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Debate pack: Access to drugs for ultra-rare diseases

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Summary

This debate pack has been compiled ahead of the debate on Access to drugs for ultra-rare diseases to be held on Tuesday 16 June 2015 at 2.30pm in Westminster Hall. The Member in charge of the debate is Greg Mulholland. Debate packs are produced quickly after the announcement of Parliamentary business. They are intended to provide a summary or overview of the issue being debated and identify relevant briefings and useful documents, including press and parliamentary material. More detailed briefing can be prepared for MPs on request to the Library.

Rare and ultra-rare diseases each affect a very small number of patients, but overall about 3 million people in the UK will be affected by one of these conditions. The UK Rare diseases Strategy, published in November 2013, and to be implemented across the UK, seeks to address some of the specific challenges relating to these conditions, such as improving diagnosis, and investing in research for these conditions.

Drugs for rare diseases (orphan medicines) are usually granted marketing authorisations through a centralised procedure at the European Medicines Agency and services for rare conditions are the direct commissioning responsibility of NHS England in England. More information on the process for consideration of new treatments is provided in the briefing in this debate pack.

Prior to the Westminster Hall debate, Digital Outreach and Genetic Alliance UK organised a digital Rare Disease debate on Twitter. The debate started at 2pm on Monday 15 June. A number of patients and their families, patient groups, charities and the Member in charge of the Westminster Hall debate, Greg Mulholland all contributed.

The idea for digital debates linked to debates in Westminster Hall came from the Speaker's Commission on Digital Democracy, which stated:

> "We believe the public want the opportunity to have their say in House of Commons debates; we also believe that this will provide a useful resource for MPs and help to enhance those debates. We therefore recommend a unique experiment: the use of regular digital public discussion forums to inform debates held in Westminster Hall."

The Twitter debate formally finished at 4pm yesterday but it is likely that people will continue to contribute using the hashtag #RDdebate throughout the Westminster Hall debate today. To keep up with the debate on Twitter please use this link: Twitter Rare Disease debate.

1. Rare and ultra-rare diseases

A rare disease is generally considered as one affecting fewer than 5 people in 10,000, ultra-rare diseases have been described as affecting 1 person in 10,000 or fewer.[1] However, large numbers of people are affected by these rare conditions, it is estimated that in the EU, 5-8000 rare diseases affect 6-8% of the population, equalling between 27-36 million people. The *UK Rare Diseases Strategy,* published in November 2013, estimated that in the UK alone, more than 3 million people will suffer from a rare disease at some point in their life.[2]

A significant proportion (around 80%) of these conditions have a known genetic cause but other causes include infections, cancer and exposure/deficiencies to substances prior to birth. Due to the fact that relatively small numbers of people are affected with a particular rare disease, there are specific challenges in ensuring speedy diagnosis and access to appropriate services and treatments.

This briefing will provide an overview of access to drugs for rare diseases and UK Government policy in this area.

In order to be funded for use in the UK with NHS funding, a medicine would usually require a marketing authorisation and then a funding decision by the relevant NHS body. In the case of drugs for rare and ultra-rare diseases, often referred to as orphan drugs, marketing authorisations are usually granted by the European Medicines Agency (EMA) through a centralised procedure.

In order to allow access to drugs to meet an unmet demand and in the interest of public health, where less than complete data is provided on a product, a conditional marketing authorisation may be granted by the EMA.[3] The EMA also provide incentives to encourage the development of these medicines, these include fee reductions and ten year marketing exclusivity from the EMA.

The UK Government has launched the Early access to medicine scheme in 2014. This aims to allow access to medicines that do not yet have a full marketing authorisation for patients with severely debilitating or life threatening conditions, where there is a clear unmet medical need. The Medicines and Healthcare Products Regulatory Agency is responsible for the scheme and provides a scientific opinion on the benefit/risk balance of the medicine.[4]

In November 2014, a new Innovative Medicines review to consider how access to new medicines can be accelerated was also announced by the UK Government.[5]

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1 European Commission, Rare diseases
2 Department of Health, *The UK Strategy for Rare Diseases*, November 2013
3 EMA, Presubmission guidance, FAQs
4 HC Deb 28 November 2014 | PQ 215820
5 GOV.uk, *News story: Major investment in life sciences*, November 2014

Commissioning of rare disease services

Commissioning of specialised services is performed centrally, in England this has been the direct responsibility of NHS England since April 2013. These services account for about 14% of the total NHS budget, around £13.8 billion every year.[6]

NHS England has recently developed a new decision making tool for the commissioning of specialised services but in November 2014, it postponed the introduction of this and decided to consult on its proposals.[7] [8] . This decision followed concerns expressed about the new process, which included whether it may be unbalanced in the consideration of drugs for rare diseases. A number of patient groups had called for further consideration of the policy.[9] This consultation closed in April 2015.

A number of charities and campaign groups representing patients with rare conditions, and their families, have raised concerns about the delay in the consideration of a number of drugs during the consultation process.[10]

In response to a Westminster Hall debate on this issue in January 2015, the Minister for Life Sciences, George Freeman, provided more information on these recent events:

> In summer 2014, a decision-making aid for the prioritisation of new interventions and treatments was developed by a partnership of stakeholders, including more than 250 patient representatives. It was due to be used for the first time in early December 2014, but on 28 November 2014 NHS England decided to postpone its introduction, in response to concerns that some patients affected by rare diseases might be disadvantaged by its application. The legal process about that must now run its course. I understand that NHS England is, rightly, reviewing the appropriate approach to prioritising new treatments and interventions within specialised commissioning in response to those concerns. A 90-day consultation on the prioritisation framework and decision-making process for commissioning decisions on new treatments will be launched by NHS England shortly. This morning, I again raised the importance and urgency of that consultation process. [11]

He also highlighted the difficult decisions involved with NHS funding for treatments:

> Ideally, of course, we would want to fund all the treatments that are shown to benefit patients in any way, but we have to make difficult decisions about how we spend the money that we have available. That is why we have put clinicians in charge of the process, so that they can make decisions based on patient benefit

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6 NHS England, Specialised Services,
7 NHS England, NHS England responds to patient feedback and consults on prioritising specialised services, January 2015
8 NHS England, Investing in specialised commissioning - public consultation
9 Health Service Journal, *Exclusive: NHS England delays treatment choices after legal threat,* 2 December 2014
10 MPS, *Interim funding for ultra-rare diseases is essential whilst NHS England re-examines its processes,* 23 January 2015
11 HC Deb 20 January 2015 c30WH

> and on the best health economic assessments that we can make. The painful truth is that with finite resources, when we make a decision in one case to accept a drug, we will make a decision elsewhere to reject, and we have a duty to all to ensure that we make those decisions fairly.[12]

Where no decision has been made on a commissioning policy on the provision of a drug, an individual funding request (IFR) can be made to NHS England.[13] Clinicians, on behalf of patients can make an IFR for treatment to NHS England for treatments that would not normally be funded. NHS England provide more guidance on this process.

Evaluation of medicines

The National Institute for Health and Care Excellence (NICE) provides evidence-based information for the NHS on the effectiveness and cost-effectiveness of healthcare interventions. It publishes mandatory technology appraisal guidance (stipulating clinical interventions – mainly medicines – which must be funded by NHS commissioners (primarily clinical commissioning groups (CCGs) and NHS England), as well as advisory clinical guidelines and public health guidance (which commissioners are not obliged to implement). CCGs are legally required to make funding available for drugs and treatments recommended by NICE as part of a technology appraisal within three months of NICE's final guidance being published.[14] NICE guidance informs decisions on funding in England, Wales (with the All Wales Medicines Strategy group) and Northern Ireland. The Scottish Medicines Consortium reviews all newly licenced drugs to provide guidance on funding in Scotland.

NICE Highly Specialised Technology (HST) evaluations are recommendations on the use of new and existing highly specialised medicines and treatments within the NHS in England.[15] The programme only considers drugs for very rare conditions. Following consideration of the drug (the process is detailed on the NICE website) the final recommendation will be issued in NICE guidance.

In 2014 the Scottish Medicines Consortium (SMC) developed a new system for appraising medicines for rare diseases. An outline of the new process is provided in this document, Process Changes for End of Life and Very Rare Conditions (orphan and ultra-orphan medicines), and more information is provided on the SMC Website.

The Rare Disease Strategy

In November 2013 the UK Department of Health and the devolved Governments published the UK Strategy for Rare Diseases[16]. This is the UK's first strategy to help build understanding of rare diseases and boost research to find effective treatments and therapies. It sets out 51 commitments to be delivered by 2020, to address 5 key areas:

- empowering those affected by rare diseases

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12 HC Deb 20 January 2015 c29WH
13 NHS England, *Interim Funding policy: Individual Funding Requests*, April 2013
14 NICE, What we do
15 NICE, Highly Specialised Technologies guidance
16 Department of Health, *The UK Strategy for Rare Diseases*, November 2013

- developing better methods of identifying and preventing rare diseases;
- improving diagnosis and earlier intervention for those with a rare disease;
- developing better coordination of care for those with a rare disease, including joined up consultation and treatment schedules; and,
- building on research to improve personalised approaches to healthcare for those with a rare disease.

The strategy also highlighted the importance of collaborative working with patients, their families, industry, research and international partners.

All 4 UK countries committed to producing implementation plans to provide more information about how the strategy will be put into action. These implementation plans have already been published in Scotland[17] and Wales[18]. The Northern Ireland Department of Health, Social Services and Public Safety (DHSSPS) published a Statement of Intent[19] in June 2014 and have developed a draft Northern Ireland Rare Diseases Implementation Plan. This has been subject to a public consultation[20] which closed in January 2015. In England, NHS England is responsible for implementing the rare disease strategy. It published a statement of Intent in February 2014 that provides information on how its commitments will be achieved and working with partners to address the other commitments within the strategy[21]. It has stated that more detailed planning for rare diseases will be provided in the NHS England Five year strategy for specialised services, which is currently under development.[22]

17 Scottish Government, *It's not rare to have a rare disease: The Implementation plan for rare diseases in Scotland* ,June 2014
18 Welsh Government, *Welsh Implementation plan for rare disease*s, February 2015
19 DHSSPS, Northern Ireland Statement of Intent for the UK Strategy of Rare Diseases, June 2014
20 DHSSPS, NI Rare Diseases Implementation Plan - Public Consultation, October 2014
21 UK Strategy for rare disease, *NHS England statement of intent*, February 2014
22 NHS England, News: *NHS England sets out plans for delivering commitments in UK Strategy for Rare Diseases*, February 2014

2. Parliamentary Material

This section provides a selection of recent Parliamentary Questions and other Parliamentary material.

2.1 Parliamentary Questions

Asked by: Nokes, Caroline When he expects NHS England to reach a decision on access to Translarna for the treatment of Duchenne muscular dystrophy; and if he will make a statement.

Asked by: Campbell, Ronnie When he expects NHS England to reach a decision on access to Translarna for the treatment of Duchenne muscular dystrophy; and if he will make a statement.

Answering Member: Gummer, Ben, Department of Health NHS England is considering the interim commissioning position for Translarna as part of its wider prioritisation process for funding in 2015-16 and expects to come to a decision by the end of this month. Translarna has also been referred for evaluation by the National Institute for Health and Care Excellence's highly specialised technologies programme. Draft NICE guidance will be available later this year, with final guidance expected in February 2016.

Caroline Nokes: I thank the Minister for that response and welcome him to his place. Yesterday my constituent Jules Geary came to see me regarding her son Jagger, who suffers from Duchenne muscular dystrophy. Jagger had been approved for Translarna treatment but then suddenly found that it had been withdrawn at the last moment. Like many other boys, he is now waiting, not knowing when a treatment that will prolong his mobility will be forthcoming. Will my hon. Friend meet me, Jules and Muscular Dystrophy UK to discuss how this process can be streamlined so that other children do not have to wait this long?

Ben Gummer: Muscular dystrophy is a terrible, debilitating illness and my sympathies go out to Jagger and his family. My hon. Friend will be aware that families and their representatives will be going to Downing Street on 10 June to make their representations on this matter. The Minister for Life Sciences has introduced an accelerated access review precisely because of the concerns that my hon. Friend has raised, and I know that he will welcome representations once it has been completed.

Mr Ronnie Campbell: Is the Minister aware of the case of my constituent, little George Pegg? At one time he could not walk, but this drug has made his life 100% better and he can now walk. Why are we dithering? This has been going on for at least a year, so why don't you get off that backside of yours and get it approved?

Mr Speaker: Order. May I just exhort Members to have some regard to considerations of taste? This is a new Minister. I call Minister Gummer.

Ben Gummer: I thank the hon. Gentleman for his question. In relation to posteriors, it is good to see his in its rightful place. I have heard of his constituent's case, which is as distressing as that of Jagger and of all

those suffering from Duchenne muscular dystrophy. It is a terrible disease that causes lasting pain to the sufferers and their families. That is precisely why we are pushing hard for a decision from NHS England by the end of this month—it could not have come as quick as he had hoped—and for interim NICE guidance by the end of this year. I am pushing officials to move as quickly as they can on this.

Greg Mulholland (Leeds North West) (LD): The reality is that NHS England has failed to respond to letters or to turn up for meetings, and it has behaved in an utterly unaccountable manner in regard not only to Translarna but to Vimizim, which is used to treat Morquio syndrome. We have still not had confirmation that an interim decision will be made on 25 June, but we are now being told that there will be a decision from NICE on 5 June. Will Ministers finally get a grip on this and give the families affected by these various conditions some sense of when they might get the treatment that could improve their quality of life?

Ben Gummer: I am sorry to hear that the hon. Gentleman has had that experience with NHS England. My hon. Friend the Minister for Life Sciences will want to speak to him about that; if it is the case, it is clearly unacceptable. As the hon. Gentleman will have heard from my previous answer, we are hoping to get quick decisions from NHS England on the interim commissioning guidance this month, and I am pushing hard for a decision from NICE as soon as possible this year, so that we can get interim guidance from it.

HC Deb 2 Jun 2015 : Column 438

Asked by: Mulholland, Greg

To ask the Secretary of State for Health, if he will take steps to commit to the referral of ultra-orphan drugs to NICE Highly Specialised Technologies; and if he will give a target date for this action.

Answering member: George Freeman | Department: Department of Health

We have referred a number of high cost, low volume technologies to the National Institute for Health and Care Excellence (NICE) for evaluation under its highly specialised technologies work programme. We will continue to do so for technologies that meet the specified criteria and where a NICE evaluation would add value. Only today, Ministers referred the drug Translarna (ataluren) for Duchenne muscular dystrophy, to NICE's work programme.

Further information about the process by which topics are selected for referral to NICE's highly specialised technologies work programme is available at:

www.nice.org.uk/about/what-we-do/our-programmes/topic-selection

HC Deb 26 March 2015 | PQ 228646

Asked by: Mulholland, Greg

To ask the Secretary of State for Health, what steps he has taken to help early diagnosis of (a) lysosomal acid lipase deficiency and (b) other very rare diseases; and what research his Department has commissioned into innovative medicines for ultra-rare diseases which have a substantial medical impact.

Answering member: Jane Ellison Department of Health

The UK Strategy for Rare Diseases commits all four countries of the United Kingdom to work to achieve reduced times for diagnosis of rare diseases and to work with the National Health Service and clinicians to establish appropriate diagnostic pathways which are accessible to, and understood by, professionals and patients.

NHS England commissions a highly specialised service for liver disease in children, including cirrhosis resulting from lysosomal acid lipase deficiency, from three expert centres in London, Birmingham and Leeds. These long established centres provide a clear referral pathway for expert diagnosis and management and are well known to paediatricians across England.

The Department's National Institute for Health Research (NIHR) funds a range of research relating to medicines for very rare diseases. The NIHR Rare Diseases Translational Research Collaboration (TRC) provides world-class NHS research infrastructure to support fundamental discoveries and translational research on rare diseases. At its core, this TRC is formed from NIHR biomedical research centres and units, and NIHR clinical research facilities, all with research expertise into rare diseases, facilities and capacity. With investment of £20 million over four years, the NIHR Rare Diseases TRC will help to increase research collaboration across the NIHR infrastructure.

In March 2014, the Government launched the NIHR BioResource to provide a national cohort of healthy volunteers, patients and their relatives who wish to participate in experimental medicine research, the initial focus for the BioResource includes rare diseases. These studies will have the potential to rapidly advance the understanding of disease mechanisms, identify potential drug targets, and improve insight into the therapeutic potential and limitations of existing and emerging therapies.

Written Question 228647 [Diseases] 26 March 2015

Asked by: Baroness Masham of Ilton

To ask Her Majesty's Government what steps they are taking to ensure that commissioning routes in respect of treatment for ultra-rare diseases are as quick and thorough as possible.

Answering: Earl Howe, Department of Health

NHS England has responsibility for the commissioning of specialised services and is currently consulting on changes to the principles and processes by which it makes decisions on prioritising investment in specialised services.

NHS England has provided assurance that any prioritisation which is urgent on clinical grounds will continue to be dealt with quickly through their existing procedures. The route for individual funding requests remains in place.

The consultation can be found at:

www.engage.england.nhs.uk/consultation/investing-in-specialised-commissioning

HL Deb 25 March 2015

Asked by: Mulholland, Greg

This is the third time in four months that I have raised at Prime Minister's questions NHS England letting down the 180 or so people with ultra-rare diseases, some of whom are outside the House today, who have been failed by a flawed process. Some of those children will lose access to their drugs from May, and their conditions will deteriorate irreversibly. We have two sessions of Prime Minister's questions left. Can he tell me that, in that time, he will announce when we will get interim funding for the drugs that these children and these people need?

Answered by: The Prime Minister

My hon. Friend is absolutely right to raise this issue, because these are very rare and debilitating conditions, and there are drugs that can help the children who have them. Having looked at this—and I know that the health and science Ministers have looked vary carefully at it and met the families and the drug companies, as well as NHS England—my understanding is that NHS England is holding a review, which will be completed by the end of April, and the companies are currently funding these drugs until the end of May. So I do not see any reason why there should not be continuity of care and continuity of drugs, and that is what I hope we can achieve.

HC Deb 11 March 2015 C292

Asked by: Huppert, Dr Julian

To ask the Secretary of State for Health, what steps he is taking to ensure that emerging treatments for Duchenne muscular dystrophy which receive conditional approval are available through the Early Access to Medicines Scheme as early as possible; what steps he is taking to encourage flexible approaches to the licensing process for potential new treatments for rare conditions; what progress NHS England has made on development of a commissioning policy for the Translarna treatment for Duchenne muscular dystrophy; and if he will make a statement.

Answering member: George Freeman | Department: Department of Health

The Early Access to Medicines Scheme (EAMS) aims to give patients with life threatening or seriously debilitating conditions access to medicines that do not yet have a marketing authorisation when there is a clear unmet medical need. The Medicines and Healthcare products

Regulatory Agency (MHRA) is responsible for the scientific aspects of the scheme and the scientific opinion will be provided after a two-step evaluation process:

- step I, the promising innovative medicine (PIM) designation

- step II, the early access to medicines scientific opinion

The PIM designation will give an indication that a product may be eligible for the EAMS (based on early clinical data) and that the development programme is on track. The PIM designation will be issued after an MHRA scientific meeting and could be given several years before the product is licensed.

The scheme is voluntary and the opinion from MHRA does not replace the normal licensing procedures for medicines.

There have been no applications from companies with products for Duchene Muscular Dystrophy.

The opinion will support the prescriber and patient to make a decision on whether to use the medicine before its licence is approved. The EAMS scientific opinion is valid for one year in the first instance and lapses at this time or at the time of the grant of a marketing authorisation e.g. conditional marketing authorisation.

For certain categories of medicines going through the centralised marketing authorisation procedure (European procedure), in order to meet unmet medical needs of patients and in the interest of public health, it may be necessary to grant marketing authorisations on the basis of less complete data than is normally required. In such cases, it is possible to recommend the granting of a marketing authorisation subject to certain specific obligations to be reviewed annually, a conditional approval. The granting of a conditional marketing authorisation will allow medicines to reach patients with unmet medical needs earlier than might otherwise be the case.

A conditional marketing authorisation for the first in class medicinal product Translarna (ataluren) was granted this year. Translarna is an orphan medicinal product that is used to treat patients aged five years and older with Duchenne muscular dystrophy (DMD) who are able to walk. Translarna is expected to slow down the loss of walking ability in DMD patients. As part of the conditional marketing authorisation, the company will be required to provide comprehensive data on the efficacy of Translarna from an ongoing confirmatory study.

Rare diseases are classified as conditions affecting no more than 5 in 10,000 people in European Union and patients with rare conditions deserve the same quality, safety and efficacy in medicines as other patients with more common conditions. Since the pharmaceutical industry has little interest, under normal market conditions, in developing and marketing medicines intended for small numbers of patients (orphan medicinal products), the European Union offers a range of incentives to encourage the development of these medicines in order to address the unmet clinical need (orphan drug legislation, Regulation (EC) No 141/2000). These incentives include a period of 10

years market exclusivity, the provision of Protocol Assistance (scientific advice specifically tailored for orphan medicinal products) and fee reductions and waivers for regulatory procedures. Products intended for treatment of Duchenne muscular dystrophy qualify for incentives in the orphan drug legislation.

Applications for the designation of orphan medicines are reviewed by the European Medicines Agency through the Committee for Orphan Medicinal Products (COMP). For orphan designation, the following criteria must be fulfilled. The medicinal product is intended for the diagnosis, prevention or treatment of a life-threatening or chronically debilitating condition affecting no more than 5 in 10,000 persons in the European Union or without incentives it is unlikely that expected sales of the medicinal product would cover the investment in its development and no satisfactory method of diagnosis, prevention or treatment of the condition concerned is authorised, or, if such method exists, the medicinal product will be of significant benefit to those affected by the condition. Via the MHRA, the United Kingdom takes an active role in the decision making process at the COMP, ensuring applications for Orphan Drug designation of potential drug candidates for rare diseases are appropriately recognised, encouraging companies to develop their products further. For licensing, it is compulsory for designated orphan medicinal products to use the centralised procedure to gain a marketing authorisation.

The UK is fully represented at the Committee on Human Medicinal Products, ensuring that applications for a Marketing Authorisation (MA) for an Orphan Drug are thoroughly and rapidly evaluated for quality, safety and efficacy and a MA is granted without undue delay for the treatment of rare diseases.

In some circumstances, marketing authorisations may undergo a more rapid regulatory review called ‘accelerated assessment’. This occurs where the Applicant can demonstrate that the medicinal product is expected to be of major public health interest (particularly from the point of view of therapeutic innovation). For drugs for rare diseases, marketing authorisation applications may be granted as a conditional authorisation or an authorisation under exceptional circumstances. The granting of a conditional marketing authorisation allow medicines to reach patients with unmet medical needs earlier than might otherwise be the case, and ensures that additional data on a product are generated, submitted, assessed and acted upon. Under exceptional circumstances, the MA Applicant must demonstrate that he is unable to provide comprehensive data on the efficacy and safety under normal conditions of use, because, for example, the indications for which the product in question is intended are encountered so rarely that the Applicant cannot reasonably be expected to provide comprehensive evidence.

NHS England can confirm that a draft clinical commissioning policy for Translarna treatment for Duchenne muscular dystrophy has been developed and is being considered as part of the annual funding prioritisation process for 2015-16.

2.2 Other Parliamentary material

Debates

Health: Rare Diseases

House of Lords Questions for short debate HL Deb 3 March 2015 | HL Deb 760 cc170-187

Drugs (Ultra-rare Diseases)

Commons Adjournment debates 20 January 2015 | Vol 591 cc22-31WH

NHS Specialised Services

Backbench Business Debate 15 January 2015 c335WH

Health: Innovative Medicine

Lords question for short debate on what steps they are taking to ensure early access to innovative medicine for life-threatening conditions.

HL Deb 01 April 2014 | Vol 753 cc906-922

Statements

Written statement: Innovative Medicines and Med-tech Review

HC Deb 20 Nov 2014 | Vol 588 c14WS

Written Statement: Rare Diseases Strategy

HC Deb 22 November 2013 c66WS

Early Day Motions

Patients with ultra-rare diseases and powers of the Health Secretary

EDM 918 24 March 2015, Mulholland, Greg

That this House notes the Public Administration Select Committee meeting of 25 February 2015 where the Secretary of State for Health referred to the document he uses to hold NHS England to account, the Framework Agreement between the Department of Health and NHS England; also notes that the Health Secretary went on to say in reference to the document that if NHS England is failing on that, we can hold them accountable on that; further notes that paragraph 4.11.3 of the Framework Agreement says if the Secretary of State considers that NHS England is significantly failing in its duties and functions he is able to intervene and issue directions to NHS England; also notes the cross-party Early Day Motion 819 thus far signed by 61 hon. Members; reiterates its strong concern at NHS England's bureaucracy blocking access to drugs to 180 patients with the ultra-rare diseases Morquio

disease, Duchenne muscular dystrophy and tuberculosis sclerosis; further notes that this matter has been raised repeatedly with the Prime Minister, Deputy Prime Minister, Health Secretary and Life Sciences Minister; further notes that Ministers are constantly saying that resolving this situation is a matter for NHS England; however is dismayed at the lack or urgency on the part of NHS England; therefore calls on the Health Secretary to immediately intervene as the Framework Agreement allows him to and announce interim funding for the drugs these 180 patients urgently need.

Access to treatments for Duchenne Muscular dystrophy, Tuberous Sclerosis and Morquio Disease

EDM 819, 25 February 2015, Mulholland, Greg

That this House is aware that the pause in NHS England's decision-making process on prioritisation and specialised commissioning has created a block to accessing effective medicines for rare diseases where there are commissioning policies under development or assessment; notes that for conditions such as Duchenne muscular dystrophy, tuberous sclerosis and Morquio disease, where a cohort of more than five patients exists, there is no alternative viable route to NHS funding; further notes that as a result patients with need of treatment are left at risk of significant harm, including organ failure and avoidable death; also notes that around 180 patients with Duchenne muscular dystrophy, tuberous sclerosis and Morquio disease are currently being denied access; believes it is unethical to place patients at risk as a result of process inefficiencies; and calls on the Government to bring into force, without delay, interim measures to provide children and adults with rare diseases access to vital and proven medicines that will prevent organ deterioration and premature death.

Access to Translarna through the NHS

EDM 639 17 December 2014, Anderson, David

That this House is deeply concerned at the decision of NHS England to halt its assessment of the Duchenne muscular dystrophy treatment, Translarna, whilst it reconsiders how it assesses new treatments; understands that Translarna, which received conditional approval from the European Commission (EC) in July 2014, is the first drug to treat an underlying genetic cause of Duchenne muscular dystrophy; notes that under the terms of the EC's conditional approval Translarna would be made available to boys whose Duchenne is caused by a nonsense mutation, who are over five and can still walk; further notes that NHS England has taken its decision at a very late stage of the assessment process; acknowledges the deep concern of families at this delay, who fear that it could prevent their sons from accessing Translarna before they lose the ability to walk; and joins the calls of the Muscular Dystrophy Campaign for NHS England to take emergency steps to ensure that Translarna is made available to the children it could help by April 2015.

3. Press notices

This section will include a small selection of recent and relevant press notices.

European Medicines Agency

Rare Disease Day 2015

More medicines becoming available for patients who previously had no treatment options

26 February 2015

The European Medicines Agency (EMA) supports Rare Disease Day 2015, taking place on Saturday 28 February.

A rare disease affects not more than 5 in 10,000 people; however, altogether about 30 million people suffer from these diseases in the European Union (EU).

The Agency plays an important role in the development and authorisation of medicines for rare diseases, known as orphan medicines. EMA's Committee for Orphan Medicinal Products (COMP) issues recommendations to grant orphan designation to medicines, and marketing-authorisation applications for designated orphan medicines are assessed by EMA rather than in each Member State separately.

Companies that have been granted an orphan designation for their medicine benefit from a number of incentives, including reduced fees for marketing-authorisation applications, scientific advice (protocol assistance) and paediatric investigation plans, as well as protection from market competition once the medicine is authorised through a 10-year market-exclusivity period. These incentives aim to bring more medicines for rare diseases to patients.

Since the entry into force of the EU orphan regulation in 2000, more than 100 medicines for rare disease have been brought to the EU market.

A number of medicines that were recommended for marketing authorisation over the past year provide first ever therapies for patients with no treatment options. Among them are:

- the first medicine for a sub-group of patients with Duchenne muscular dystrophy (Translarna);
- the first treatment for erythropoietic protoporphyria, a rare genetic disease which causes intolerance to light (Scenesse);
- the first treatment for Morquio A syndrome, a very rare and life threatening disease (Vimizim);
- the first medicine for the treatment of multicentric Castleman's disease, a chronically debilitating and life threatening condition (Sylvant).

2014 also saw the first recommendation of a therapy based on stem cells, Holoclar, for the treatment for limbal stem-cell deficiency, a rare eye condition that can result in blindness.

A number of medicines have been recommended for the treatment of rare cancers, including Gazyvaro, Imbruvica and Zydelig. Targeted treatments for melanoma and breast-cancer patients whose cancer has a specific mutation (Mekinist and Lynparza, respectively) were also recommended for approval in 2014.

EMA expects that 2015 will also bring more medicines to patients with a rare disease. The Agency started the evaluation of 21 applications for the marketing authorisation for orphan designated medicines in 2014, compared to 14 in 2013.

Involving patients

It is a priority for EMA to ensure that medicines brought to the market are meaningful to patients and respond to their needs. This is why patients are involved in a number of EMA activities so that their views, real-life experience and preferences can be taken into account.

In 2014, a pilot project was launched to involve patients in the assessment of the benefits and risks of a medicine during Committee for Medicinal Products for Human Use (CHMP) plenary meetings. The first medicine to be included in the pilot was Scenesse (afamelanotide), a medicine for the treatment of erythropoietic protoporphyria, a rare genetic blood disorder which causes intolerance to light. Patients with this disease shared their experiences of living with the condition and answered specific questions from the Committee. Their inputs were considered by the CHMP as part of its assessment of the medicine.

The COMP has had a patient representative as Vice-chair since its creation in 2000. Patient representatives are also increasingly involved in scientific-advice meetings with medicines developers so that they can bring their unique perspective on the clinical development plans. They are also involved as experts in the various scientific advisory groups, which provide specialised advice to the Agency's scientific committees on the benefit-risk evaluation of specific types of medicines or treatments.

About Rare Disease Day

Rare Disease Day is held every year on the last day of February. Launched in 2008, it seeks to raise awareness of rare diseases, and to improve access to treatment and medical representation for individuals with rare diseases and their families. It is coordinated by the European Organisation for Rare Diseases (EURORDIS).

The Rare Disease Day 2015 theme 'living with a rare disease' recognises the millions of families, friends and carers whose daily lives are impacted by rare diseases. The 2015 slogan 'day-by-day, hand-in-hand' calls for solidarity as together patients, families, carers, patient organisations and healthcare professionals can participate in the improvement the lives of people living with a rare disease.

NHS England

News

NHS England responds to patient feedback and consults on prioritising specialised services

27 January 2015

NHS England today launches a new consultation about how it will prioritise which specialised services and treatments to invest in. Patient groups asked it to consult on changes to the principles and process by which it makes these decisions.

NHS England directly commissions around 145 specialised services. In order to ensure the maximum number of patients benefit from new innovative treatments coming on stream, choices need to be made about which of these to fund.

In carrying out a public consultation, NHS England will ensure the principles and process for making these decisions are well informed, evidence-led and in line with the expectations of patients and the public. The consultation will last for 90 days from 27 January 2015.

Richard Jeavons, Director of Commissioning Specialised Services at NHS England, said: "We want to ensure patients have access to the very best innovative, evidence-based treatments and services that are being developed all the time. We need to get the most out of every pound for the benefit of patients. They asked us to consult on the principles and process behind making these decisions, which is exactly what we are doing."

This process is looking at the specialist treatments and services that will be routinely available for groups of patients on the NHS. Clinicians, on behalf of their patients, will continue to be able to make a request (an Individual Funding Request) to NHS England for treatment that is not routinely available.

NHS England is also undertaking an engagement exercise to seek views on which specialised services should be prioritised for 'service reviews' as part of a rolling programme of reviewing how each specialised service is delivered. NHS England is writing to all providers of specialised services, Clinical Reference Groups and associated patient groups seeking their views on where to concentrate efforts over next 12 – 24 months. A number of engagement events and workshops are also being planned. Views on which areas to prioritise for service reviews can also be emailed to us.

NHS England also said today that it has listened to patients and clinicians and would continue national commissioning of specialist renal and morbid obesity services in 2015/16. It said it will keep under review whether to transfer responsibility to clinical commissioning groups, but any changes would not happen before April 2016.

Commenting on the decision to continue commissioning specialist renal and morbid obesity services at a national level, Richard Jeavons added: "We all want what is best for patients, and we have listened extremely carefully to their views and those of CCGs. For the time being we will continue to directly commission these services, which will give us head

room to work through the proposals in more detail and address the issues that have been raised with us."

Department of Health

Press release

Better support, treatment and research for millions of patients with rare diseases

22 November 2013

The first UK Rare Diseases Strategy was launched by Health Minister Lord Howe today.

Better support, treatment and research for millions of patients with rare diseases

The UK's first strategy to help build understanding of rare diseases and boost research to find effective treatments and therapies was launched by Health Minister Lord Howe today.

The UK Rare Diseases Strategy aims to ensure that none of the three million people in the UK who are affected by rare diseases are left behind.

For the first time, it sets out a UK-wide vision for building on our reputation as a world leader in rare disease research, including revolutionary genomic research to help transform diagnosis and treatment.

Key elements of the strategy include:

- a clear personal care plan for every patient that brings together health and care services, with more support for them and their families
- support for specialised clinical centres to offer the best care and support
- better education and training for health and social care professionals to help ensure earlier diagnosis and access to treatment
- promoting the UK as a world leader in research and development to improve the understanding and treatment of rare diseases

Health Minister Lord Howe said:

Millions of people in the UK are affected by the thousands of different kinds of chronically debilitating illnesses that are defined as rare diseases. So when looked at as a whole, rare diseases are not rare and our focus should be on making sure that no one who suffers from one is left behind.

For the first time, we are strengthening the links between research and the treatment and care of patients with rare diseases. This is about putting those patients first, with better diagnosis, treatment and support for them and their families.

The UK already leads the way with ground-breaking research to better understand and treat these illnesses and this strategy will help cement our reputation as the driving force in this field.

A rare disease is a life-threatening or chronically debilitating disease that affects five people or fewer in 10,000, and requires special, combined efforts to enable patients to be treated effectively.

There are already more than 5,000 rare diseases identified and the total is steadily rising as new genetic discoveries explain previously unexplained disease patterns. It is estimated that one in 17 people will suffer from a rare disease in the course of their lifetime. In the UK, this means more than three million people will have a rare disease.

Therefore, when looked at as a whole, rare diseases are not rare. They represent a significant cause of illness, making considerable demands on the resources and capacity of the NHS and other care services.

The United Kingdom is a recognised leader in the field of rare disease research, treatment and care. The genomics revolution has the potential to radically transform the diagnosis and treatment of patients with rare diseases over the next ten years. Last year, the Prime Minister, David Cameron, announced that the personal DNA code — known as a genome — of up to 100,000 patients or infections in patients will be sequenced over the next five years. This will improve understanding, leading to better and earlier diagnosis and personalised care.

The project is being led by Genomics England, a company entirely owned by the Department of Health, and rare diseases is one of its key priorities.

In parallel with the launch of the UK Rare Diseases Strategy, the Government announced that the National Institute for Health Research (NIHR) is establishing a Rare Diseases Translational Research Collaboration (TRC). With investment of £20 million over four years, the NIHR Rare Diseases TRC will help to increase research collaboration that will lead to improved diagnosis, treatment and care for people with rare diseases.

Alastair Kent OBE, Chair of Rare Disease UK, which was part of the stakeholder forum that helped shaped the strategy, said:

Patients and families affected by life limiting rare diseases have often struggled to get a diagnosis and to access services that meet their needs. While the NHS has demonstrated that it is capable of delivering a world class service, the experience of patients has been that this is by no means a universal experience.

With the development of the UK Strategy for Rare Diseases patients and families can have a clear expectation of what to expect from the NHS, wherever they live in the UK. For the first time the four nations of the UK have come together to recognise and respond to the needs of rare disease patients systematically, structurally and in accordance with the founding principle of the NHS that treatment should be a response to need.

Turning the UK Strategy's aims into practical benefits for patients will require hard work and detailed planning. Now, at least, patients can have a clear expectation of what the NHS aspires to provide for them, wherever they live in the UK. This is a huge step forward, and the publication of the Strategy will give hope and comfort to the 3.5 million people affected by rare diseases across the UK.

To mark the launch of the strategy, Lord Howe visited Great Ormond Street Hospital, which hosts a large number of nationally commissioned services for rare diseases.

Professor David Goldblatt, Director of Clinical Research and Development at Great Ormond Street Hospital and Director of the NIHR Biomedical Research Centre, said:

Many of the children attending Great Ormond Street Hospital suffer from rare diseases. Their families' experience of delays in the recognition of their syndrome or the absence of a clear diagnosis can be profoundly affecting. We look forward to continuing our work, in partnership with local NHS Trust and Primary Care physicians, in improving the experience of families with rare diseases.

Rare diseases are under-researched at present and in response, the NIHR-funded Biomedical Research Centre at GOSH/UCL is now focussing almost exclusively on rare disease experimental research. While Great Ormond Street Hospital has made some great strides, for example in pioneering gene therapy, much more work needs to be done.

Our ambition is to invest in rare disease research and advance understanding and treatments for the benefit of patients across the world. The hospital's charity is building a new Centre for Children's Rare Disease Research for the hospital and UCL Institute of Child Health, which will play a key role in developing therapies for many disorders for which no treatment or cure currently exists.

Background information

Click here to view the UK Rare Diseases Strategy.

Rare Disease UK (RDUK) is the UK alliance for people with rare diseases and all who support them. RDUK has over 1,400 members including over 240 patient organisations, individual patients and carers, clinicians, health professionals, researchers and industry.

RDUK is an initiative of Genetic Alliance UK, the national charity of over 160 patient organisations supporting all those affected by genetic conditions.

4. Press articles

This section will include links to a number of recent and relevant press articles.

Leeds MP hails Nick Clegg's decision to look at Morquio case again

ITV News-25 Mar 2015

He urged Nick Clegg to intervene to help get the drugs for the 180 patients in total across the UK affected by rare diseases like Morquio ...

Morquio Syndrome: 'Without the drug, I know I don't have long left'

The Guardian-12 Mar 2015

Anna Eaton has two sons: Archie, 13, and Isaac, 12. Both have Morquio Syndrome, also known as MPS IVA, which affects bone development ...

Rare Disease Day 2015: Morquio Syndrome - the condition that ...

International Business Times UK-27 Feb 2015

Only 105 people in Britain and one in 200,000 to 300,000 in the United States are affected by a hereditary condition called Morquio syndrome.

New Scottish funding for rare disease drugs

BBC Online 7 October 2014

Spare a thought for “orphan” drugs: the rare disease medicines that prove health is a numbers game

The New Statesman 5 March 2015

Why coordinated care is key for rare disease patients

Health Service Journal 13 February 2014

5. Further reading

European Medicines Agency: Designation of Orphan Drugs

http://www.ema.europa.eu/ema/index.jsp?curl=pages/regulation/general/general_content_000029.jsp

National Institute for Health & Care Excellence Highly Specialised Technologies Evaluation Committee

http://www.nice.org.uk/get-involved/meetings-in-public/highly-specialised-technologies-evaluation-committee

UK Strategy for Rare Diseases

https://www.gov.uk/government/publications/rare-diseases-strategy

Information about the UK Strategy for Rare Diseases and how it will be implemented.

DoH, last updated March 2015

https://www.gov.uk/government/collections/rare-diseases

All Party Parliamentary Group for Muscular Dystrophy, Access to High cost drugs for rare diseases, September 2013

http://www.muscular-dystrophy.org/assets/0004/4260/Access_to_high_cost_drugs_report_FINAL.pdf

UK Rare Disease Forum

https://www.gov.uk/government/groups/uk-rare-disease-forum

Rare Disease UK

http://www.raredisease.org.uk/

Genetic Alliance UK

http://www.geneticalliance.org.uk/

MPS Society

http://www.mpssociety.org.uk/

Muscular Dystrophy UK

http://www.musculardystrophyuk.org/

BRIEFING PAPER
Number 2015/0011, 15 June 2015

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