description/care pathway

All paediatric specialised services have a component of primary, secondary, tertiary and even quaternary elements.

The efficient and effective delivery of services requires children to receive their care as close to home as possible dependent on the phase of their disease.

Services should therefore be organised and delivered through 'integrated pathways of care' (National Service Framework for Children, Young People and Maternity Services (Department of Health & Department for Education and Skills, London 2004)

Interdependencies with other services

All services will comply with Commissioning Safe and Sustainable Specialised Paediatric Services: A Framework of Critical Inter-Dependencies – Department of Health

Imaging

All services will be supported by a three tier imaging network ('Delivering quality imaging services for children' Department of Health 13732 March 2010). Within the network:

- It will be clearly defined which imaging test or interventional procedure can be performed and reported at each site
- Robust procedures will be in place for image transfer for review by a specialist radiologist, these will be supported by appropriate contractual and information governance arrangements
- Robust arrangements will be in place for patient transfer if more complex imaging or intervention is required
- Common standards, protocols and governance procedures will exist throughout the network.
- All radiologists, and radiographers will have appropriate training, supervision and access to continuous professional development (CPD)
- All equipment will be optimised for paediatric use and use specific paediatric software

Specialist Paediatric Anaesthesia

Wherever and whenever children undergo anaesthesia and surgery, their particular needs must be recognised and they should be managed in separate facilities, and looked after by staff with appropriate experience and training.[1] All UK anaesthetists undergo training which provides them with the competencies to care for older babies and children with relatively straightforward surgical conditions and without major co-morbidity. However, those working in specialist centres must have undergone additional (specialist) training[2] and should maintain the competencies so acquired[3] *. These competencies include the care of very young/premature babies, the care of babies and children undergoing complex surgery and/or those with major/complex co-morbidity (including those already requiring intensive care support).

As well as providing an essential co-dependent service for surgery, specialist anaesthesia and sedation services may be required to facilitate radiological procedures and interventions (for example MRI scans and percutaneous nephrostomy) and medical interventions (for example joint injection and intrathecal chemotherapy), and for assistance with vascular access in babies and children with complex needs such as intravenous feeding.

Specialist acute pain services for babies and children are organised within existing departments of paediatric anaesthesia and include the provision of agreed (hospital wide) guidance for acute pain, the safe administration of complex analgesia regimes including epidural analgesia, and the daily input of specialist anaesthetists and acute pain nurses with expertise in paediatrics.

*The Safe and Sustainable reviews of paediatric cardiac and neuro- sciences in

England have noted the need for additional training and maintenance of competencies by specialist anaesthetists in both fields of practice.

References

1. Guidelines for the Provision of Anaesthetic Services (GPAS) Paediatric anaesthetic services. RCoA 2010 www.rcoa.ac.uk
2. Certificate of the Completion of Training (CCT) in Anaesthesia 2010
3. Continuing Professional Development (CPD) matrix level 3

Specialised Child and Adolescent Mental Health Services (CAMHS)

The age profile of children and young people admitted to specialised CAMHS day/in-patient settings is different to the age profile for paediatric units in that it is predominantly adolescents who are admitted to specialised CAMHS inpatient settings, including over-16s. The average length of stay is longer for admissions to mental health units. Children and young people in specialised CAMHS day/in- patient settings generally participate in a structured programme of education and therapeutic activities during their admission.

Taking account of the differences in patient profiles, the principles and standards set out in this specification apply with modifications to the recommendations regarding the following:

- Facilities and environment – essential Quality Network for In-patient CAMHS (QNIC) standards should apply (http://www.rcpsych.ac.uk/quality/quality,accreditationaudit/qnic1.aspx)
- Staffing profiles and training - essential QNIC standards should apply.
- The child/ young person's family are allowed to visit at any time of day taking account of the child / young persons need to participate in therapeutic activities and education as well as any safeguarding concerns.
- Children and young people are offered appropriate education from the point of admission.
- Parents/carers are involved in the child/young persons care except where this is not in the best interests of the child / young person and in the case of young people who have the capacity to make their own decisions is subject to their consent.
- Parents/carers who wish to stay overnight are provided with accessible accommodation unless there are safeguarding concerns or this is not in the best interests of the child/ young person.

Applicable national standards e.g. NICE, Royal Colleges

Children and young people must receive care, treatment and support by staff registered by the Nursing and Midwifery Council on the parts of their register that permit a nurse to work with children (Outcome 14h Essential Standards of Quality and Safety, Care Quality Commission, London 2010)

- There must be at least two Registered Children's Nurses (RCNs) on duty 24

hours a day in all hospital children's departments and wards.
- There must be an Registered Children's Nurse available 24 hours a day to advise on the nursing of children in other departments (this post is included in the staff establishment of 2RCNs in total).

Accommodation, facilities and staffing must be appropriate to the needs of children and separate from those provided for adults. All facilities for children and young people must comply with the Hospital Build Notes HBN 23 Hospital Accommodation for Children and Young People NHS Estates, The Stationary Office 2004.

All staff who work with children and young people must be appropriately trained to provide care, treatment and support for children, including Children's Workforce Development Council Induction standards (Outcome 14b Essential Standards of Quality and Safety, Care Quality Commission, London 2010).

Each hospital which admits inpatients must have appropriate medical cover at all times taking account of guidance from relevant expert or professional bodies (National Minimum Standards for Providers of Independent Healthcare, Department of Health, London 2002).'Facing the Future' Standards, Royal College of Paediatrics and Child Health.

Staff must carry out sufficient levels of activity to maintain their competence in caring for children and young people, including in relation to specific anaesthetic and surgical procedures for children, taking account of guidance from relevant expert or professional bodies (Outcome 14g Essential Standards of Quality and Safety, Care Quality Commission, London 2010).

Providers must have systems in place to gain and review consent from people who use services, and act on them (Outcome 2a Essential Standards of Quality and Safety, Care Quality Commission, London 2010). These must include specific arrangements for seeking valid consent from children while respecting their human rights and confidentiality and ensure that where the person using the service lacks capacity, best interest meetings are held with people who know and understand the person using the service. Staff should be able to show that they know how to take appropriate consent from children, young people and those with learning disabilities (Outcome 2b) (Seeking Consent: Working with Children Department of Health, London 2001).

Children and young people must only receive a service from a provider who takes steps to prevent abuse and does not tolerate any abusive practice should it occur (Outcome 7 Essential Standards of Quality and Safety, Care Quality Commission, London 2010 defines the standards and evidence required from providers in this regard). Providers minimise the risk and likelihood of abuse occurring by:
- Ensuring that staff and people who use services understand the aspects of the safeguarding processes that are relevant to them.
- Ensuring that staff understand the signs of abuse and raise this with the right person when those signs are noticed.

- Ensuring that people who use services are aware of how to raise concerns of abuse.
- Having effective means to monitor and review incidents, concerns and complaints that have the potential to become an abuse or safeguarding concern.
- Having effective means of receiving and acting upon feedback from people who use services and any other person.
- Taking action immediately to ensure that any abuse identified is stopped and suspected abuse is addressed by:
    - having clear procedures followed in practice, monitored and reviewed that take account of relevant legislation and guidance for the management of alleged abuse
    - separating the alleged abuser from the person who uses services and others who may be at risk or managing the risk by removing the opportunity for abuse to occur, where this is within the control of the provider
    - reporting the alleged abuse to the appropriate authority
    - reviewing the person's plan of care to ensure that they are properly supported following the alleged abuse incident.
- Using information from safeguarding concerns to identify non-compliance, or any risk of non-compliance, with the regulations and to decide what will be done to return to compliance.
- Working collaboratively with other services, teams, individuals and agencies in relation to all safeguarding matters and has safeguarding policies that link with local authority policies.
- Participates in local safeguarding children boards where required and understand their responsibilities and the responsibilities of others in line with the Children Act 2004.
- Having clear procedures followed in practice, monitored and reviewed in place about the use of restraint and safeguarding.
- Taking into account relevant guidance set out in the Care Quality Commission's Schedule of Applicable Publications
- Ensuring that those working with children must wait for a full CRB disclosure before starting work.
- Training and supervising staff in safeguarding to ensure they can demonstrate the competences listed in Outcome 7E of the Essential Standards of Quality and Safety, Care Quality Commission, London 2010

All children and young people who use services must be

- Fully informed of their care, treatment and support.
- Able to take part in decision making to the fullest extent that is possible.
- Asked if they agree for their parents or guardians to be involved in decisions they need to make.

(Outcome 4I Essential Standards of Quality and Safety, Care Quality Commission, London 2010)

Key Service Outcomes

Evidence is increasing that implementation of the national Quality Criteria for Young People Friendly Services (Department of Health, London 2011) have the potential to greatly improve patient experience, leading to better health outcomes for young people and increasing socially responsible life-long use of the NHS. Implementation is also expected to contribute to improvements in health inequalities and public health outcomes e.g. reduced teenage pregnancy and STIs, and increased smoking cessation. All providers delivering services to young people should be implementing the good practice guidance which delivers compliance with the quality criteria.

Poorly planned transition from young people's to adult-oriented health services can be associated with increased risk of non adherence to treatment and loss to follow-up, which can have serious consequences. There are measurable adverse consequences in terms of morbidity and mortality as well as in social and educational outcomes. When children and young people who use paediatric services are moving to access adult services (for example, during transition for those with long term conditions), these should be organised so that:

- All those involved in the care, treatment and support cooperate with the planning and provision to ensure that the services provided continue to be appropriate to the age and needs of the person who uses services.

The National Minimum Standards for Providers of Independent Healthcare, (Department of Health, London 2002) require the following standards:

- **A16.1** Children are seen in a separate out-patient area, or where the hospital does not have a separate outpatient area for children, they are seen promptly.
- **A16.3** Toys and/or books suitable to the child's age are provided.
- **A16.8** There are segregated areas for the reception of children and adolescents into theatre and for recovery, to screen the children and adolescents from adult patients; the segregated areas contain all necessary equipment for the care of children.
- **A16.9** A parent is to be actively encouraged to stay at all times, with accommodation made available for the adult in the child's room or close by.
- **A16.10** The child's family is allowed to visit him/her at any time of the day, except where safeguarding procedures do not allow this
- **A16.13** When a child is in hospital for more than five days, play is managed and supervised by a qualified Hospital Play Specialist.
- **A16.14** Children are required to receive education when in hospital for more than five days; the Local Education Authority has an obligation to meet this need and are contacted if necessary.
- **A18.10** There are written procedures for the assessment of pain in children and the provision of appropriate control.

All hospital settings should meet the Standards for the Care of Critically Ill Children (Paediatric Intensive Care Society, London 2010).

There should be age specific arrangements for meeting Regulation 14 of the Health

and Social Care Act 2008 (Regulated Activities) Regulations 2010. These require:

- A choice of suitable and nutritious food and hydration, in sufficient quantities to meet service users' needs;
- Food and hydration that meet any reasonable requirements arising from a service user's religious or cultural background
- Support, where necessary, for the purposes of enabling service users to eat and drink sufficient amounts for their needs.
- For the purposes of this regulation, "food and hydration" includes, where applicable, parenteral nutrition and the administration of dietary supplements where prescribed.
- Providers must have access to facilities for infant feeding, including facilities to support breastfeeding (Outcome 5E, of the Essential Standards of Quality and Safety, Care Quality Commission, London 2010)

All paediatric patients should have access to appropriately trained paediatric trained dieticians, physiotherapists, occupational therapists, speech and language therapy, psychology, social work and CAMHS services within nationally defined access standards.

All children and young people should have access to a professional who can undertake an assessment using the Common Assessment Framework and access support from social care, housing, education and other agencies as appropriate

All registered providers must ensure safe use and management of medicines, by means of the making of appropriate arrangements for the obtaining, recording, handling, using, safe keeping, dispensing, safe administration and disposal of medicines (Outcome 9 Essential Standards of Quality and Safety, Care Quality Commission, London 2010). For children, these should include specific arrangements that:

- ensure the medicines given are appropriate and person-centred by taking account of their age, weight and any learning disability
- ensure that staff handling medicines have the competency and skills
- needed for children and young people's medicines management
- ensure that wherever possible, age specific information is available for people about the medicines they are taking, including the risks, including information about the use of unlicensed medicine in paediatrics.

Many children with long term illnesses have a learning or physical disability. Providers should ensure that:

- They are supported to have a health action plan
- Facilities meet the appropriate requirements of the Disability
- Discrimination Act 1995
- They meet the standards set out in Transition: getting it right for young people. Improving the transition of young people with long-term conditions from children's to adult health services. Department of Health, 2006, London.