The Scottish Parliament
Pàrlamaid na h-Alba

HEALTH AND SPORT COMMITTEE

AGENDA

12th Meeting, 2012 (Session 4)

Tuesday 27 March 2012

The Committee will meet at 10.00 am in Committee Room 2.

1. **Decision on taking business in private:** The Committee will decide whether to take items 6 and 7 in private.

2. **Petitions PE1398, PE1399 and PE1401 on access to medicines for orphan diseases and Individual Patient Treatment Requests (IPTRs) - witness expenses:** The Committee will be invited to delegate to the Convener responsibility for arranging for the SPCB to pay, under Rule 12.4.3, any expenses of witnesses who attend to give evidence on the petitions.

3. **Subordinate legislation:** The Committee will take evidence on the National Health Service (Superannuation Scheme and Pension Scheme) (Scotland) Amendment Regulations 2012 (SSI/2012/69) from—

   Nicola Sturgeon, Cabinet Secretary for Health, Wellbeing and Cities Strategy, Chad Dawtry, Director of Policy, Strategy and Development, Scottish Public Pensions Agency, and Eleanor Guthrie, Senior Policy Manager, Scottish Public Pensions Agency, Scottish Government.

4. **Subordinate legislation:** The Committee will consider the following negative instruments—

   The National Health Service (Superannuation Scheme and Pension Scheme) (Scotland) Amendment Regulations 2012 (SSI/2012/69);
   The Community Care (Joint Working etc.) (Scotland) Amendment Regulations 2012 (SSI/2012/65);
   The Community Care and Health (Scotland) Act 2002 (Incidental Provision) (Adult Support and Protection) Order 2012 (SSI/2012/66);
   The National Assistance (Sums for Personal Requirements) (Scotland) Regulations 2012 (SSI/2012/67);
   The National Assistance (Assessment of Resources) Amendment (Scotland) Regulations 2012 (SSI/2012/68);

The National Health Service (Optical Charges and Payments) (Scotland) Amendment Regulations 2012 (SSI/2012/73);
The National Health Service (Free Prescriptions and Charges for Drugs and Appliances) (Scotland) Amendment Regulations 2012 (SSI/2012/74);
The Food Hygiene (Scotland) Amendment Regulations 2012 (SSI/2012/75); and
The Personal Injuries (NHS Charges) (Amounts) (Scotland) Amendment Regulations 2012 (SSI/2012/76).

5. **Petitions PE1398, PE1399 and PE1401 on access to medicines for orphan diseases and Individual Patient Treatment Requests (IPTRs):** The Committee will take evidence from—

   Stephen Nutt, Executive Officer, Rare Disease UK;

   Joan Fletcher, Family Support Officer, Association for Glycogen Storage Disease (UK);

   Lesley Loeliger, Founder and Chair, PNH Scotland.

6. **Petitions PE1398, PE1399 and PE1401 on access to medicines for orphan diseases and Individual Patient Treatment Requests (IPTRs):** The Committee will consider its approach to future scrutiny of the issues raised in relation to the petitions.

7. **Work programme:** The Committee will consider its work programme.

8. **Social Care (Self-directed Support) (Scotland) Bill (in private):** The Committee will consider its approach to scrutiny of the Bill at Stage 1.

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The papers for this meeting are as follows—

Agenda Item 3

National Health Service (Superannuation Scheme and Pension Scheme) (Scotland) Amendment Regulations 2012 (SSI/2012/69)	HS/S4/12/12/1

Agenda Items 3 and 4

Note by the clerk	HS/S4/12/12/2

Agenda Items 5 and 6

PRIVATE PAPER	HS/S4/12/12/3 (P)
SPICe briefing	HS/S4/12/12/4
Correspondence from the Scottish Government	HS/S4/12/12/5
Submission from Rare Disease UK	HS/S4/12/12/6
Submission from the Association for Glycogen Storage Disease (UK)	HS/S4/12/12/7
Submission from PNH Alliance	HS/S4/12/12/8

Agenda Item 7

PRIVATE PAPER	HS/S4/12/12/9 (P)

Agenda Item 8

PRIVATE PAPER	HS/S4/12/12/10 (P)

Health and Sport Committee

12th Meeting, 2011 (Session 4), Tuesday 27 March 2012

Subordinate Legislation Briefing

Negative Instrument

Overview

There are nine negative instruments for consideration at today's meeting.

A brief explanation of the instruments, along with the comments of the Subordinate Legislation Committee, is set out below. If members have any queries or points of clarification on the instrument which they wish to have raised with the Scottish Government in advance of the meeting, please could these be passed to the Clerk to the Committee as soon as possible.

In keeping with existing practice, these instrument have not been provided in hard copy but can be accessed online.

Name	Deadline	Motion to annul	Purpose	Drawn to attention by Subordinate Legislation Committee (SLC)?
Community Care (Joint Working etc.) (Scotland) Amendment Regulations 2012 (SSI/2012/65)	25 April	No	These Regulations amend the Community Care (Joint Working etc.) (Scotland) Regulations 2002 ("the 2002 Regulations") to specify new functions which may be delegated between NHS bodies and local authorities. The Regulations also make some adjustments to the accounting requirements for arrangements under sections 13 to 15 of the Community Care and Health (Scotland) Act 2002 ("the 2002 Act").	No attention drawn

Community Care and Health (Scotland) Act 2002 (Incidental Provision) (Adult Support and Protection) Order 2012 (SSI/2012/66)	25 April 2012	No	This Order makes provision which is incidental to the Community Care (Joint Working etc.) (Scotland) Amendment Regulations 2012. Those Regulations amend the Community Care (Joint Working etc.) (Scotland) Regulations 2002 to add various new functions to those functions which may be delegated by both local authorities and NHS bodies to each other. Those amendments include the functions of Part 1 of the Adult Support and Protection (Scotland) Act 2007 ("the 2007 Act").	No attention drawn
National Assistance (Sums for Personal Requirements) (Scotland) Regulations 2012 (SSI/2012/67)	25 April 2012	No	Under section 22(4) of the National Assistance Act 1948, in assessing a resident's ability to pay for residential accommodation, local authorities are required to allow the resident an amount for personal expenses which is usually increased each April at the same time as Social Security benefits are uprated. The amount is the same for residents whether they are placed in local authority or independent sector homes. It is proposed that from 9 April 2012 the minimum weekly rate of this allowance will increase in line with average earnings from £22.60 to £23.50.	No attention drawn

National Assistance (Assessment of Resources) Amendment (Scotland) Regulations 2012 (SSI/2012/68)	25 April 2012	No	These Regulations amend the National Assistance (Assessment of Resources) Regulations 1992 ("the principal Regulations"). The principal Regulations concern the assessment of a person's liability to pay for accommodation provided under the Social Work (Scotland) Act 1968 ("the 1968 Act"). By virtue of section 87(3) of the 1968 Act, accommodation provided under the 1968 Act or section 25 of the Mental Health (Care and Treatment) (Scotland) Act 2003 shall be regarded as accommodation provided under Part III of the National Assistance Act 1948.	No attention drawn
National Health Service (Superannuation Scheme and Pension Scheme) (Scotland) Amendment Regulations 2012 (SSI/2012/69)	25 April 2012	No	The instrument amends the National Health Service Superannuation Scheme (Scotland) Regulations 2011 (S.I. 1995/117 "the 1995 Section of the scheme") and the National Health Service Pension Scheme (Scotland) Regulations 2008 (S.S.I. 2008/224 "the 2008 Section of the scheme").	No attention drawn

National Health Service (Optical Charges and Payments) (Scotland) Amendment Regulations 2012 (SSI/2012/73)	25 April 2012	No	These Regulations amend the National Health Service (Optical Charges and Payments) (Scotland) Regulations 1998 ("the principal Regulations") which provide for payments to be made by means of a voucher system, in respect of costs incurred by certain categories of persons in connection with the supply, replacement and repair of optical appliances.	No attention drawn
The National Health Service (Free Prescriptions and Charges for Drugs and Appliances) (Scotland) Amendment Regulations 2012 (SSI/2012/74)		No	These Regulations amend the National Health Service (Free Prescriptions and Charges for Drugs and Appliances) (Scotland) Regulations 2011 ("the principal Regulations"). The principal Regulations provide that where a person provides pharmaceutical services to a patient who presents an English prescription form, that person must make and recover from the patient the charges specified in the National Health Service (Charges for Drugs and Appliances) Regulations 2000.	No attention drawn

Food Hygiene (Scotland) Amendment Regulations 2012 (SSI/2012/75)	26 April 2012	No	These Regulations amend the Food Hygiene (Scotland) Regulations 2006 ("the 2006 Regulations") by updating the definitions of certain EU instruments that are referred to in those Regulations and providing that a reference to those EU instruments shall be an ambulatory reference to them as any annex to them is amended from time to time. In addition, these Regulations extend the availability of remedial action notices, provide for compensation on successful appeal and provide for an identification mark for certain minced meat and other meat products.	The SLC has drawn the instrument to the attention of the Parliament on the reporting ground that it raises a devolution issue. THE SLC Committee Report is available at Annexe A
Personal Injuries (NHS Charges) (Amounts) (Scotland) Amendment Regulations 2012 (SSI/2012/76)	26 April 2012	No	These Regulations amend the Personal Injuries (NHS Charges) (Amounts) (Scotland) Regulations 2006 ("the principal Regulations") which make provision for the charges which a person who pays compensation to an injured person is liable to pay where that injured person has received National Health Service treatment or ambulance services.	No attention drawn

Annexe A

The Scottish Parliament
Pàrlamaid na h-Alba

15th Report, 2012 (Session 4)

Subordinate Legislation

Remit and membership

The Committee reports to the Parliament as follows—

1. At its meeting on 20 March 2012, the Committee agreed to draw the attention of the Parliament to the following instruments—

- Civil Legal Aid (Scotland) Amendment Regulations 2012 (SSI 2012/64); and
- Food Hygiene (Scotland) Amendment Regulations 2012 (SSI 2012/75).

2. The Committee's recommendations in relation to these instruments are set out below.

Food Hygiene (Scotland) Amendment Regulations 2012 (SSI 2012/75) *(Health and Sport Committee)*

19. This instrument amends the Food Hygiene (Scotland) Regulations 2006. The 2006 regulations are the means by which the directly applicable European Union rules on food hygiene are enforced in Scotland. They provide authorisation to various bodies to enforce the EU and national rules and various tools in order to require food business operators to rectify non-compliance. They also create criminal offences in relation to non-compliance.

20. The purpose of this instrument is to amend the 2006 regulations to:

- provide enforcement measures in relation to a number of new directly applicable EU regulations concerning food hygiene;
- provide that amendments to any annexes to the EU instruments specified in Schedule 1 of the 2006 regulations will automatically be captured by the enforcement measures contained in those regulations (without the need for further amending regulations);
- extend the use of remedial action notices as a means of requiring compliance with food hygiene law to food businesses which are not required to be approved (those which do not manufacture products of animal origin);

- provide for compensation to be payable to a food business operator who has suffered loss by complying with a remedial action notice where that notice is subsequently cancelled on appeal;
- specify the form of special health mark which is to be applied to meat from animals which have been subject to emergency slaughter outside a slaughterhouse; and
- provide for enforcement of the rules relating to the sale of meat from animals subject to emergency slaughter.

21. This instrument is subject to the negative procedure. It comes into force on 1 April 2012.

22. In considering the instrument, the Committee asked the Scottish Government for clarification of certain points. The correspondence is reproduced in Appendix 2.

23. The Committee is concerned with the effect of regulation 2(6) and (8) and Schedule 2 of this instrument which insert new regulations 32A and new Schedule 6A into the Food Hygiene (Scotland) Regulations 2006.

24. These new provisions concern the special health mark which is to be applied to meat derived from animals which have been subject to emergency slaughter outside the slaughterhouse. They also create new criminal offences in connection with the marketing of meat products derived from such animals.

25. These provisions are necessary in order to allow for the lawful marketing of meat from such animals from establishments which require to be approved under EU hygiene regulations. (They do not apply to the supply of meat directly to shops which supply meat to the final consumer.)

26. In order to adopt the practice of marketing emergency slaughter meat, member states require to make national measures in a manner that is compatible with the option to do so provided in the EU regulations. In particular, the Committee considers that in order to implement this measure effectively Scots law requires to adopt a uniform approach to UK products. The Committee accepts that these provisions would be capable of doing so if similar national measures existed in the rest of the United Kingdom. Unfortunately, they presently do not.

27. The Committee is concerned that it would not be possible for this difference in approach to the national measure between Scots law and the law of the rest of the UK to be maintained long term. This would not appear to be compatible with what the EU hygiene rules permit.

28. The Committee recognises that to achieve a uniform UK approach requires action in other jurisdictions beyond the scope of the Scottish Ministers' powers. The committee also recognises that this situation is not of the Scottish Ministers' making as the decision not to proceed on the same timetable does not appear to have been notified by the other administrations until it was too late. That does not however address the question of what is to be done about the situation that has arisen. The Committee considers that it is the Scottish Ministers' responsibility to consider whether remedial action should be taken to address this in light of the delay in the rest of the United Kingdom.

29. The Food Standards Agency as the national food safety authority has taken what it considers to be a pragmatic approach to prevent enforcement of these measures and deter prosecutions for non-compliance being brought. It has issued recommendations to that effect to enforcement agencies and the prosecution service. The Committee considers it implicit from this action that the FSA recognises that it can no longer continue with the adoption of the national measure in Scotland in isolation from the rest of the United Kingdom.

30. The Committee notes that it is not of course possible for the FSA to direct how the prosecution service exercises its functions. In addition, the Committee considers that it is not satisfactory to maintain laws which the Government does not intend to enforce whether temporarily or not. It considers that it is an important principle of the criminal law that there is certainty as to which actions are subject to criminal sanction and those which are not. The Committee is therefore most dissatisfied with the approach to resolution of this situation adopted by the Food Standards Agency. The Committee is also dissatisfied with the lack of co-ordination at a United Kingdom level in bringing forward complementary and contemporaneous legislation to achieve a national marking scheme.

31. **The Committee draws this instrument to the attention of the Parliament on reporting ground (f). It raises the following devolution issue.**

32. **The national measures in relation to the specification of the special health mark for meat from animals which have been subject to emergency slaughter and the prohibitions on marketing meat products from such animals set out in new regulation 32A and new Schedule 6A have been brought into force in relation to Scotland in advance of such measures having been made in respect of the rest of the United Kingdom.**

33. **The provision made by this instrument is capable of recognising similar measures in the rest of the United Kingdom but those measures do not presently exist. In the absence of objective justification, it is incompatible with EU law for the United Kingdom as a whole to discriminate between producers in Scotland and the rest of the UK when implementing the national measure provided in respect of emergency slaughter in EU Regulations 853/2004 and 854/2004. It would not therefore be possible to maintain this position indefinitely.**

34. **The Committee acknowledges that this situation occurred as a result of the remaining administrations in the United Kingdom delaying their implementation after the Scottish Ministers had made these regulations rather than as a result of a fault with these regulations themselves. Nevertheless the situation that has arisen requires to be addressed as regards Scotland.**

35. **In recognition of the difference of treatment between Scotland and the rest of the UK which results, the Food Standards Agency will be writing to enforcement authorities and the Crown Office and Procurator Fiscal Service to advise that no enforcement action should be taken until equivalent provisions are in force throughout the UK.**

36. **The Committee considers it most unsatisfactory to adopt a policy of ignoring the effect of subordinate legislation and electing not to enforce it. It is important that there is certainty as regards what actions are to be subject to criminal sanction and those which are not.**

37. **The Committee considers it would be possible to resolve the matter were the implementation of these measures to be postponed until the point at which it has been agreed that identical measures should be made throughout the rest of the United Kingdom.**

APPENDIX 2

Food Hygiene (Scotland) Amendment Regulations 2012 (SSI 2012/75)

On 9 March 2012 the Scottish Government was asked:

(1) When will mutual recognition arrangements for the special mark provided by new regulation 32A and new Schedule 6A be brought into force in the rest of the United Kingdom?

(2) Why it is considered competent under EU law to make provision for a new national measure laying down the format of a special health mark to be applied to carcases of animals which have undergone emergency slaughter outside the slaughterhouse when the measure only applies to Scotland and in the absence of mutual recognition arrangements within the UK national law does not permit such meat to be marketed throughout the UK Member State or provide for the health mark specified for Scotland to be recognised in the rest of the UK?

(3) What is the practical effect of the absence of mutual recognition arrangements for Scottish producers and can prosecutions be brought for non-compliance with the regulations or EU law in this respect?

The Scottish Government responded as follows:

(1) The detailed specifications of the special health mark provided by new regulation 32A and new Schedule 6A have been agreed by the 4 UK administrations and the special health mark will be identical throughout the UK. It was originally intended that all 4 administrations would make implementing provisions to come into force on 1 April 2012, in relation to the special health mark, and all the other provisions now contained within the Food Hygiene (Scotland) Amendment Regulations ("the Scottish Regulations"). England proceeded with 2 separate draft Regulations, to allow the time required for clearance with regulatory committees. In February 2012 they confirmed revised timetables, with the Regulations implementing the provisions relating to hygiene, including the special health mark, to come into force on 1 April, and the Regulations implementing the provisions on remedial action notices, to come into force on 6 April.

In Scotland it was decided to continue to combine into one instrument, to come into force on 1 April 2012, all of the provisions dealing with remedial action notices, food hygiene amendments and the special health mark. This decision was made to avoid the necessity of 2 separate Regulations coming into force on 1 and 6 April, both amending the Food Hygiene (Scotland) Regulation 2006, which it was felt would have been an inefficient use of parliamentary time, which might attract criticism, and in addition, would be confusing to food business operators.

It was only on 2 March 2012, after the Scottish Regulations had been made on 29 February 2012 and laid on 2 March, that the Food Standards Agency in Scotland were advised that the timetables for the English Regulations had slipped further. England now intend to bring into force in May 2012 their Regulations which deal with the special health mark, and those dealing with remedial action notices as soon as

they have passed all parliamentary scrutiny. Wales has considered its position and aims to follow a similar timetable to England. Northern Ireland has considered its position and aims to implement the provisions relating to remedial action notices by 6 April 2012 and those relating to Hygiene, including the special health mark, during May 2012.

Whilst this situation is not ideal, the Scottish Government takes the view that there will still be only a short gap, of a few weeks, when the provisions relating to the special health mark in Scotland are in force, before the provisions for the rest of the UK come into force.

For the intervening period, and as explained in the answer to question 3, below, the Food Standards Agency will be writing to stakeholders, local authorities and COPFS to advise that no enforcement action should be taken until legislation in the rest of the UK has come into force. Once that legislation in the rest of the UK is in force, an identical special health mark will be required throughout the UK. Accordingly, there is no need for a mutual recognition provision and the measure will be fully implemented under EU law.

(2) This instrument provides for a "special health mark" as required by paragraph 9 of Chapter VI of Section I of Annex III to EC Regulation 853/2004 and paragraph 7 of Chapter III of Section I of Annex I to EC Regulation 854/2004. These EC Regulations do not prohibit a mark being implemented in one part of a member state in advance of another part. In terms of EU law, this measure has been implemented in Scotland.

(3) In the meantime, to fully cover the period between 1 April 2012 and the date that equivalent legislation comes into force in the rest of the UK, the Food Standards Agency in its capacity as Central Competent Authority, will be writing to stakeholders, local authorities and the Crown Office and Procurator Fiscal Service, in advance of the application date of 1 April, to advise that no enforcement action should be taken until equivalent provisions are in force throughout the UK. The letter will also give general advice advocating a graduated approach to enforcement for these new provisions, once they are adopted in each of the UK countries.

With respect to the practical effect of the new provisions, in so far as they relate to special health marks applied to carcases of animals subject to emergency slaughter, a square health mark has been in use in the UK for these purposes since 2006 when the EU provisions took effect. FSA guidance is provided in the UK Manual for Official Controls. Application of the special health mark has been custom and practice in all UK countries since then. The Scottish Regulations simply set out the formal legal basis for the format of the mark, as required by the EU, for controls that have been consistently applied since 2006. Therefore there will be no practical effect for the Scottish Regulations providing a new legal basis for this, a few weeks in advance of the rest of the UK.

With respect to the practical effect of the application of the new identification mark on further processed minced meat and meat products and meat preparations derived from emergency slaughtered meat, this will be a new requirement. However the overall number of emergency slaughtered animals is relatively small. A large proportion of these carcases are supplied directly to the final retail butcher for delivery directly to the final consumer sector, and an identification mark will not be

required. In light of the small numbers involved, and the effect of the letter being sent to all local authorities advising against enforcement until the equivalent legislation is in force throughout the UK, there will be no practical adverse effect of the earlier introduction of the legislation in Scotland.

Agenda Items 5 and 6 27 March 2012		HS/S4/12/12/4

HEALTH & SPORT COMMITTEE

CONSIDERATION OF PE1398, PE1399, PE1401 CONCERNING ORPHAN MEDICINES

INTRODUCTION

This briefing aims to provide Members with background information concerning the issues raised by the petitioners. The briefing begins with a discussion of orphan medicine designation, licensing and the process used in Scotland for the approval of medicines for use in the NHS with a focus on orphan medicines. The second part outlines some of the key concerns common to all three petitions and the Government's response to these so far.

ORPHAN MEDICINE DESIGNATION

Orphan designation is handled on an EU basis through EU regulations, where were introduced in the EU in 2000 in an attempt to improve the availability of medicines for rare diseases, or "orphan medicines". The process is handled through the European Medicines Agency (EMA) and designation is granted by the European Commission. The EMA states[1] that in order for a medicine to qualify for orphan designation, a medicine must meet one of two criteria:

1. it is intended for the diagnosis, prevention or treatment of a life-threatening or chronically debilitating condition affecting no more than 5 in 10,000 people in the EU at the time of submission of the designation application
2. it is intended for the diagnosis, prevention or treatment of a life-threatening, seriously debilitating or serious and chronic condition and without incentives it is unlikely that the revenue after marketing of the medicinal product would cover the investment in its development

In both cases, there must either be no satisfactory method of diagnosis, prevention or treatment of the condition concerned, or, if such a method does exist, the medicine must be of significant benefit to those affected by the condition.

Given orphan medicines are intended for small numbers of patients, the pharmaceutical industry is less likely, in normal market conditions, to develop and market such medicines. Therefore, the European Commission offers a number of incentives to pharmaceutical companies to encourage the development of these medicines, including fee reductions, access to the centralised authorisation (or "licensing") procedure (see below) and ten years of market exclusivity once authorised. There may also be a range of other incentives at EU and Member State level.

[1] European Medicines Agency (Online) *Orphan designation*. Available at: http://www.ema.europa.eu/ema/index.jsp?curl=pages/regulation/general/general_content_000029.jsp&mid=WC0b01ac05800240ce&jsenabled=true

LICENSING OF MEDICINES AND ASSESSING THEM FOR USE IN THE NHS IN SCOTLAND

Licensing of medicines

Pharmaceutical companies wishing to bring a medicine to market must apply for a marketing authorisation or "license". The purpose of licensing is to consider whether the medicine has a measurable effect against a placebo or comparator in a clinical trial ("efficacy"), and whether, on balance, the medicine is likely to have an acceptable level of safety and quality. In the UK the licensing body is the Medicines and Healthcare products Regulatory Agency (MHRA).

Pharmaceutical companies have two choices in obtaining a license for the UK – either they can apply for a UK license through the MHRA or can apply through the EMA. If the latter, this is referred to as the "centralised authorisation procedure"[2]. This procedure results in a single marketing authorisation valid in all EU countries, together with Iceland, Liechtenstein and Norway. As noted above, an orphan medicine will be taken through this procedure.

Process for assessing medicines for use in NHS Scotland

When a new medicine is licensed for use, the pharmaceutical company is asked to make a submission on the product, including results of clinical trials and cost effectiveness data, to the Scottish Medicines Consortium (SMC). The SMC's role is to undertake an evaluation of the medicine's clinical efficacy and cost effectiveness, and then determine whether the medicine should be recommended for use in the NHS in Scotland.

In coming to a determination, the SMC has a two stage process[3]. Firstly, its New Drugs Committee (NDC) evaluates the submission with the support of medical, pharmaceutical, and health economics experts. The NDC then makes a provisional recommendation that is shared with the pharmaceutical company concerned. The advice from NDC, together with feedback from the company is then considered by the SMC committee. Patient interest group submissions, focusing on the difficulties the disease presents for patients and the place of the medicine in addressing patient needs, are also taken into account.

Assessment of orphan medicines

In their submissions to the Public Petitions Committee, the SMC[3] and Scottish Government[4] discussed the process for assessing orphan medicines.

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[2] EMA (Online) *Central authorisation of medicines*. Available at: http://www.ema.europa.eu/ema/index.jsp?curl=pages/about_us/general/general_content_000109.jsp&mid=WC0b01ac0580028a47

[3] SMC (2011) *Scottish Medicines Consortium response to the Public Petitions Committee on PE1398, PE1399 and PE1401*. Available at: http://www.scottish.parliament.uk/S4_PublicPetitionsCommittee/General%20Documents/PE1398_F_Scottish_Medicines_Consortium_07.11.11.pdf

[4] Scottish Government. (8 November 2011) Public Petitions committee consideration of PE1398, PE1399 and PE1401. Available at: http://www.scottish.parliament.uk/S4_PublicPetitionsCommittee/General%20Documents/PE1398_H_Scottish_Government_08.11.11.pdf

The SMC notes that for an orphan medicine the submitting company is required to make the case for clinical and cost-effectiveness in the same way as for all new medicine submissions. The SMC has stated that it recognises that efficacy data are very often limited in orphan medicines due to the rarity of the condition, and as such it is willing to accept greater uncertainty in the health economic case when assessing a medicine with an orphan indication.

The health economics tool used to measure the benefit of a medicine is the quality-adjusted life year (QALY). This takes into account how a treatment affects a patient's quantity of life (how long they live for) and the quality of life (the quality of their remaining years of life). These factors are then combined into a single measure that puts a figure on the health benefits for a medicine. The resulting QALY can then be used to benchmark the benefits each medicine is likely to offer. Then, to consider the cost effectiveness of the medicine, the QALY is combined with the cost of the medicine to produce a ratio called the cost per QALY. Both the SMC and Scottish Government assert that this is an accepted method in health economics, and that it is used by both the SMC and NICE in England.

Orphan medicines are more than likely to have a high cost to the NHS. However, the SMC states that there are situations where it may be willing to accept a higher cost per QALY in excess of normal parameters[5]. It has developed "modifiers" to be used when appraising medicines in particular categories, including orphans. These have been created in order to allow the SMC to view the cost per QALY more flexibly, with the potential to recommend a medicine notwithstanding the economic evidence provided. The modifiers include:

- evidence of a substantial improvement in life expectancy
- evidence of a substantial improvement to quality of life
- evidence that a sub-group of patients may derive specific or extra benefit and the medicine can be targeted at this sub-group
- absence of other therapeutic options of proven benefit
- possible bridging to another definitive therapy (e.g. curative surgery)
- emergence of a licensed alternative to an unlicensed therapy

The SMC also states that its overall judgement on a medicine will be influenced by input from clinical experts and patient interest groups, as well as the clinical and cost effectiveness data on the new medicine submitted by the manufacturer.

Patient Access Schemes

Patient Access Schemes were introduced through the Pharmaceutical Price Regulation Scheme (PPRS). The PPRS is the mechanism which the Department of Health (DoH) in England, on behalf of the four UK health departments, uses to regulate the prices of branded medicine. It is a voluntary scheme, usually negotiated every five years, between the DoH and the pharmaceutical industry though is underpinned by statutory powers. It seeks to

[5] A cost per QALY of under £20,000 is generally considered acceptable value for money. For a medicine with a cost per QALY between £20,000 and £30,000 SMC might accept this if the medicine gives significant benefits over existing treatments. Through the application of modifiers to medicines with a cost per QALY of £30,000 may allow its approval in some cases.

achieve a balance between reasonable prices for the NHS and a fair return for the industry to enable it to research, develop and market new and improved medicines.

As noted by the Scottish Government[6], the 2009 PPRS sought the introduction of more flexible pricing options which enable drug companies to improve the value of specific medicines to the NHS. One of these is the Patient Access Schemes (PAS), which offer discounts or rebates to reduce the cost of a drug to the NHS. When submitting a medicine (including an orphan medicine) for consideration by the SMC, manufacturers can propose a PAS in order to improve the cost-effectiveness and availability of the medicine.

The Patient Access Scheme Assessment Group has been established under the auspices of NHS National Services Scotland to deliver a national service to conduct an objective and independent assessment of PAS submitted by pharmaceutical companies. This a process independent of the SMC. Where a PAS is considered feasible, the SMC is able to take account of the discount offered under the terms of the PAS. Where a PAS is not considered feasible, SMC appraises the drug on its standard costs.

The Scottish Government[6] has stated that this scheme has played an important role in helping more patients to access drugs that would not otherwise be assessed as cost-effective by the SMC.

Medicines accepted for use by SMC

Where SMC has accepted a new medicine, NHS Boards are expected to make it (or its equivalent) available. Therefore, NHS Boards will review the medicine in the context of other existing comparable medicines available within the Board's own formulary/approved list to treat the same condition. However, the Scottish Government has recognised the need for a consistent and standardised approach to such considerations. On 13 February 2012 the Chief Medical Officer published guidance, part of which set out a framework for NHS Boards to apply when updating their written policy in relation to NHS Board formulary decision-making for SMC accepted medicines. NHS Boards have been requested to confirm by 1 April 2012 that their policies on formularies have been updated to reflect this additional guidance.

Medicines not recommended for use by the SMC

Where SMC has issued "not recommended" advice in relation to a medicine, NHS Boards are not expected to make it routinely available. However, these medicines, including those medicines not recommended due to a non-submission by the manufacturer to the SMC, can be made available under certain circumstances through individual patient treatment requests (see below). The petitioners are all interested in this process because they believe that it is more likely that patients with rare diseases will have to access this system because the medicines they need are less likely to be approved by the SMC.

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[6] Scottish Government (1 February 2012) *Public Petitions committee consideration of PE1398, PE1399 and PE1401*. Available at: http://www.scottish.parliament.uk/S4_PublicPetitionsCommittee/General%20Documents/PE1398_R_Scottish_Government_01.02.11.pdf

The use of NICE technology appraisals in Scotland

NICE (or the National Institute for Health and Clinical Excellence) is the body which has, amongst its other roles, the responsibility for advising on whether newly licensed medicines should be made available on the NHS in England and Wales. It carries out two main types of assessment on medicines, and only one has a formal status in Scotland:

- Single Technology Appraisal (STA) - is typically where a medicine is assessed for one single indication. As this is similar to the process undertaken by the Scottish Medicines Consortium, STAs have no formal status in the NHS in Scotland.
- A Multiple Technology Appraisal (MTA) - normally covers more than one medicine for one or more indications. The MTA process is more involved and includes an independent assessment of the evidence surrounding a medicine. Experts from Healthcare Improvement Scotland (HIS) are involved in the appraisal process, and when the guidance is finally published, HIS publishes a statement advising whether or not the advice is relevant to Scotland. If it is then this supersedes any advice on a medicine that has been produced by the Scottish Medicines Consortium.

Individual Patient Treatment Requests

Prior to April 2011, there was no formal structure for NHS Boards to make decisions on requests by patients to be treated with a medicine not recommended for use by the SMC. However, it was generally the case that in order for a Board to agree to such a request, the patient would need to be: a) significantly different to the general population of patients with the condition in question; and, b) likely to gain more benefit from the medicine than the average patient. These criteria were referred to as “exceptional circumstances”. NHS Boards had their own procedures for dealing with such decisions.

Following the Public Petition’s Committee Report on the issues raised by petition PE1108, concerning the provision on the NHS of cancer treatment medicines, the Scottish Government published guidance in May 2010. Annex D of this sets out a specific guidance framework for NHS Boards to apply when developing a written policy for what became known as individual patient treatment requests (IPTRs) for medicines not recommended by the SMC. As part of the process NHS Boards were to have written policies in place for dealing with such requests by 1 April 2011.

In March 2011, the Chief Medical Officer (CMO) set out further good practice guidelines on circumstances under which IPTR should be granted and how decisions should be made. This guidance (para 11-12) states that the responsibility for an application for an IPTR rests with the clinician who supports prescribing the requested medicine. It is the clinician who is expected to demonstrate the clinical case for the patient to be prescribed a medicine within its licensed indication(s) where the following criteria apply, namely that the patient’s clinical circumstances (condition and characteristics) are significantly different from either: (i) the general population of patients covered by the medicine’s licence; or (ii) the population of patients included in the clinical trials for the medicine’s licensed indication as appraised. It goes on to

state that these circumstances imply that the patient is likely to gain significantly more benefit from the medicine than would normally be expected. Finally, it states that such considerations should be taken on a "case by case" basis reflecting clinical opinion and, as such, should not be generalised. The Public Petitions Committee received letters from the vast majority of NHS Boards confirming that they had IPTR policies in place.

SPECIFIC MATTERS RAISED BY THE PETITIONERS

Each of the petitioners raises a number of issues specific to those petitions. This section of the briefing considers some of the key matters of common concern to them all. It does not seek to address all the issues raised by the petitioners, all of which are contained in the submissions provided by the petitioners to the Committee.

The SMC assessment process for orphan medicines

Each of the petitioners questions whether the assessment process used by SMC is appropriate for orphan medicines. They note the data supplied to the Public Petitions Committee from the SMC[3] showing that the acceptance rate for orphan medicines submitted to the SMC is 61%, compared to 75% for medicines without orphan status (as at October 2011). In addition, there are concerns that the QALY economic analysis tool is inappropriate for assessing orphan medicines and that it is not clear how the modifiers are applied. Rare Disease UK[7] stated that "evaluation should be based on an appraisal of the technology against multiple criteria and not simply a cost utility analysis". It asked the Scottish Government to review the mechanism and methodology used by the SMC to appraise the value of medicines for orphan diseases.

In its submissions to the Public Petitions Committee, the Scottish Government[4,6] rejected this, arguing that the SMC processes for appraising orphan medicines are "robust and comprehensive". It reiterated the current process and felt it was based on multiple criteria (through the modifiers) and not simply a cost-utility analysis. In its submission the SMC[3], discussing the lower acceptance rate for orphan medicines stated:

> "SMC believes that these figures are reassuring because *de facto* the evidence base for orphan medicines is often weaker than for other medicines, the SMC modifiers described above do not always apply to the medicine under review and the prices charged for these drugs can make it impossible for them to meet conventional measures of good value." (p 3).

However, the Scottish Government agreed to give consideration to existing arrangements for the appraisal of orphan medicines. The latest letter from the Scottish Government states that the matter is still under consideration.

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[7] Rare Disease UK. (8 November 2012) *Submission to Public Petitions Committee*. Available at: http://www.scottish.parliament.uk/S4_PublicPetitionsCommittee/General%20Documents/PE1398_N_Petitioner_01.12.11.pdf

Ultra orphan medicines and national commissioning

As noted above, the standard definition of an orphan medicine is one for the prevention or treatment of a life-threatening or chronically debilitating condition affecting no more than 5 in 10,000 people. However, all the petitioners are concerned about medicines for those rare diseases that affect fewer people (1:50,000 prevalence). For example, the PNH Alliance / PNH Scotland explain that paroxysmal nocturnal haemoglobinuria[8] (PNH) is an ultra rare disease and that the appraisal system of the SMC is weighted against ultra orphan medicines. The petitioners call either for a separate body to appraise "ultra orphan" medicines, or an alternative pathway within the SMC, or both. In particular, they point to the system in England, where there is the Advisory Group for National Specialised Services (AGNSS).

Advisory Group for National Specialised Services

AGNSS will consider a small number of highly specialised new drugs and technologies, usually consisting of no more than 500 patients and/or four centres in England. Its role is to make recommendations to Ministers about whether the drugs and technologies it considers are appropriate for commissioning at a national level. AGNSS only considers drugs and technologies that NICE decides are not suitable for a NICE appraisal because of the very small patient numbers involved. The criteria for referral are shown on the NHS Specialist Service website.

Scottish Government response

The response from the Scottish Government[6] states:

> "Whilst the Scottish Government is aware of the term "ultra orphan" used by NICE, we are not aware of any formal recognition of this term by relevant regulatory agencies and therefore do not believe it necessary for the SMC to develop a policy for medicines that would fall within this description. The SMC's arrangements for appraising orphan medicines including its modifiers will capture those medicines which would be deemed by NICE to come under the description of "ultra orphan"." (Para 35).

The Scottish Government also notes that, in Scotland, national commissioning for highly specialised services is carried out by National Services Division (NSD) of NHS National Services Scotland (NSS). New services for designation or consideration of case(s) for de-designation are considered by the National Services Advisory Group whose advice is in turn provided to Scottish Ministers for decision. These arrangements are currently under review by the National Planning Forum. This should be complete by summer 2012.

Outlining the current situation, the Scottish Government noted that, where it is safe, sustainable and effective to do so, such services are commissioned and provided within NHS Scotland. Where the incidence of the relevant disease or condition is so low that it is not practicable to commission a service in Scotland the National Specialised Commissioning Team (NSCT) commission and provide services on a UK basis, i.e. for the whole of the population of the UK.

[8] Paroxysmal nocturnal hemoglobinuria (PNH) is an acquired blood stem cell disorder.

The Scottish Government further explains that where UK commissioned services are provided for residents in Scotland, these services are most often commissioned and paid for via NSD. However, NHS Boards may also seek appropriate treatment and care for individual patients directly with relevant providers in England, in which case the Board concerned accepts responsibility for payment on an extra contractual basis.

Individual Patient Treatment Requests

The operation of the IPTR process by NHS Boards is of particular concern to the petitioners. The petitions and submissions on behalf of PNH Alliance / PNH Scotland and the Association of Glycogen Storage Disorders both discuss how they believe the process does not adequately take account of the needs of their members with PNH and Pompe disease[9]. The Rare Disease UK submission[10] from February 2012 summarises the key concerns with the process:

> "In rare diseases it is extremely difficult to demonstrate the criteria. The small patient numbers who make up the clinical trial populations are those patients with the greatest clinical need for the drug and therefore the license will be based on this group of patients. It is therefore extremely difficult to show that a patient with genuine clinical need will be "More likely to benefit from the medicine than might be expected for other patients with the condition". The patients who are likely to have the greatest need for the treatment will be the same as those patients within the clinical trials upon whom the license is based. Unlike in some of the more common conditions and even certain cancers where there is often more than one licensed treatment available, in the majority of rare diseases there is likely to be only one licensed treatment available, apart from just supportive care. In orphan diseases the above criteria are therefore more likely to lead to those patients with the greatest clinical need being refused access to therapies, which may be life changing and / or life saving." (p 1).

The petitioners asked for a review of the criteria for assessing IPTRs. In its submission to the Public Petitions Committee on 8 November 2012, the Scottish Government stated that the Chief Medical Officer and Chief Pharmaceutical Officer had been asked to review existing processes for IPTRs as well as other matters. In its submission[6] of 1 February 2012, the Scottish Government advised that the group set up to consider these matters had concluded that the IPTR guidance had produced benefits for NHS Boards and should be allowed to "bed in" before any further changes should be considered. Nevertheless it was felt that a number of "key messages to underpin the extant guidance" would be published. These were published in

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[9] Pompe disease is a metabolic muscle disorder. It is a rare neuromuscular genetic disorder that occurs in babies, children, and adults who inherit a defective gene from each of their parents. The disorder has a number of synonyms, the most common are: Acid Maltase Deficiency and Glycogen Storage Disease Type II.

[10] Rare Disease UK. (23 February 2012) *Submission to the Public Petitions Committee*. Available at: http://www.scottish.parliament.uk/S4_PublicPetitionsCommittee/General%20Documents/PE1398_S_Petitioner_23.02.11.pdf

guidance on 13 February 2012 by the CMO. NHS Boards have been asked to confirm by 1 April 2012 that their policies on the management of IPTRs have been updated to reflect this additional guidance.

In addition, the Scottish Government has stated that it will keep the IPTR guidance under review, and will take forward any recommendations from the Parliamentary consideration of the three petitions as appropriate.

National Plan for Rare Diseases

Rare Disease UK has shown interest in the development and implementation of a National Plan for Rare Diseases. The need for a UK National Plan followed European Council Recommendation of 8 June 2009 on action in the field of rare diseases, which recommended that every Member state should have a national strategy.

The Department of Health in England published a consultation on behalf of the four UK health departments on 29 February 2012. It makes a number of recommendations, including:

- using specialist centres to make exact diagnosis, which will make sure people are treated earlier and in some cases could save lives
- acknowledges that all doctors should have the right training to be aware of the possibility of a rare disease
- recommends that the care of patients with rare diseases should be better co-ordinated

The consultation recognises that where effective new medicines become available for the treatment of rare and very rare conditions, patients are concerned that they should have access to the medicines their doctors recommend. However, it does not discuss or make recommendation on the separate appraisal systems that exist within the four UK countries, rather it discusses the working being undertaken through the PPRS to introducing a new system of pricing for branded medicines, where the price of a drug will be linked to its assessed value.

The consultation closes on 25 May 2012.

Jude Payne
SPICe Research
22 March 2012

HS/S4/12/12/5

Deputy First Minister and Cabinet Secretary for Health, Wellbeing and Cities Strategy
Nicola Sturgeon MSP

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The Scottish Government

Duncan McNeil MSP
Convener of the Health and Sport Committee
The Scottish Parliament
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DELIVERING
A GAMES LEGACY FOR SCOTLAND

XX February 2012

Thank you for your letter dated 25 January 2012 regarding the present system of Individual Patient Treatment Requests (IPTRs) in which you asked for a progress report on the Clinically-led Short Life Working Group (SLWG) which was established at the end of last year to consider how the extant guidance in relation to the managed introduction of new medicines across the NHS in Scotland could be further strengthened.

I can advise that the SLWG held two very well-attended and productive meetings in December 2011 which resulted in consensus agreement on guidance to further strengthen the safe and effective use of new medicines. This guidance has now issued and can be viewed via the following link:
http://www.sehd.scot.nhs.uk/cmo/CMO(2012)01.pdf

I hope this is helpful and would be pleased to provide further information as required.

NICOLA STURGEON

PE1398

Petition by Alastair Kent, on behalf of Rare Disease UK, calling on the Scottish Parliament to urge the Scottish Government to review the mechanism and methodology used by the Scottish Medicines Consortium to appraise the value of medicines for orphan diseases and to instruct the Chief Medical Officer to revise the criteria for accessing Individual Patient Treatment Requests by removing the term 'exceptional' from all health boards IPTR requests in relation to orphan diseases.

Rare Disease UK (RDUK) would like to thank the Scottish Parliament Health and Sport Committee for taking an interest in the issues raised by Public Petition PE1398 with regards Individual Patient Treatment Requests. In addition to the documents submitted by RDUK during the Public Petitions hearings, we thought it would be useful to summarise the views of RDUK regarding access to medicines for orphan diseases in Scotland.

RDUK acknowledges the revised guidelines on the 'end to end' process from licensing of medicines through to the individual patient treatment requests in CEL17(2010) and the further guidance (SGHD/CMO(2012)1). RDUK and our members continue to be concerned that this guidance does little to affect the problem that current criteria for accessing orphan medicines through an Individual Patient Treatment Request (IPTR) is too onerous for patients with rare diseases.

The specific characteristic of rare diseases mean that, for these conditions, the current criteria for IPTRs are extremely difficult to demonstrate. The criteria for IPTRs state that:

The patient's clinical circumstances (condition and characteristics) are significantly different from either:

- The general population of patients covered by the medicine's license; or
- The population of patients included in the clinical trials for the medicine's licensed indication as appraised.'

The small patient numbers who make up the clinical trial populations are also those patients with the greatest clinical need for the drug and therefore, the license will be based on this group of patients. It is therefore extremely difficult to show that a patient with genuine clinical need will be 'more likely to benefit from the medicine than might be expected for other patients with condition". The patients who are likely to have the greatest need for the treatment will be the same as those who patients within the clinical trials upon whom the license is based. Unlike in some of the more common conditions and even certain cancers where there is often more than one licensed treatments available, in the majority of rare diseases there is likely to be only one licensed treatment available. In orphan diseases the current IPTR criteria are therefore more likely to lead to those patients with the greatest clinical need being refused access to potentially life changing therapies.

RDUK calls upon the Health and Sport Committee to consider undertaking a thorough review of the current criteria in relation to the implementation of their IPTR process specifically in regards to orphan medicines. Such a review should involve all relevant stakeholders, including patient organisations, industry, health boards and clinicians.

RDUK would also urge the Health and Sport Committee to consider PE1398's assertion that the current methodology used by the Scottish Medicines Consortium to appraise the cost-effectiveness of medicines does not appraise orphan drugs equitably. A recommendation from the report 'Improving Lives, Optimising Resources: A Vision for the UK Rare Disease Strategy' is that 'Evaluation [for orphan medicines] should be based on an appraisal of the technology against multiple criteria and not simply a cost utility analysis'. Despite the addition of modifiers to the SMC process, there is no significant difference in the distribution of decisions before and after (61% were 'not recommended' in the period 2003-2007 and 63% in the period 2008-2011). RDUK would like the Health and Sport Committee to consider undertaking an open and transparent public review into the SMC's arrangements for the appraisal of orphan medicines and to consider the possibility of using a separate mechanism for appraising orphan or ultra-orphan medicines in Scotland.

We would also like to notify the Committee that a public consultation on a UK plan for rare diseases as referred to previously has been launched by the Scottish Government jointly with the other UK health departments. We have submitted this consultation document.

We look forward to meeting with the Committee on the 27th of March and discussing our concerns about the current system for Individual Patient Treatments and the difficulties in accessing medicines for orphan diseases in Scotland.

We would also like to draw the Committee's attention to:

- Consultation on the United Kingdom Plan for Rare Diseases: http://www.dh.gov.uk/prod_consum_dh/groups/dh_digitalassets/@dh/@en/documents/digitalasset/dh_132883.pdf

- SGHD/CMO(2012)1 - Guidance to further strengthen the safe and effective use of new medicines across the NHS in Scotland: http://www.sehd.scot.nhs.uk/cmo/CMO(2012)01.pdf

Petition PE1399

Petition by Allan Muir calling on the Scottish Parliament to urge the Scottish Government to instruct the Chief Medical Officer (CMO) to revise the criteria to access Individual Patient Treatment Requests (IPTRs) for Orphan diseases as these criteria are detrimental to patients suffering from Pompe disease.

Attached are six documents that Petitioner Allan Muir (PE1399) wishes to draw to the Committee‟s attention to support his petition. The Petitioner has indicated their relevance below:

1. Letter regarding funding for Patient to Nicola Sturgeon from wife of Patient. Myozyme was applied for after the SMC did not recommend the drug for use in Scotland. The Patient eventually got treatment through Exceptional Case Use in May 2008.

2. Letter from Steve Waldek, asking the AGSD for support. In the letter he explains the situation from his point of view as a treating clinician. This is evidence for supporting patients access for treatment from an expert clinician in the field. This also supports the statement in the Scotsman: http://www.scotsman.com/news/family-forced-to-fund-life-saving-treatment-1-1606896
   An SNP spokesperson said: "Everyone recognises the importance of decisions on medication and treatment being made by health professionals and not politicians. When consultants recommend access to medicines we expect health boards to respond flexibly and favourably to requests."

3. Personal letter from Stephen Waldek which is also supported by the written evidence he sent to Ayrshire & Arran NHS Board.

4. The appeal refusal letter from NHS Ayrshire and Arran talks about „opportunity cost implications‟ and not the clinician‟s recommendation.

5. “PB18 Article”, AGSD-UK publication, the Pompe Bulletin, illustrating the plight of untreated Pompe sufferers in Scotland.

6. “The Healthcare Quality Strategy for Scotland: http://www.scotland.gov.uk/Resource/Doc/311667/0098354.pdf which reads:

The Quality Ambitions

- Mutually beneficial partnerships between patients, their families and those delivering healthcare services which respect individual needs and values and which demonstrate compassion, continuity, clear communication and shared decision-making.
- There will be no avoidable injury or harm to people from healthcare they receive, and an appropriate, clean and safe environment will be provided for the delivery of healthcare services at all times.
- The most appropriate treatments, interventions, support and services will be provided at the right time to everyone who will benefit, and wasteful or harmful variation will be eradicated.

Nicola Sturgeon
Cabinet Secretary for Health and Wellbeing
Better Health, Better Care Consultation
Scottish Executive
St Andrew's House
Edinburgh
EH1 3DG

15 October 2007

THE CHALLENGE FOR HEALTH AND WELLBEING

I write on behalf of Scottish families, including my own, who live, as a sufferer or carer, with Pompe Disease, a very rare debilitating, often fatal, condition. There is at long last hope for those suffering with this disease as an Enzyme Replacement Therapy (ERT), Myozyme, is now available however not to patients who live in Scotland.

The Scottish Medicines Consortium (SMC) took the decision earlier this year not to recommend Myozyme for use within NHSScotland for the treatment of Pompe Disease. This decision is scandalous as it was based purely on cost. Infants die within the first year of their life and children and adults suffer severe muscle wastage resulting in a huge loss of mobility, breathing problems resulting in ventilator support and untimely death.

The number of Pompe patients in Scotland is few and to be disadvantaged by having a rare condition and living in Scotland is unforgivable.

Meeting the needs and expectations of all in Scotland is a huge challenge indeed however as you state '*We must do all we can to put the expectations of people and patients at the heart of the decision making in NHSScotland'.* Health and Wellbeing does matter to us all and Pompe patients and their families deserve a Scottish healthcare system they truly deserve.

Therefore having been given the opportunity to comment on the document Better Health, Better Care I would like to make the following points:

1. IMPOVING YOUR EXPERIENCE OF CARE

You state that in order to deliver your strategic vision you need to:
*'Improve our **patients' experience of care,** delivering care as locally as possible…'*

There are no specialist centres for the diagnosis, management or treatment of Lysosomal Storage Diseases (LSDs) in Scotland. England has 8 centres and 1 is currently being proposed for Wales. Currently patients' have to depend on physicians with differing levels of expertise at whatever hospital happens to be in travelling distance.

Looking ahead and with hope to the time the SMC recommend Myozyme where do the patients go to receive treatment? Delivering care as locally as possible would be impossible as no specialist centres exist in Scotland. However as the National Services Division (NSD) has responsibility for ensuring the provision of both national screening programmes and specialist services on behalf of NHSScotland patients would have to travel to England. NSD already manages, on behalf of the 15 Scottish NHS Boards, drug therapies for LSDs, such as Gaucher's Disease and Anderson- Fabry's Disease. The clinical decision on whether to approve treatment, (funding) remains the responsibility of the Director of Public Health of the NHS Board of residence therefore when SMC do recommend Myozyme for use within NHS Scotland there may still be inequality. NHSScotland should ensure this whole process including funding is equitable and this would improve Pompe patients' experience of care. Although ERT is provided for some LSDs in a Glasgow PCT currently there are no specialists trained to give Myozyme.

On a personal note, my understanding of the SMC is that it is a central point of advice to NHS Boards and their Area Drug and Therapeutics Committees (ADTCs) about the effectiveness of all newly licensed medicines. SMC advice will also help the NHS plan the speedy introduction of beneficial treatments across Scotland. When treatment is available in England for a Scottish patient, where this therapy has been approved, all decisions concerning this therapy have been vetoed. It seems until SMC decide to recommend Myozyme for use within NHS Scotland all other decisions concerning this therapy are put on hold even although as previously said for use within NHSScotland in this case would be impossible.

- Patients should be fully informed of what their care will be
- Professionals should treat patients with dignity and honesty
- There needs to be more transparency, openness and cooperation between agencies that need to work together.

5. ANTICIPATORY CARE AND LONG TERM CONDITIONS

*'Work in partnership to provide **anticipatory care** and improve services for **long term conditions'***

This is crucial for Pompe patients as early diagnosis and treatment would prevent long term conditions developing. NHSScotland should ensure they have specialists in LSDs to ensure Pompe disease is diagnosed as early as possible to avoid other complications developing due to misdiagnosis.

More information needs to be made available and accessible to practitioners as not all fully understand the long term conditions they are treating.

On a personal note, when a patient is suffering a chronic long term condition and has two very different consultants in two hospitals this can be difficult and stressful. Hospital 1 is a round trip of 90 miles, hospital 2 a round trip of 56 miles. The treatment in the 2 hospitals differs also, one being patient centred with good teamwork the other lack of cooperation or support and consultant

centred. Improving the management of this long term condition would be beneficial as when the patient knows more about his/her condition than the consultant it is worrying.

- Patients should be partners
- All professionals should treat patients with respect
- Everyone involved with care communicates

6. THE BEST POSSIBLE START

*'Invest in early intervention and prevention to **give children the best possible start** in life'*

All children deserve the best start in life; Pompe children should be no different. Myozyme gives infants the chance to thrive, without it they die. Without Myozyme older children suffer from severe disability. By having a specialist centre for LSDs in Scotland the children affected would receive the best possible start.

- Readily available information for the families of sick children
- All agencies involved need to work together
- Families need to be supported and fully informed

7. CONTINUOUS IMPROVEMENT IN HEALTHCARE

*'Ensure **continuous improvement** in services, with a determined focus on patient safety'*

People with orphan diseases suffer not only from having a horrible disease but also because it is so rare they find themselves in a minority with few specialised centres or those with the expertise to treat them. By having specialised diagnostic and treatment centres with ongoing and up to date treatments for LSDs and other rare conditions this would be continuous improvement NHSScotland could be truly proud of.

- Deliver the services Scotland needs and deserves
- Improved services for chronic and acute conditions
- Patients and carers viewed as partners

IN CONCLUSION

It seems that those with rare conditions, who really need specialised care, support and an inclusive approach to their health care, are very often left struggling to cope because of the lack of cooperation and collaboration between the NHS and its partners. NHSScotland has to ensure the standard of care is equitable as any variation in clinical practice is unacceptable.

At this moment in time to live with a rare muscle wasting condition like Pompe Disease and know that there is treatment available but, because you happen to live in Scotland, are unable to access it is cruel. By living on the other side

of border with treatment infants have the chance of life; children and adults have hope and a greatly improved life. By living on this side of the border without treatment infants will die and children and adults will lead shortened lives with ever increasing disabilities and reliance on others, including NHSScotland. That is heartbreaking.

I urge you to look on the above points favourably.

Yours sincerely

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NHS Foundation Trust

University Teaching Hospital

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Clinical Science building, Room A209, 1st Floor, A Block

safe • clean • personal

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27th August 2010

Allan Muir

Dear Allan,

I would like to bring to your attention the situation regarding the prescribing of Myozyme for patients living in Scotland and at the same time see whether you cannot help alleviate the problem.

A little while ago the Scottish Medicines Committee reviewed Myozyme and decided that it would not approve its use. This was well before any of the trials published on its efficacy in adults.

Subsequently, we have seen a number of patients from Scotland and the situation currently is, in my opinion, both against natural justice and not in the patient's best interests. I will, in this letter, not mention the patient's names but I would be happy to give you details following discussion with the patient to get their permission. However, I think the details of the case are quite important.

The first patient we saw from Scotland is a gentleman who has disease diagnosed according to the nationally agreed guidelines. He is wheelchair bound and has some night time ventilation. We applied for funding and received it. He has now been on funded treatment for over 2 years. He has shown some improvement in certain aspects of his condition and he is now quite stable showing no progressive deterioration. We feel there is some major improvement in this patient.

We were then referred a lady with enzymatically proven Acid Maltase Deficiency. She is moderately severely affected in terms of her muscle power but does not have any significant respiratory problems. We applied for funding and after an appeal, which I supported, funding has been turned down.

More recently, we have seen another lady with enzymatically proven Acid Maltase Deficiency. She too has mild to moderate muscle weakness with some minimal respiratory symptoms. She lives in a different area to the other lady. Nevertheless, we have been refused funding. An appeal is pending although I am not very confident of winning it.

It seems to me bizarre that we can have three patients with the same condition in different parts of Scotland where one gets treatment and the other two do not. It is also unclear as to what criteria's are being used.

There is nothing further that I can do at this stage although we will lodge an appeal around the decision on the last patient. However, I wonder whether the Pompe Association would like to take this up with the Scottish authorities. Far be it for me to suggest how this might be done, but it seems to me against the rules of natural justice to have one patient in Scotland on treatment and two patients's being denied it.

I would be more than happy to discuss this with you over the telephone. I am away at the SSIEM the first week in September but back thereafter and will be more than happy to explore this with you. As you can see, I have copied this to Joan Fletcher who is aware of our three patients.

Kind regards

Yours sincerely

Dr S Waldek
Consultant in Adult Inherited Metabolic Diseases

cc

Sister Joan Fletcher
Family Liaison Officer/Clinical Nurse Specialist
Genetic Medicine, 6th Floor
Manchester Academic Heath Science Centre
Central Manchester University Hospitals NHS Foundation Trust
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M13 9WL

Dear Ms. Mawson,

I have just received your e-mail following the panel meeting to discuss the application to prescribe enzyme replacement therapy for patient LM. I am sorry that I cannot send a formal letter but I am just about to leave for a lecture tour in Australia. I have answered the questions raised and I also attach some PowerPoint slides of results from published trials and natural history studies. I did provide the reference for the recently published outcome study with my original letter.

1. Acid Maltase deficiency is a RELENTLESSLY PROGRESSIVE muscle disease. I attach slides from two studies on the natural history of the disease. In the case of the patient in question, I have already detected deterioration in her condition over a relatively short time frame and I am therefore sure that without treatment she will slowly get worse with increasing muscle weakness and become progressively less mobile and able to conduct acts of daily living. This in turn will make it necessary for her to have help from carers. In addition, she is more than likely to get involvement of her muscles of respiration and eventually be in need of long term ventilatory support. I am afraid that IO cannot make it planer than this!
2. The question of efficacy was raised. What makes this patient different from others is that all the other patients we have under our care (36 at present) are able to access enzyme therapy. From a clinical point of view she is no different from the other patients. She has a proven diagnosis and progressive disease. I attach slides from the recent results of a trial published in the NEJM. From our own, now quite extensive, experience patients respond to enzyme by showing a degree of improvement in well being and muscle power (respiratory function as well if impaired). This occurs over two years and then patients stabilize. There is nothing to indicate that the patient in question here will not show some improvement and then stabilize. I have not seen any adults who have deteriorated despite treatment. Our aim, in the long term for this patient would be to ensure that she does NOT deteriorate and can maintain or improve on her current function.
3. She would have met the criteria for the trial
4. Response to treatment would be measured clinically as with all the other patients. Once on therapy the patient are seen on a 6 monthly basis. The minimal; assessment is as follows:-
    a. Full history with special reference to nutrition, respiratory function and mobility.
    b. Full examination with special reference to muscle power and function musing the MRC scale.
    c. 6 minute walk test where applicable.
    d. Pulmonary function tests. We are especially interested in the FEV1 and FVC lying and standing together with the SNIP—sniff Inspiratory pressures---as these give good indication of diaphragm involvement.

    e. Where indicated patients have nutritional blood tests, cardiac evaluation, and DEXA bone scans. These are all as per the NCG published guidelines.
5. I have been asked about outcomes of patients treated for more than 12 months. As already mentioned patients continue to improve till about 24 months after starting and then stabilize. A few patients continue to improve.
6. Reference has been made to other therapies. There are NO VALIDATED trials of alternative therapies. The following is a brief outline on what has been tried and our evidence based approach to these.
    a. Exercise: There have been anecdotal reports of the positive effect of various exercise programmes on the course of acid maltase deficiency. None of these have shown any long term gains. However, it is quite clear that well regulated aerobic exercise and attention to posture and gait do have a place in the overall management of patients. We are fortunate in having our own departmental physiotherapist and she assesses all patients giving them appropriate advice and working with local colleagues if needed. Our experience is that physiotherapy alone has no effect, and if done to intensively can be detrimental. However, a proper regulated exercise programme in combination with ERT does give better results than ERT alone in many patients.
    b. Diet: There have been several studies of various types of special diet—mostly based on protein and/or calorie supplements. None of these have been validated and have not stood the test of time in the pre-ERT era. However, with ERT diet has a part to play. Again we are fortunate in having our own metabolic dietitian. All patients have a dietary assessment at baseline and that includes blood tests. Many patients have significant vitamin D deficiency. Patients are encouraged to eat a healthy diet and supplements are given where necessary. (This patient has just been advised to take calcium and vit D tablets.) Patients on ERT eating a good healthy diet and exercising as well, will put on a small amount of weight. Much of this is accounted for by muscle bulk.
7. One last point to make, not covered previously, is that we tell the patients that we would treat them for one year and stop therapy if the disease has continued to progress despite enzyme replacement therapy.

I hope that this is satisfactory. As I mentioned at the beginning, I am due to travel overseas but will be able to pick up my e-mails all the time so will be able to answer further questions if needed.

Yours sincerely

Stephen Waldek
Consultant in Adult Inherited Metabolic Diseases

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NHS
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Date 30 November 2010
Your Ref
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Dear

Thank you for taking the time to attend the Medicine Appeals Panel (MAP) on Wednesday 3 November 2010, in Eglinton House, Ailsa Hospital, and for your patience while awaiting the date of the panel reconvening, on 26 November 2010.

As you are aware, the remit of the MAP is to provide an opportunity to appeal a decision regarding availability of a medicine for a particular person where it can be demonstrated that:

- Important facts may not have been taken into account when considering the request,
  and/or
- The officer of NHS Ayrshire & Arran making the decision, did not follow reasonable process of the principles of their remits.

Therefore, in line with the Medicine Appeals Procedure, I write to confirm the Panel's decision to uphold the IPTR (Individual Patient Treatment Request) Panel's decision to decline the request for Myozyme®, (algucodidase alfa), to treat late onset Pompe's disease.

The Appeals Panel were extremely sympathetic to your case, which made the decision a difficult one. However, after reviewing the additional article presented by Dr Deegan, the Panel concurred that it provided no further important information to uphold the appeal, or impact on the QALY cost, and subsequently on the opportunity cost implications for NHS Ayrshire & Arran.

The Appeals Panel also concurred that the process followed by the Executive Medical Director when considering the original request for Myozyme®, (algucodidase alfa), was both reasonable and thorough.

The Appeals Panel recommend that NHS Ayrshire & Arran should consider any future resubmission by your clinician, which provides objective clinical evidence of an increasing rate of deterioration of overall severity in your condition.

Should you remain dissatisfied with this decision, you have the right to seek legal advice as there is no further route for appeal on this occasion.

Yours sincerely

Dr Wai-yin Hatton
Chief Executive

Scots Who Dinna Hae:
Cauld Comfort for Pompe and the Campaign for Justice

With the situation in Scotland with regards to the funding of Myozyme reaching a critical point, campaign groups, individuals and the media have been increasingly involved in a bid to finally change national policy. By Luke Fraser and Allan Muir.

It is given to say that one of the cornerstones of the UK social democratic welfare state is the NHS. The establishment in 1948 of four national healthcare systems across the UK brought with it an era of universal healthcare which, despite innumerable on-going crises and threats of privatisation, has continued uninterrupted to this day. As a result the NHS as a whole has long been lauded internationally, rooted as it is in its fundamental principal of providing equality of access to free treatment for all.

'There are currently only 11 known patients with Pompe in Scotland. Yet whilst all have now been diagnosed, to date only three have each been prescribed treatment'

Yet some, it would seem, are more equal than others. In the case of ultra-orphan diseases such as Pompe, with significantly higher per-patient treatment costs - albeit offset by extremely low population numbers, the situation has not always been so clear, and there has often been great disparity across between the four UK NHS systems.

'The AGSD-UK has been working closely with the Muscular Dystrophy Campaign (MDC) since 2010 to apply pressure at a number of levels'

In England, Enzyme Replacement Therapy (ERT) for Pompe has been fully funded since market approval in 2006. With NHS Wales, which had dragged its heels with regard to funding, success was eventually achieved in 2008 with the establishment of an LSD centre at the University Hospital of Wales in Cardiff, guaranteeing treatment for infants, children and adults with a history of Pompe since childhood.

Yet the situation in Scotland has continued to lag behind. There are currently only 11 known patients with Pompe in Scotland. Yet whilst all have now been diagnosed, to date only three have each been prescribed treatment following an Individual Patient Treatment Request (IPTR). These include one adult and the two infants - Sean Tye (3) and Sabeel Hussain (8), who we have mentioned in previous issues.

As the Bulletin reported back in 2010, a cost-sharing agreement was set up between the 14 Scottish local health boards. This arrangement was supposed to have covered infantile and late-onset Pompe patients, yet so far neither group has benefitted. As the AGSD-UK has pointed out, the agreement simply isn't working in the manner in which it was set up to do.

As time goes on and as the symptoms of these untreated patients progress, the need for treatment becomes ever more pressing and its withholding by the health boards more unacceptable. It is also inexplicable in view of the fact that equally costly drugs for other LSD orphan diseases such as Gaucher and Fabry are treated in Scotland whilst access to ERT for Pompe is denied.

Against this background the AGSD-UK has been working closely with the Muscular Dystrophy Campaign (MDC) since 2010 to apply pressure at a number of levels, both locally and nationally, with the objective of showing the gross unfairness of access to treatment in a number of ways and ensuring political change.

A key aim of the campaign has been to lay out the importance of early treatment and why a protracted and ill-defined process to it is so dangerous. It is commonplace by now that once a diagnosis for Pompe is confirmed patients need to start therapy without delay, and that infants are permanently disabled by delays in treatment.

With such objectives in mind, the campaign took various guises. The MDC has made representations to the Scottish Muscle Network of physicians who met with Minister for Public Health and Sport, Shona Robison, with a view to influencing her review of health services. Earlier, AGSD-UK Development Director Allan Muir successfully pushed for the insertion of a sentence on Pompe into the recent Cross-Party Group of the Scottish Parliament's Mackie Report. The recommendation was made that 'the Scottish Government reviews the situation regarding the unequal treatment of the small number of patients with Pompe disease living in Scotland.'

The AGSD-UK was also represented by Allan Muir, Joan Fletcher, Lynn Millar and Tom Addyman – the father of two Pompe boys, in the Rare Disease Day meeting at the Scottish Parliament in February. This was chaired by Jackie Baillie, Labour MSP for Dumbarton and Shadow Cabinet Secretary for Health and Wellbeing. Jackie is also Chair of the cross-party-group on Muscular Dystrophy. After the meeting she tabled a written parliamentary question on access to treatment for Pompe. There is a mounting legal angle to all this. The AGSD-UK is of the opinion that the only way to succeed currently is through Judicial Review of hospital board decisions. Meanwhile patients are being encouraged to investigate taking legal action against local health boards. The AGSD-UK has stated that it will fully support any individuals or groups who wish to do so.

In terms of the media, a joint press release was put out with the aim of clearly highlighting the situation for Pompe patients and families. As a result coverage was favourable. Stories were featured in Scottish national press in print and online, and on National TV and radio. At a conservative estimate, coverage is highly likely to have been seen, heard or read by over 300,000 Scots.

On TV, BBC Scotland carried the story in their lunchtime bulletin. The report included an interview with Sheena Burton, a mother of three adult children with Pompe. Sheena discussed her fears for her children and the difference Myozyme could make to their symptoms.

On STV North there was a full report of two and a half minutes featuring Paul Jupp – one of Sheena Burton's sons, and an interview with Sheena explaining that if two of her children had access to treatment now, they would 'never have to suffer'. In the interview Sheena talked of the terrible consequences of the conditions, with permanent ventilation being a potential outcome for all of her children.

An interview with Nic Bungay, Director of Care Support and Campaigns at the MDC then followed, in which he called on the Scottish Medicines Consortium to review access to the drug.

'At a conservative estimate, coverage is highly likely to have been seen, heard or read by over 300,000 Scots'

On radio, BBC Radio Scotland featured a minute long story also featuring an interview with Nic Bungay. The key message in the piece was of the discrepancy between the drug availability in Scotland and the rest of the UK. Nic argued that with so few patients needing the drug, it is affordable. A slot was also devoted to the issue on BBC Radio Aberdeen, giving focus to Sheena Burton's family as local patients affected by the issue.

Meanwhile The Herald newspaper ran two stories in March. The first focused on 3 year old Sean Tye, who in 2008 was the first person in Scotland to receive treatment, only after the personal intervention of First Minister Alex Salmond. The article quoted Sean's father Ching Shen (Sam) Tye as saying 'We are immensely proud of our son and are so grateful to have received treatment that means he is here with his family today.'

The MDC's Nic Bungay

Sam continued: 'However, for parents here in Scotland who have a child with Pompe disease, medication is hard-won. The sooner that treatment begins for a baby with Pompe, the better their chance of a childhood. I dread to think of other families watching time pass by as they wait to hear whether their child will get the medication that they need.'

Allan Muir was also interviewed. He said 'Access to therapy for Pompe disease patients in Scotland currently depends on which health board a patient belongs to, and the vast majority of patients are being denied treatment. This is hugely frustrating for the families affected, especially given that patients with other related rare diseases are being treated with similar medications

Nic Bungay also commented, saying 'we know of only 11 Scottish Pompe disease patients, yet the vast majority of them are being denied access to a ground-breaking treatment automatically available to people living in England, on the grounds that the therapy is not cost-effective. We are backing the Scottish Pompe disease patients fully in their call for an urgent review into the treatment for this painful disease.'
The article concluded by quoting a spokesman for the SMC as saying: 'We evaluated Myozyme in 2007 and were disappointed to find that it wasn't a value for money medicine. This decision was based on analysis of the manufacturer's own information. All NHS boards have mechanisms in place in order to consider drugs that are not deemed to be cost-effective. The manufacturer is at liberty to make a submission with new or revised data at any time, and we would welcome such a submission.'

'Nic Bungay argued that with so few patients needing the drug, it is affordable'

The Herald also ran a story on Paul Jupp, mentioned above. It described how when Paul was growing up he would tire easily, that he struggled to walk long distances and that his legs would sometimes collapse underneath him. His mother Sheena said 'I knew there was something wrong, but it was years before they picked up what it was.' Paul was 14 when he was diagnosed. The impact of the condition on the muscles in his face has made it difficult for him to talk and he often stops breathing in his sleep. Sheena Burton says her biggest fear is that one day he will not wake up. She continued: 'I find it so frustrating that there is no support for them here in Scotland. Knowing that there is a drug that can halt this disease and that it is not available to patients here, I find unbearable.'

The Daily Record newspaper ran a feature on Lynn Millar, 42. It stated that if Lynn does not receive 'the drug which eases the incurable muscle-wasting condition, she will be confined to a wheelchair and need round-the-clock oxygen.'

It described the determination of Lynn, who will not let her illness hold her back in her career – continuing as she does to work full-time as a supply chain team leader with Spirit Aerosystems, a job she's been in for 26 years. When not at work Lynn campaigns for Myozyme to be available to all Pompe sufferers in Scotland.

Lynn, who was diagnosed with Pompe after developing a limp, said 'I walk with a limp because my legs have a weakness. But Pompe can affect every muscle in your body and it will get worse. I struggle with walking up hills or stairs, getting out of a chair. But I try to get on with life because I have a daughter. Lauren is great. She'll dust, vacuum and shop without moaning.'

Lynn was diagnosed in 1999, the same year as Lauren was born. She worries every day about being wheelchair-bound and the impact it will have on Lauren. 'I don't want to hold Lauren back', she said. 'I know it will happen if I don't get the drug. That's why I am going to do everything I possibly can to get it.'

Last year, Lynn met medical specialists from NHS Ayrshire and Arran where her case for Myozyme was heard. It was thrown out and Lynn appealed. This was also rejected. In a bid to have it overturned, and

have the drug more readily available to Pompe sufferers in Scotland, Lynn has now contacted her local MSP John Scott.

Lauren, 11, has been forced to grow up fast and throughout her childhood has been responsible for many household chores; but she does not mind. She said 'My mum is such a loving and kind person. If she got the drug, life would be very different and it would prevent her from being in a wheelchair.'

"For parents here in Scotland who have a child with Pompe disease, medication is hard-won"

Lynn's parents find it tough to watch their daughter struggle. Mum Margaret, 64, said: 'She deals with it as well as she can. She's not in pain but it breaks our hearts to watch her suffer. Lynn lives for Lauren. She wants a quality of life for her, more than herself, but that won't happen if she doesn't get the drug.'

These stories all too clearly illustrate the plight of untreated Pompe sufferers in Scotland. It is strange at best that a health authority can put free hospital parking and prescriptions above the needs of its patients requiring treatment. The European Orphan Drug legislation was put in place to find a solution to the inequalities faced by those living with ultra-rare diseases. But Scotland, it seems, does not value its less fortunate citizens as highly as do all of its European neighbours. At least Scots won't clock up huge parking fees as they wait in clinic for treatment, and they can always stockpile prescription analgesics to ease their muscle pain.

The AGSD-UK is currently considering other measures that may be successful in raising the profile and needs of Pompe families in Scotland.

"Knowing that there is a drug that can halt this disease and that it is not available to patients here, I find unbearable"

The organisation will continue to lobby MSPs, Health Ministers and Health Boards in collaboration with other organisations such as the MDC, the Scottish Patient's Association, Genetic Alliance, and the UK LSD collaboration. The fight for equitable treatment across the UK must be sustained.

A full version of the Mackie Report can be viewed online at:
www.muscular-dystrophy.org/assets/0001/9335/Mackie_Report.pdf

The reception at Rare Disease Day

Rare Disease Day 2010

PE1401

Petition by Lesley Loeliger, on behalf of PNH Scotland, and Professor Peter Hillmen, on behalf of the PNH Alliance, calling on the Scottish Parliament to urge the Scottish Government to review the mechanism and methodology used by the Scottish Medicines Consortium to appraise medicines for rare diseases and to instruct the Chief Medical Officer to revise the criteria by which health boards assess Individual Patient Treatment Requests in order to improve access to therapy for patients with paroxysmal nocturnal haemoglobinuria.

PNH Alliance and PNH Scotland: supporting evidence for Public Petition (PE1401)

1. Introduction

The PNH Alliance and PNH Scotland are grateful for the work of the Health and Sport Committee in taking further inquiries to ascertain the reason for the lack of access for PNH patients to Eculizumab.

The Scottish Medicines Consortium (SMC) considered Eculizumab for the treatment of PNH in 2010 and despite proving demonstrable efficacy[i], the SMC decided not to recommend it. While it is nationally commissioned for patients in England and Wales, and fully funded in Northern Ireland, patients in Scotland can therefore only access the medicine via Individual Patient Treatment Requests (IPTRs) made to local NHS Boards by their consultant haematologist.

However, there presently exists a very serious situation in Scotland where the designated outreach clinic for all Scottish PNH patients is held in Monklands Hospital in Lanarkshire but the individual health boards of respective patients determine funding (or not as the case may be) for those that require Eculizumab.

Globally recongised expert PNH physicians working at the Monklands Hospital can therefore only recommend Eculizumab for treatment with patient not guaranteed to receive it despite clinical opinion to the contrary.

The situation is particularly acute for patients within the NHS Greater Glasgow & Clyde Board where several patients have been denied treatment. This has led to the death of one patient in May 2011 and is putting further lives at risk.

2. Public Petition

In order to address patients' concerns in regard to access to therapy for PNH, the PNH Alliance and PNH Scotland submitted a public petition in August last year, asking the Scottish Parliament to urge the Scottish Government to review the mechanism and methodology used by the SMC to appraise medicines for rare diseases and to review the decision-making criteria used for the IPTR process.

The Public Petition Committee considered the petition and gathered evidence from the Scottish Government and relevant stakeholders involved. An evidence session was held by the Committee on the 4th October 2011, at which PNH Alliance and PNH Scotland provided oral evidence to the Committee.

3. Issues raised within the inquiry by the Public Petitions Committee

(i) Scottish Government

In response to the inquiry by the Public Petitions Committee, the Scottish Government stated that the "SMC operates independently from the Scottish Government". Whilst the PNH Alliance and PNH Scotland understand that there has to be some degree of independence, the fact remains that the SMC is responsible for recommendations on considerable amounts of government spending for which elected politicians have to take responsibility. The PNH Alliance and PNH Scotland believe that it is the responsibility of the Scottish Government to set the criteria and framework within which the SMC operates in order to improve access.

In March 2011 the Scottish Government issued IPTR guidance for NHS Boards, which is however not being implemented consistently as demonstrated by the inequity of access to Eculizumab. The update guidance to further strengthen the safe and effective use of new medicines across the NHS in Scotland, which was published in February this year, further fails to ensure that funding requests by PNH patients are comprehensively assessed by the NHS Boards' IPTR panels (see 3.iii).

Overall, the SMC and the NHS Boards claim to act in accordance with government guidelines, while the Scottish Government states that the funding decisions are taken by independent bodies and it would therefore be inappropriate for Ministers to intervene in the decision-making process. There is therefore a lack of accountability and a need for the Scottish Government to comprehensively review the appraisal process for medicines for rare diseases such as PNH.

(ii) Scottish Medicines Consortium

PNH is an ultra-orphan condition and we recommend that the SMC formally recognises such conditions in line with definitions stipulated in England by NICE and in Wales by the All Wales Medicines Strategy Group. England and Wales have further recognised the need for separate policy mechanisms to review ultra-orphan therapies.

The SMC does not provide data on recommendations on ultra-orphan medicines. However, analysis[ii] by the PNH Alliance and PNH Scotland shows that of the 12 medicines licensed by the European Medicines Agency for ultra-orphan conditions, the SMC only recommends one for restricted use only.

The PNH Alliance and PNH Scotland believe that the SMC system of modifiers does not work because successive studies of Eculizumab demonstrate "evidence of substantial improvement in life expectancy and quality of life". [iii] However, the SMC still declined to recommend it for reimbursement by NHS Boards.

The very small patient populations suffering from ultra-orphan diseases make it extremely difficult for manufacturers and clinicians to generate robust clinical cost effectiveness data required by the SMC HTA process. The very concept of health economic analysis was originally developed to facilitate comparison of products for large patient populations and for which there are alternative treatment options. Conversely, given the small patient populations and general lack of treatment alternatives, it is inappropriate to apply conventional cost effectiveness analyses and thresholds to ultra-orphan therapies. Doing so serves only to entrench the systemic denial of patient access to ultra-orphan medicines as it is extremely unlikely that the HTA Quality Adjusted Life Years (QALY) mechanism (central consideration of the SMC) will ever come close to traditionally accepted levels when assessing ultra-orphan medicines.

The PNH Alliance and PNH Scotland would support work by the SMC to understand more regarding public views on health spending associated with rare diseases. Such discussions should be held with a high degree of public transparency.

(iii) Individual Patient Treatment Requests

In regard to the IPTR process there currently is a gross inequity for PNH patients, affecting those living within the NHS Greater Glasgow and Clyde (NHSGGC) area. In relation to a recent case of a PNH patient highlighted extensively in the media and Parliament, the NHSGGC released the following statement: "The patient's new consultant has completed a detailed clinical review and has presented the case to a multi-disciplinary team of 16 haematologists from across the west of Scotland for their expert opinion as to whether her condition supported the prescription of eculizumab. After assessment of her case the multi-disciplinary team did not support the view that her condition had changed significantly for the prescribing of eculizumab."

There were however no haematologists who are considered experts in PNH or who have published clinical papers on treatment of PNH in NHSGGC. Prof Peter Hillmen, Consultant Haematologist, St James University Hospital, Leeds and renowned expert in PNH has confirmed that another patient, whose appeal for funding of Eculizumab was rejected by NHSGGC last year, died shortly after with severe anaemia, which was caused by his PNH. Another PNH patient had an IPTR declined by NHS Greater Glasgow and Clyde and was unsuccessful in her appeal. It is our understanding that it was only when she suffered a potentially fatal blood clot that her IPTR was accepted. The PNH Alliance and PNH Scotland are therefore of the firm opinion that there is overwhelming clinical evidence for PNH patients to be treated with Eculizumab and considers the lack of treatment to be a financial decision as opposed to a clinical one.

The situation for PNH patients living within the NHSGGC catchment area is further exacerbated by some patients receiving Eculizumab and some not despite living in close proximity. The NHSGGC bases decisions on whether to fund therapy on exceptionality criteria. However, given the very small amount of patients in NHSGGC the catchment area (estimated to be 4) there is a strong argument to suggest that all PNH patients are exceptional and exceptionality cannot be defined for such low patient numbers. Furthermore, PNH is a life threatening disease and therefore exceptionality is based on the patient becoming gravely ill. The patient, who passed away earlier this year, was not considered an exceptional case but nevertheless died from PNH, largely as a result of failure to fund treatment by NHSGGC.

The Scottish Government published in February this year updated guidance to further strengthen the safe and effective use of new medicines across the NHS in Scotland, which provides that IPTR panels "are expected to include a practicing medical consultant with (or with access to) specialist knowledge of the relevant clinical area". However, the PNH Alliance and PNH Scotland believe that the updated guidance is not strong enough to ensure that PNH specialist clinical opinion is taken into the IPTR panel's consideration when assessing funding requests by PNH patients. In rare diseases such as PNH, there only exists a very small number of specialist clinicians, who have particular expertise and experience in treating patients with PNH, all of which have strongly recommended the treatment of with Eculizumab.

The PNH Alliance and PNH Scotland therefore believe that a general haematologist who only *has access to* clinical data on PNH will not have the necessary expertise to assess the impact of the treatment on patients' outcome. It is therefore vitally important that a specialist clinician, who is widely recognised as having particular expertise and clinical interest in PNH is included in the IPTR decision-making process to ensure that patients' funding requests are assessed by an appropriate medical expert.

4. Recommendations for future development

It is noted from the responses that all of the organisations that the Committee contacted claimed to be correctly following agreed Scottish Government policy. Whilst this may be correct, PNH patients are still being denied access to life saving therapy, which suggests that there is a systematic failure between NHS Boards, the SMC and the Scottish Government.

The PNH Alliance and PNH Scotland therefore propose that the Scottish Government establishes and implements a separate body or alternative pathway within the SMC for the appraisal of ultra-orphan medicines. Such a body or process would be in line with arrangements in England and ambitions set out by the Scottish Government for NHS Scotland in their Quality Strategy which states that: "The most appropriate treatments, interventions, support and services will be provided at the right time to everyone who will benefit." [iv]

The proposed body should undertake a holistic evaluation of the value of a ultra-orphan medicine to patients taking into consideration the following criteria:

- Data on clinical effectiveness showing improvements in quality and/or quantity of life and an assessment of patient outcomes on therapy
- The number of patients whose condition improves as a result of the treatment compared to the total number of patients treated (NNT)
- Burden of illness and severity of condition
- Availability of treatment alternatives
- Where possible, comparison with existing treatments and to what extent the medicines meets unmet need
- Safety and risk profile of the medicine
- Societal value of the medicine including impact on carers or families, needs and expectations of patients (including productivity quality of life), patient voice and NHS Scotland priorities
- Benefit to society from research and innovation in the relevant area where conventional rules of investment may not apply

5. Suggested future actions:

- **Request that the Scottish Government undertake a public consultation on new means for appraising ultra-orphan medicines in line with arrangements in England and Wales.**

- **Request that in the interim the Scottish Government designates funding for PNH patients to prevent putting further lives at risk.**

- **Further strengthen guidance for IPTR decision-making to ensure that patients' funding request are comprehensively assessed including the medical opinion of recognised clinical experts in the treatment of PNH.**

- **That NHS Greater Glasgow & Clyde make publicly available their "expert clinical opinion" on the use of Eculizumab and that this is compared with clinical evidence developed by recognised clinical leaders in the treatment of PNH.**

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[i] R. Kelly; A. Hill, L. Mitchell, P. Hillmen et.al.: *Long term treatment with eculizumab in paroxysmal nocturnal hemoglobinuria: sustained efficacy and improved survival*, American Society of Hematology, April 1, 2011; R. Brodsky, N.Young, et.al: *Multicenter phase 3 study of the complement inhibitor eculizumab for the treatment of patients with paroxysmal nocturnal hemoglobinuria;* American Society of Hematology ,February 12, 2008; P. Hillmen, C.Hall, et.al.:*Effect of Eculizumab on Hemolysis and Transfusion Requirements in Patients with Paroxysmal Nocturnal Hemoglobinuria,* The New England Journal of Medicine, 2004

[ii] EMA website; accessed November 2011

[iii] R. Kelly; A. Hill, L. Mitchell, P. Hillmen (ibid).

[iv] THE SCOTTISH GOVERNMENT, NHS Quality Strategy, May 2010: http://www.scotland.gov.uk/Topics/Health/NHS-Scotland/NHSQuality ; Accessed 8th November 2011