

# Equality Analysis: UK Plan for Rare Diseases

February 2012



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<b>Policy</b>	Estates
HR/Workforce Management	Commissioning
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Clinical	Finance
	Social Care/Partnership Working
<b>Document purpose</b>	Consultation/Discussion
<b>Gateway reference</b>	16959
<b>Title</b>	Consultation on the United Kingdom Plan for Rare Diseases
<b>Author</b>	UK Health Departments
<b>Publication date</b>	29 February 2012
<b>Target audience</b>	PCT CEs, NHS Trust CEs, SHA CEs, Foundation Trust CEs, Medical Directors, Directors of PH, Directors of Nursing, Allied Health Professionals, GPs, Royal Colleges, Scottish NHS Boards
<b>Circulation list</b>	Specialist societies, academies & organisations, Royal Colleges, UK GTN, Specialised Commissioning Groups, Patient-related organisations, charities.
<b>Description</b>	This consultation document on the UK Plan for Rare Diseases is in response to the 2009 European Council Recommendation on Rare Diseases. The consultation has been developed jointly by the four nations of the UK. The consultation responses will be used to develop the final UK Plan for Rare Diseases to be published later in 2012.
<b>Cross reference</b>	N/A
<b>Superseded documents</b>	N/A
<b>Action required</b>	N/A
<b>Timing</b>	<b>Responses are invited to this consultation by 25 May 2012</b>
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# **Equality Analysis: UK Plan for Rare Diseases**

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## Introduction

An evidence review was undertaken as part of the Equality Analysis for the consultation on the UK Plan on Rare Diseases (UKPRD). The aim of the evidence review was to establish what positive and negative impacts the UKPRD may have upon groups with protected characteristics identified by the Equality Act 2010. The term “protected characteristics” comes from the Equality Act 2010 and covers the following: disability, sex, sexual orientation, race, age, gender reassignment, religion or belief and pregnancy or maternity.

People who suffer from a rare disease (defined as five or less cases per 10,000 population; The Council of the European Union, 2009) are faced with a number of obstacles due to the small number of people affected and the high complexity of the diseases. These obstacles include but are not limited to (Kole and Faurisson, 2010): (i) lack of scientific knowledge of their disease, (ii) lack of access to correct diagnosis, (iii) delays in diagnosis, (iv) lack of appropriate multidisciplinary healthcare, (v) lack of quality information and support at the time of diagnosis, (vi) undue social consequences, (vii) inequities and difficulties in access to treatment, rehabilitation and care, (viii) dissatisfaction with and loss of confidence in medical and social services, (ix) denied treatment by health professionals and (x) lack of availability of orphan drugs.

These obstacles have led to inequalities in access to and quality of care between people who have a rare disease and the rest of society who use “mainstream” health services (Kole and Faurisson, 2010).

Individually these diseases are rare, but there are more than 5,000 rare diseases, so that collectively they add up to a significant problem for a great many people. As a result of this the European Union have identified rare diseases as a priority for action to improve the coordination and coherence of plans and strategies to tackle rare diseases across Europe. This collaborative approach being taken by Member States across Europe should improve the quality/availability of care and access to care for people with rare diseases, therefore reducing inequalities that exist.

Equality means ensuring that every individual has an equal opportunity to make the most of their lives and talents, and believing that no one should have poorer life chances because of where, what or whom they were born, what they believe, or whether they have a disability.

The general equality duty in the Equality Act (2010) requires public authorities, in the exercise of their functions, to have a due regard to the need to:

- eliminate unlawful discrimination, harassment and victimisation and other conduct prohibited by the Act;
- advance equality of opportunity between people who share a protected characteristic and those who do not;
- foster good relations between people who share a protected characteristic and those who do not.

As a result, the UKPRD must be considered for its potential to impact on people who have protected characteristics.

Around 80% of rare diseases are of genetic origin and therefore can be linked with particular sexes, genders or disabilities (Guillem *et al.*, 2007). Rare diseases are often identified in childhood and therefore disproportionately affect the younger generation (Chief Medical Officer for England, 2009). Likewise, some rare diseases are very debilitating and therefore they impact on the family/carers of a person with a rare disease.

The United Kingdom already has a strong history of supporting and treating people with rare diseases; through healthcare for complex conditions, world class research into rare diseases, and patient organisations who express the needs and priorities of people with rare diseases.

The UK recognises the opportunity to build on this position, while considering the broader European context and the benefits of cross-border collaborations. The UK is therefore supporting a European Council Recommendation that asks all Member States to develop a national plan or strategy by the end of 2013.

The NHS Constitution (England, but the principles are the same in Northern Ireland, Scotland and Wales) states that:

*“The NHS provides a comprehensive service, available to all irrespective of gender, race, disability, age, sexual orientation, religion or belief. [...] It has a duty to each and every individual that it serves and must respect their human rights. Everyone counts. We will use our resources for the benefit of the whole community, and make sure nobody is excluded or left behind.”*

The consultation on the UKPRD aims to improve co-ordination of services, result in better outcomes, strengthen research and monitoring activities, engage and empower patients and their families/carers, and raise awareness in the

public and in professionals. As well as this the plan will encourage the recognition of diversity, in other words differences amongst and between individuals. Diversity recognises that we need to respond differently to both individuals and to groups if we are to provide accessible services to promote an inclusive working environment.

Therefore within both the NHS and broader societal context the UKPRD should have a positive impact on people with protected characteristics as outlined by the Equality Act. In order to establish if there is any evidence to the contrary, a literature search was conducted to investigate any impact on inequalities by rare diseases.

## Search Strategy

Research articles were obtained by a computerised search in NHS Evidence: Health Information Resources (2011, accessed from [www.evidence.nhs.uk](http://www.evidence.nhs.uk)). NHS Evidence searches:

- evidence-based reviews (Bandolier, Cochrane Library, Database of Abstracts of Reviews of Evidence [DARE], Health Technology Assessment [HTA] Database, NHS Economic Evaluation Database [EED] and UK Database of Uncertainties about the Effects of Treatments [DUETs]);
- guidance (Clinical Knowledge Summaries [CKS] formerly PRODIGY, National Library of Guidelines, and selected International Guidelines);
- specialist collections (collections of the best available evidence for different communities of practice);
- books, journals and healthcare databases (AMED – Allied and Complimentary Medicines, British Nursing Index, CINAHL, EMBASE – Excerpta Medica Database, E-books, Health Business Elite, Health Management Information Consortium, Medical Literature Analysis and Retrieval System Online [MEDLINE] – U.S. National Library of Medicine, My Journals, and PsycINFO).

Key search terms used were; “rare disease\*”, inequalit\*, and each of the protected characteristics listed above.

These search terms were used as outlined in the table below.

<b>Search</b>	<b>Hits (with duplications)</b>	<b>Results (excluding duplications)</b>
“rare disease*” and inequalit*	8	2 relevant titles
“rare disease*” and disabilit*	158	6 relevant titles
“rare disease*” and (sex or gender)	465	2 relevant titles
“rare disease*” and (race or ethnic*)	186	3 relevant titles
“rare disease*” and age	3164. Search was repeated in the title only and yielded 5 results	2 relevant articles
“rare disease*” and “gender reassignment”	0	0 relevant titles
“rare disease*” and (“sexual orientation” or sexuality)	5	0 relevant titles
“rare disease*” and (religion or belief)	11	1 title, but relevant to maternity/pregnancy  0 relevant titles for religion or belief
“rare disease*” and (pregnan*)	570	10 relevant titles
“rare disease*” and (carer* or famil*)	1704. Search was repeated in the title only and yielded 13 results	1 title, but relevant to ethnicity  0 titles relevant to families/carers

## Selecting and Reviewing Evidence

Search results were initially screened by title to reduce the number of results. The basis of the screen was to identify articles that examined the impact of rare diseases or a rare disease on the defined category (both positive or negative). Article titles were also screened to identify if rare diseases or a rare disease are linked with multiple categories, for example, rare diseases in disabled children.

The abstracts of selected titles were reviewed to further investigate if the article was relevant to the objective of the literature search. Articles which were deemed relevant were used to form the evidence below.

Due to time restrictions, the full articles selected were not reviewed in detail for methodological quality. Articles were only accepted from scholarly journals that

are peer reviewed or from key rare disease organisations (Specialised Healthcare Alliance, Rare Disease UK, and the European Organisation for Rare Diseases [EURORDIS]). This was to ensure the quality of the evidence articles identified.

## Disability

Several studies have shown that although some rare diseases do not necessarily affect life expectancy, the majority lead to physical, emotional and/or psychosocial limitations with a wide range of disabilities. Guillem *et al.* (2008) found that 26% of children with mental, sensorial, neurological (skeletal and neurological) impairments have a rare disease.

It is also to be underlined that relatively common conditions can hide underlying rare diseases, e.g. autism (in Rett syndrome, Usher syndrome type II, Sotos Cerebral Gigantism, Fragile X, Angelman, Adult Phenylketonuria, Sanfilippo,...) or Epilepsy (Shokeir syndrome, Feigenbaum Bergeron Richardson syndrome, Kohlschutter Tonz syndrome, Dravet syndrome...). For many conditions described in the past as clinical ones such as mental deficiency, cerebral palsy, autism or psychosis, a genetic origin is now suspected or has already been described. In fact, a rare disease can be masked by a host of other conditions, which may lead to misdiagnosis (EURORDIS, 2005).

Given the link between rare diseases and disabilities progressing work on rare diseases should also have a positive impact on people with disabilities in the UK; particularly around improving diagnosis and reducing what is often termed as “diagnostic odyssey” (i.e. delay). This will help improve access to care and reduce the stress and anxiety associated with a lack of diagnosis.

## Sex/Gender

The genetic causes of rare diseases can affect males and females differently – for example systemic sclerosis (scleroderma) is four times more common in women than in men and systemic lupus erythematosus (SLE) is nine times more common in women than in men.

In some cases a disease may be rare in one sex but not in another. For example, male breast cancer is classified as a rare disease, representing only 1% of the total number of breast cancers worldwide (Rudlowski, 2008).

Any action to improve the UK's current position in relation to rare diseases may therefore differentially affect males and females. However, this will be due to the needs of people with rare diseases and not because of any discrimination.

## Race/Ethnicity

As previously mentioned 80% of rare diseases have a genetic cause which means that they can sometimes be more common in certain races and ethnicities. For example, de Serres (2003) summarised that articles in the literature on alpha-1 antitrypsin (AAT) deficiency have been interpreted as indicating that AAT deficiency is a rare disease that affects mainly Caucasians (whites) from northern Europe. De Serres (2003) goes on to report that a recent publication on the worldwide racial and ethnic distribution of AAT deficiency, new data were presented demonstrating that it is also found in various populations of African blacks; Arabs and Jews in the Middle East; and Central, Far East, and Southeast Asians, as well as whites in Australia, Europe, New Zealand, and North America.

Similarly, Familial Mediterranean fever (FMF) is an autosomal recessive disorder, characterised by short, recurrent attacks of fever with abdominal, chest or joint pain and erysipelas-like erythema. It is an ethnically restricted genetic disease, found commonly among Mediterranean populations, as well as Armenians, Turks, Arabs and Jews (La Regina *et al.*, 2003).

These examples show that rare diseases can impact on certain groups in society in different ways. This means that working with a variety of ethnic groups calls for ethnically sensitive approaches; largely in communication.

Engagement with and empowerment of people with rare diseases is a key tenet of the UKPRD. Work on this should remain sensitive to the differing needs of people with rare diseases – this includes language and cultural sensitivities. The UKPRD will stress the importance of supplying information to people with rare diseases (and their families/carers) in a variety of languages and formats. Treatment and care also needs to be handled in a culturally sensitive way, recognising that there could be issues around diet (Ramadam), gender, carers, providing blood samples and so on.

## Age

According to EURORDIS (2007) 75% of rare diseases affect children and 30% of these children die before the age of 5. There is also great diversity in the age at which the first symptoms occur. Symptoms of many rare diseases appear at

birth or in childhood, including Infantile Spinal Muscular Atrophy, Neurofibromatosis, Osteogenesis Imperfecta, Rett syndrome and most metabolic diseases, such as Hurler, Hunter, Sanfilippo, Mucopolysaccharidosis Type II, Krabbe diseases, Chondrodysplasia.

In some cases, the first symptoms of the disease, such as Neurofibromatosis, may occur in childhood, but this does not prevent much heavier symptoms to occur at a later stage of life. Other rare diseases, such as Huntington disease, Spinocerebellar Ataxias, Charcot-Marie-Tooth disease, Amyotrophic Lateral Sclerosis, Kaposi's Sarcoma and thyroid cancer, are specific to adulthood. Whilst many diseases cause symptoms in childhood, these symptoms may not translate into a specific rare diagnosis for years. (EURORDIS, 2005).

One area of difficulty highlighted by people with rare diseases is the progression from childhood to adulthood. Limb, Nutt and Sen (2010) found that almost 30% of patients reported experiencing problems in the transition from paediatric to adult services. Many of these problems lay with the way in which services are delivered and unmet needs during this difficult period in a person's life. This transition between paediatric and adult services is a key area of action for the UKPRD. The plan stresses the importance of networks of care which support people with rare diseases throughout the life-course and reduce disruptions to their care.

The impact of rare diseases in children is far greater than on the individual because it impacts on their parents and other family members too. A number of studies cite the need to consider the wider/holistic needs of a child and their family/carers, that spread from pure health needs to social needs – family relationships, economic well-being, daily living (Gaité Pindado *et al.*, 2008).

## Gender Reassignment

No articles were identified when searching for gender reassignment and rare diseases. Gender Dysphoria Services are currently commissioned as a specialised service in England by Specialised Commissioning Groups as defined within the 3rd edition of the Specialised Services National Definitions Set.

Subject to the passage of the Health and Social Care Bill through Parliament, in England, it is the Government's intention that specialised services will in future be the responsibility of the NHS Commissioning Board. By commissioning specialised services in this way it is expected that any suggestions of a "postcode lottery" will end. No final decisions have been taken on Gender Dysphoria Services.

## Sexuality/Sexual Orientation

No relevant articles were identified when searching for sexual orientation/sexuality and rare diseases.

## Religion/Belief

No relevant titles were identified when searching for religion/belief and rare diseases.

However, the consultation on the UKPRD contains recommendations around Genetic Testing and Screening which can often raise ethical questions. There is a UK Genetic Testing Network which advises the NHS (Genetics Commissioning Advisory Group) on genetic testing and a UK National Screening Committee which advises ministers on screening. Each of these committees provide expert advice and will consider each genetic test/screening programme on their own merits.

Therefore, systems are already in place to consider and manage any ethical issues that may arise.

## Maternity/Pregnancy

Pregnancy in people with rare diseases can often be a challenge. For example Hereditary angioedema type III (HAE) is a rare disease which often affects young women and during pregnancy, symptoms worsened for 50% of them. Some deliveries can be very difficult with still birth (Bouillet, 2010).

Pregnancy can be particularly difficult for women with diseases affecting the immune system. Pregnancies in patients with systemic lupus erythematosus (SLE) are mostly successful when well planned and monitored interdisciplinarily, whereas a small proportion of women with antiphospholipid syndrome (APS) still have adverse pregnancy outcomes in spite of the standard treatment. New prospective studies indicate better outcomes for pregnancies in women with rare diseases such as systemic sclerosis (scleroderma) and vasculitis. Fertility problems are not uncommon in patients with rheumatic disease and need to be considered in both genders (Ostensen *et al.*, 2011).

Pregnancy amongst women with a rare disease is an area where research is increasing, particularly as children with rare diseases survive childhood (through improved management) and move into adulthood. For example, Glatter *et al.* (2005) describe successful pregnancies in two women with Postural orthostatic tachycardia syndrome (POTS) which (at the time) were thought to be the first

reports of this. The consultation on the UKPRD has a strong emphasis on research and developing the evidence-base which should result in improved care for people with rare diseases.

Around 80% of rare diseases have identified genetic origins, involving one or several genes or chromosomal abnormalities. They can be inherited or derived from a new gene mutation or from a chromosomal abnormality. They concern between 3% and 4% of births (EURORDIS, 2005).

There are a number of antenatal and newborn screening programmes offered across the UK (these do vary from country to country) that cover a number of rare diseases. This has the potential to improve health outcomes by minimising mortality and morbidity, and for some conditions alerting parents to their risks of having other affected children. Equity of uptake of testing/screening is often an issue. All screening programmes should be assessed for their equity of uptake.

Some rare diseases can be prevented through improved diets in pregnancy, reducing the risk of infections, and avoiding hazards preconception and during pregnancy (Taruscio et al. 2011). For example, preventing some birth defects (such as neural tube defects) with folic acid during preconception and the first trimester through improved diets and/or vitamin supplements. The UKPRD supports efforts to prevent rare diseases from occurring where possible.

## Carers

Wallenius, Moller and Berglund (2009) studied (n= 2,983) the physical, psychosocial, emotional and financial impact of having a rare diagnosis. The results were presented under the categories 'contacts with health care and society: 'consequences of getting the wrong diagnosis', 'family experiences in daily life', 'extra expenses related to the rare diagnosis' and 'positive effects of having a rare diagnosis.' The result showed that these groups are at risk of being treated arbitrarily when needing service from the society. The respondents also reported the high price of having a rare disease; both on monetary costs to the individual, and time costs to friends, families and carers. The study concluded by suggesting that the care that individuals receive for their rare disease can be a result of chance and coincidence, rather than co-ordinated and consistent care.

Not only do rare diseases impact upon the quality of life for people with a disease, they also have a major impact upon carers of people with a disease (Rajmil, Perestelo-Perez and Herdman).

Dellve *et al.* (2006) found that by empowering the parent/carers of children with rare diseases (through an intensive family [and carer] competence intervention) there was decreased strain (particularly among working mothers and fathers), increased perceived knowledge, and improved overall life satisfaction. Given that a key strand of the consultation on the UKPRD is to improve patient/family/carer empowerment then this should have a positive impact.

## Geography

The funding for drugs and services for managing and treating rare diseases varies across Europe and within the UK and this has led to geographical variations in access to care (Hughes, Tunnage and Yeo, 2005).

Limb, Nutt and Sen (2010) highlighted inequalities in the services received by patients with different rare diseases and even between those affected by the same rare disease in different parts of the UK. These geographical variations are difficult to manage across borders and even within countries, however the UKPRD should reduce these variations by encouraging collaborations and networking.

In England, subject to the passage of the Health and Social Care Bill through Parliament, specialised services will in future be the responsibility of the NHS Commissioning Board, mandated by ministers. This will ensure that all patients can access equitable high quality services, regardless of which rare condition they have and regardless of where they live. It will ensure comprehensive arrangements for all rare diseases and is expected to end any suggestions of a “postcode lottery”.

## Conclusion

The inequalities highlighted by the evidence review relate to the inequalities that exist in access to treatment and support for people with rare diseases and the rest of society who access the NHS. The aim of the UKPRD is to begin to reduce this inequality through better co-ordination and collaboration within the UK and also across borders with European partners.

The evidence review highlighted that the main source of inequality comes from having a rare disease, not explicitly from having any protected characteristics outlined by the Equality Act 2010. The evidence review showed that rare diseases are associated with a number of protected characteristics; therefore, any improvement in the UK’s action on rare diseases will also lead to an improvement for people with protected characteristics.

There is evidence that investing in improving identification, management and treatment in rare diseases can save society and the health service money by improving the quality of life for people with rare diseases and their carers, reducing morbidity and mortality and reducing demand on health services. (Olauson, 2002; Toscani and Riedl, 2011).

A survey by EURORDIS (EurordisCare2) focusing on diagnostic delays for rare diseases, has revealed that, for Ehlers Danlos syndrome, one out of four patients waited for more than 30 years before being given the right diagnosis. Forty percent of patients participating in the survey received a wrong diagnosis before being given the right one. Among them:

- one out of six underwent surgical treatment based on this wrong diagnosis;
- one out of 10 underwent psychological treatment based on this wrong diagnosis.

The consequences of diagnosis delay can be:

- other children born with the same disease;
- inappropriate behaviour and inadequate support from family members;
- clinical worsening of the patient's health in terms of intellectual, psychological and physical condition, even leading to the death of the patient;
- loss of confidence in the healthcare system.

This strengthens the argument that the UKPRD should have a positive impact on people with rare diseases, and often (as illustrated above) people with rare diseases fall into the protected characteristics identified by the Equality Act 2010.

In order to ensure that the UKPRD does not inadvertently exacerbate existing inequalities (i.e. differential access to services between groups who have a rare disease e.g. geographically different access) the plan should encourage equity audit as part of routine rare disease activity. This could be done by through improved recording and monitoring of protected characteristics in disease registers, in service delivery (and care co-ordination) and in access to medication/health technologies. In addition, any screening programmes endorsed by the UK National Screening Committee should continue to be assessed/audited for fair/equal access.

Engagement with and empowerment of people with rare diseases is a key tenet of the consultation on the UKPRD. Work on this should remain sensitive to the differing needs of people with rare diseases – this includes language and cultural sensitivities. The UKPRD will stress the importance of supplying information to people with rare diseases (and their families/carers) in a variety of languages and formats.

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2900004 Feb 2012

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