EUROPLAN
European Project for Rare Diseases National Plans Development
Coordinated by the Italian National Centre for Rare Diseases
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RARE DISEASE | UK

EURORDIS
Rare Diseases Europe

United Kingdom

EUROPLAN NATIONAL CONFERENCE

FINAL REPORT

24 June 2014, London

EUCERD
Joint Action

FOREWORD

The EUROPLAN National conferences are aimed at fostering the development of a comprehensive National Plan or Strategy for Rare Diseases addressing the unmet needs of patients living with a rare disease in Europe.

These national plans and strategies are intended to implement concrete national measures in key areas from research to codification of rare diseases, diagnosis, care and treatments as well as adapted social services for rare disease patients while integrating EU policies.

The EUROPLAN National conferences are jointly organised in each country by a National Alliance of rare disease patients' organisations and EURORDIS – the European Organisation for Rare Diseases. For this purpose, EURORDIS nominated 10 EURORDIS-EUROPLAN Advisors - all being from a National Alliance - specifically in charge of advising two to three National Alliances.

EUROPLAN National conferences share the same philosophy, objectives, format and content guidelines. They involve all stakeholders relevant for developing a plan/strategy for rare diseases. According to the national situation of each country and its most pressing needs, the content can be adjusted.

During the period 2008-2011, a first set of 15 EUROPLAN National Conferences were organised within the European project EUROPLAN. Following the success of these conferences, a second round of up to 24 EUROPLAN National Conferences is taking place in the broader context of the Joint Action of the European Committee of Experts on Rare Diseases (EUCERD) over the period March 2012 until August 2015.

The EUROPLAN National Conferences present the European rare disease policies as well as the EUCERD Recommendations adopted between 2010 and 2013. They are organised around common themes based on the Recommendation of the Council of the European Union on an action in the field of rare diseases:

1. Methodology and Governance of a National Plan;

2. Definition, codification and inventorying of RD; Information and Training;

3. Research on RD;

4. Care - Centres of Expertise / European Reference Networks/Cross Border Health Care;

5. Orphan Drugs;

6. Social Services for RD.

The themes "Patient Empowerment", "Gathering expertise at the European level" and "Sustainability" are transversal along the conference.

EUROPLAN

UK National Conference Report

www.raredisease.org.uk

General information

Country	United Kingdom
Date & place of the National Conference	24 June 2014, London
Website	www.raredisease.org.uk/
Organisers	Rare Disease UK
Members of the Steering Committee	Dr Peter Corry - Paediatrician, Bradford Dr Stephen Jolles - Clinical Immunologist, University of Wales Hospital Cardiff Richard Lynn - Scientific Coordinator, British Paediatric Surveillance Unit Marie McGill - National Lead, Single Gene Complex Need Project (SGCN) Barbara McLaughlan - Head of External Affairs, Novartis Pharmaceuticals Dr Liz Philpots - Head of Research, Association of Medical Research Charities Patti Simonson - Head of Social Work and Welfare Benefits Administration, Royal Hospital for Neuro-disability Nick Sireau Chairman and CEO, AKU Society Val Stevenson - External Affairs, Genzyme Therapeutics Dr Fiona Stewart - Consultant in Medical Genetics, Belfast City Hospital Alison Wilson - All Ireland Advocacy Support Officer, Society for Mucopolysaccharide Diseases Mark Barrett - Freelance consultant Jamie Holyer - Managing Director, Advocate Policy and Public Affairs Consulting Ltd Becky Purvis - Head of Public Affairs, The Royal Society
Names and list of Workshops	Workshop 1: Methodology, Governance and Monitory of the UK Strategy for RD Workshop 2 : Research Workshop 3: Centres of Expertise
Workshop Chairs (and Rapporteurs, where applicable)	Chair: Alastair Kent OBE Chair Workshop 1 Nick Sireau

	Chair Workshop 2 Chair Katie Bushby
Annexes	1. Programme in English 2. List of participants

Introduction

Almost 100 attendees from across the rare disease community gathered in London on 24 June 2014 for the EUROPLAN National Conference in the United Kingdom (UK). The day was focused on discussion around the UK Strategy for Rare Diseases and the respective plans being developed in each home nation.

The UK is in a unique position as the union is made up of four countries; England is governed wholly by the UK Government whereas Wales, Scotland and Northern Ireland are also governed by a devolved Parliament or Assembly. Each country is responsible for the majority of health matters in their nation, including provisions for rare diseases.

As a result of the unique makeup of the UK and the publication of the UK Strategy for Rare Diseases in November 2013, Rare Disease UK (RDUK) chose not to address all EUROPLAN themes at the National conference. Priority was instead given to the themes most relevant to the national context and situation:

- Methodology, Governance and Monitory of the UK Strategy for RD;
- Research;
- Centres of Expertise

This report provides a summary of the conference, draws attention to the issues identified on the day and outlines recommendations which have been formulated to address these outcomes, with the guidance of the RDUK steering committee.

Background

In 2012, in response to the 2009 EU Council Recommendation on an action in the field of rare diseases, all four UK governments came together to publish the **'Consultation on the United Kingdom Plan for Rare Diseases'**.

In February 2013, at the annual RDUK parliamentary reception to mark Rare Disease Day in England, Earl Howe, Minister in charge of rare diseases, announced that the Department of Health would establish a **Stakeholder Forum** to steer the UK Strategy for Rare Diseases. Director of Genetic Alliance UK and Chair of RDUK, Alastair Kent OBE, was selected to chair the group.

Rare Disease UK, working with The Genomics and Rare Diseases Team at the Department of Health in England, spearheaded the development of the UK Strategy for Rare Diseases. In November 2013, the **UK Strategy for Rare Diseases, signed by the health ministers of all four countries** of the UK was published. The document is a comprehensive Strategy that contains **51 wide ranging commitments**, which seek to empower patients with rare diseases and address issues around diagnosis, information, healthcare, genomics, registries and research.

The four home nations are now developing implementation plans which are at varying stages of development. The implementation plans will be specific to individual countries' needs and will take account of the structure, resource and capabilities of national systems.

Methodology, Governance and Monitoring of the UK Strategy for Rare Diseases

Recommendations:

1. **A single, UK-wide approach to implementing the recommendations of the UK Strategy for Rare Diseases should be the default solution for all four home nations**
2. **Relevant stakeholders should be informed and actively involved in the delivery of the 51 commitments**
3. **Concrete outcome measures need to be in place to allow for effective monitoring of the implementation of the 51 commitments in each nation**

1 A single, UK-wide approach to implementing the recommendations of the UK Strategy for Rare Diseases should be the default solution for all four home nations

Action by: Government Health Departments

There was widespread agreement from attendees that efforts should be focused on ensuring **UK-wide implementation** of the recommendations of the UK Strategy for Rare Diseases, where appropriate.

Attendees expressed concerns that separate plans for each devolved nation could lead to duplicated effort and result in a waste of resources. In order to minimise these pitfalls and ensure equitable access to services, attendees felt **that all four health departments should work together** to implement as many of the 51 commitments in the UK Strategy as possible.

Delegates identified a number of key areas for interaction and joint working by the four health departments. These include:

Registries: A number of registries exist in Scotland but attendees felt that they should be **linked with the rest of the UK**. Participants welcomed developments by Public Health England to create a **register for rare diseases** in England and noted the potential for this to be UK-wide initiative. Encouragingly, officials from the devolved nations demonstrated their willingness to be involved in this rare disease registration service.

Centres of Expertise: Attendees felt that there were a number of areas where Centres of Expertise for rare diseases could collaborate, these include: best practice guidelines, sharing knowledge and expertise, standardising electronic health records and utilising telemedicine. For more information see 'Centres of Expertise'.

Screening: In order to deliver the commitments related to identifying and preventing rare diseases attendees felt that a more joined up approach to screening for rare diseases across the UK would be valuable to families affected by rare diseases. Attendees welcomed the Welsh Government's intention to work with the other UK countries to see whether additional screening services can be introduced.

Research and training: Participants noted a number of areas for joint work, including the development of a central repository for all research studies and working together to inform and educate academia and health professionals on the benefits of research into rare diseases. Attendees also noted that collaborative research frameworks were a positive step forward. For more information **see 'Research into rare diseases'.**

Attendees from the devolved nations expressed their concerns regarding the **inequity of access to effective treatments across the UK.** For example patients in Wales noted that they were not always able to access medicines that are available in England. Patients felt that **effective implementation** of the UK Strategy for Rare Diseases in each nation would lead **to better access to treatment and care for patients** affected by rare conditions.

The **Commission Expert Group on Rare Diseases**, formerly the **European Union Committee of Experts on Rare Diseases (EUCERD) strives for a unified voice across Europe** for those affected by rare conditions. Delegates and country representatives agreed that the UK needs to work with Europe to tackle the issues associated with rare diseases.

Action by Government Health Departments: All four nations to identify areas for joint working within the UK with a view to linking into the work being undertaken at a European level. The UK Rare Disease Forum is ideally placed to oversee this process.

2 Relevant stakeholders should be informed and actively involved in the delivery of the 51 commitments

Action by: Government Health Departments and organisations leading on the delivery of commitments

Participants observed that all relevant stakeholders were not always engaged in the delivery of commitments. For example, some questioned the lack of involvement of the Medical Research Council despite the large number of commitments focusing on research. Others noted the poor engagement and involvement of the Royal Colleges - all have access to expertise and valuable information which would be useful in the delivery of the 51 commitments.

Attendees were keen to emphasise the importance of **updating and informing all stakeholders of progress** towards the implementation of the UK Strategy for Rare Diseases. This should be an **active process led by the Government Health Departments** in the each nation and delivered by bodies who are leading on the delivery of individual commitments.

It was felt that **active engagement with patients and patient representatives** was particularly important as patient groups work across borders to support patients but do not have the resources to actively campaign in each nation to ensure that the needs of the patients they represent are met.

Participants raised concerns over the lack of communication, engagement and transparency by NHS England in their **Baseline Analysis**, which is focused on identifying key issues related to highly specialised services and is part of a larger review of specialised services by NHS England. Of the 63 highly specialised services, only a dozen services are being looked at as part of the **Baseline Analysis**. Attendees were concerned that the list of highly specialised services which were being looked at was

not publicly available. In particular, those representing patients were concerned about the **limited patient involvement** in the Baseline Analysis and questioned whether the analysis was purely financially motivated, fearing that rare disease patients were particularly vulnerable at this stage.

Attendees agreed that Government Health Departments in each nation should ensure that all stakeholders are **updated and informed of progress towards implementation** of the UK Strategy for Rare Diseases.

3 Concrete outcome measures need to be in place to allow for effective monitoring of the implementation of the 51 commitments in each nation

Action by: Government Health Departments

There was **widespread agreement** from all attendees regarding **the necessity for outcome measures to be developed** and put in place to allow for the **effective monitoring** of the implementation of each commitment. Regarding Scotland and Wales, attendees welcomed progress towards a final country specific implementation plan but noted that the plans needed detail around delivery and must be supported with **timelines and key outcome measures**.

Attendees welcomed the progress being made in Wales and the message that the Welsh Government is committed to working with the other countries of the UK, and linking into work being undertaken at a European level, to agree a range of specific indicators to judge success, including outcomes. They aim to have the first set in place by March 2015.

Participants were keen to stress the importance of engaging with patients to ensure accountability and were pleased to hear of the **UK Rare Disease Forum**'s new remit to monitor implementation. The **UK Rare Disease Forum,** which was originally established to develop the UK Strategy for Rare Diseases, is now **responsible for ensuring each country implements all commitments**. Every two years each nation will have to report on their progress. The UK Rare Disease Forum is chaired by Alastair Kent OBE, Director of Genetic Alliance UK and Chair of RDUK.

Delegates expressed concerns at the reduction of patient representatives sitting on the Forum from the original group.

Action by Rare Disease UK: RDUK has already started to address this issue by bringing together almost 30 patient organisations from across the UK to form the Patient Empowerment Group, with the aim of helping to increase patient participation in the discussions around the implementation of the UK Strategy for Rare Diseases.

Research into Rare Diseases

4. **Collaborative research frameworks should be flexible and support contributions from centres outside the core partnership**
5. **Organisations involved in the appraisal of medicines should work with patient organisations and other stakeholders to review standards for evidence submissions**
6. **Academia and health professionals must be informed and educated on the benefits of research into rare diseases**
7. **All bodies involved in research into rare diseases should recognise that patient organisations are a vital resource to inform and improve approaches to rare disease research**
8. **A central repository with information of all research studies should be developed by Government Health Departments**

4 Collaborative research frameworks should be flexible and support contributions from centres outside the core partnership

Action by: Health and Social Care Research and Development, National Institute for Health Research, National Institute for Social Care and Health Research, NHS Research Scotland and Chief Scientist Office

Delegates felt that collaborative frameworks for research are a positive step forward in increasing research into rare diseases, but should be flexible enough to allow for, and encourage, contributions from additional centres.

Attendees were positive about the National Institute for Health Research's (NIHR) **Rare Diseases Translational Research Collaboration** (TRC), which will provide the English NHS with research infrastructure to support fundamental discoveries and translational research on rare diseases in England. The TRC brings together **Biomedical Research Centres** (BRCs), **Biomedical Research Units** (BRUs) and **Clinical Research Facilities** (CRFs) with research expertise into rare diseases. However, delegates noted that BRCs were not originally developed with rare diseases in mind and that a reliance on BRCs risks omitting centres which could also contribute expertise. Furthermore, attendees were concerned that the devolved nations would be excluded from such a partnership.

The UK Strategy for Rare Diseases notes that the basic and translational research needed to achieve faster access to evidence-based care will require '**international collaboration, particularly for extremely rare diseases**'. In light of this, it is apparent that research frameworks should encourage contributions from additional centres within the UK but also further afield.

5 Organisations involved in the appraisal of medicines should work with patient organisations and other stakeholders to review standards for evidence submissions

Action by: All Wales Medicines Strategy Group (AWMSG), National Institute for Health Care and Expertise (NICE) and Scottish Medicines Consortium (SMC)

Industry representatives noted that pharmaceutical companies are keen to address unmet need and are actively looking to develop medicines and therapies for small patient populations. They noted, however, that NICE's evaluation framework for medicines posed a significant challenge to the process.

The UK Strategy for Rare Diseases states that each nation should:

'Ensure that there are appropriate procedures for evaluating the costs and benefits of treatments for patients.'

Organisations involved in the appraisal of medicines, in particular NICE, should work with patient organisations and other stakeholders to review the necessary standards for evidence submissions. NICE's upcoming review of Highly Specialised Technologies (HST) is an ideal time to consult with stakeholders.

The views of participants at the EUROPLAN National Conference is supported by Genetic Alliance UK's 2014 **Patient Charter,** which examined NICE's HST evaluation framework and recommended a joined-up approach to research and development, regulation and evaluation of new treatments, ensuring that the data required and the data collected are aligned. The Charter made the following recommendations:

'[Medicine appraisal bodies] should work with medicine regulators in a coordinated way to provide advice to companies regarding clinical trial design and data collection; and guidelines for the manufacturer's evidence submission.

To facilitate equitable access to new treatments, research recommendations should not be subject to restrictive eligibility criteria.'

NICE should ensure that it always takes into account the limitations of data derived from studies on small numbers of patients when appraising all medicines for rare conditions, as medicines and therapies for rare conditions continue to be appraised via single technology appraisals and multiple technology appraisals.

6 **Academia and health professionals must be informed and educated on the benefits of research into rare diseases**

Action by: Health Education England, NHS Education Scotland, NHS Wales, Northern Ireland Department of Health, Social Services and Public Safety and the Royal Colleges

Participants voiced their concerns that academia and health professionals were not always informed and educated about the benefits of research into rare diseases. For example, attendees voiced concerns about health professionals and academics who have stated that funds for rare disease research would be better spent on mainstream NHS services.

Participants welcomed initiatives to increase the amount and quality of research into rare diseases. The Welsh Draft Implementation Plan states that the National Institute for Social Care and Health Research will be responsible for creating a culture supportive of research and development in health and social care organisations, increasing capacity to support high quality commercial and non-commercial health research and facilitating strong partnerships with key individuals and organisations in the UK and internationally. The Plan has initiatives to help all health professionals '**to be aware of the importance of access to research for those with rare diseases and where information on research opportunities can be accessed**', the Plan also states that the Welsh Health Specialised Services Committee (WHSSC) as part of its commissioning cycle (the process of planning, agreeing and monitoring services) will aim to review specialist services providers and ensure that they '**support research activity**'. The NIHR's Rare Diseases TRC in England has already been discussed above.

7 **All bodies involved in research into rare diseases should recognise that patient organisations are a vital resource to inform and improve approaches to rare disease research**

Action by: all bodies involved in research into rare diseases

The value that patient organisations bring into rare disease research was universally accepted by all participants. Speakers noted that it would be difficult to carry out research without the **expertise of charities and patient organisations** that are up to speed in their specific disease area and can make patients aware of research studies.

Patient organisations can help to increase the quality and also the success of research and should be consulted and also supported in contributing to studies. Speakers at the EUROPLAN National Conference noted that patient organisations should, where possible, keep good records to help identify potential candidates for research studies.

Attendees noted that patient groups do not always exist for rare conditions and a handful of patients may be all that is necessary for meaningful patient involvement. In discussions, there was agreement on the benefit of collaboration between groups supporting different conditions with linked indications such as similar symptoms. In the UK some patient organisations are already working together in this way and combining funding for research.

8 **A central repository with information of all research studies should be developed by Health Departments**

Action by: Government Health Departments

Speakers and delegates both agreed that there should be **a central repository for all research studies in the UK**. This should be given the cooperation and investment needed by all Government Health Departments.

It was noted that a central, UK-wide, online repository would bring a number of benefits including **bringing to light the results of lesser-known studies** e.g. those carried out by medical students and helping patients identify ongoing studies in order to facilitate participation, particularly when no patient group exists to carry out this function. The repository was identified as a key area where all four nations could pool resources and work together to deliver advancements in research.

Centres of Expertise

9. **Experts working in Centres of Expertise should develop UK-wide best practice guidelines**
10. **Centres of Expertise must work across borders to ensure the transfer of knowledge and expertise**
11. **National Health Services should invest in standardised electronic health records**
12. **Telemedicine should be explored as a potential means of enhancing healthcare for patients and families affected by rare conditions**
13. **Stakeholders should not overlook excellent services that do not have the 'Centre of Expertise' label**

9 **Experts working in Centres of Expertise should develop UK-wide best practice guidelines**

Action by: Centres of Expertise

Participants felt that the development of UK best-practice guidelines by Centres of Expertise would be a valuable resource and that these **guidelines should be appropriate for all four home nations**, within a framework and regulated.

Experts working as part of a Centre of Expertise were identified as those best placed to develop and monitor such guidelines.

In Northern Ireland, Centres of Expertise and **specialist clinics have an important role to play in producing guidelines** and disseminating information. For example, the Fabry clinic has produced high quality guidelines that can be used to keep doctors involved and informed regarding a patient's care.

Both speakers and participants agreed that it is important for patients and clinicians to be are **aware of the guidance** that is in place and know how best to access it.

Attendees stressed that **guidelines should not be duplicated** across the four home nations but that best practice should be shared. Centres in all nations should accept best practice guidelines that are generated by one centre, if it is done so under an **approved accredited process.**

The UK Strategy for Rare Diseases and the **European Union Committee of Experts on Rare Diseases (EUCERD)** has laid down what it deems to be the minimum standards that Centres of Expertise should achieve for patient care - these act as an important baseline for the development of a guideline.

Whilst all participants agreed that 'no patient should be left out', it was accepted that it would be too difficult to have Centres of Expertise for the rarest conditions. What was considered the minimum number of patients required to justify a Centre was, however not agreed upon by attendees.

10 **Centres of Expertise must work across borders to ensure the transfer of knowledge and expertise**

Action by: Centres of Expertise

In keeping with the running theme of the UK EUROPLAN National Conference, attendees felt that **Centres of Expertise must collaborate and communicate nationally and internationally.** This is supported by an earlier report on **Centres of Expertise** carried out by Rare Disease UK in 2013[1], which recommends that:

'It would be beneficial for medical professionals to liaise across the UK and globally, so they are informed of new treatments and upcoming clinical trials. In addition, **Centres of Expertise should share their knowledge and expertise** with local healthcare providers, which would raise awareness on a much broader scale'.

The report also found that there is room for improvement in this area:

'Although collaboration across the UK and internationally currently takes place for some conditions, this needs to become accepted good practice for all rare conditions. **Networking allows for research opportunities to be identified and developed**'.

Amongst many speakers and guests at the EUROPLAN National Conference, there was very little knowledge of **European Reference Networks** (ERNs). ERNs will, once the networks have been developed, provide the framework for healthcare pathways for rare disease patients through high-level integrated expertise. Recommendations for ERNs were unanimously adopted by EUCERD in January 2013.

[1] Centres of Excellence, 2013 Rare Disease UK
http://www.raredisease.org.uk/documents/Website%20Documents%20/centres-of-Expertise-10-a4.pdf

ERNs will:

- Apply EU criteria to tackle rare diseases requiring specialised care;
- Serve to connect research and knowledge centres treating patients from other EU countries;
- Ensure the availability of treatment facilities where necessary;
- Help professionals and centres of expertise in different countries to share knowledge'.[2]

11 National Health Services should invest in standardised electronic health records

Action by: National health services in each nation with the support of respective Health Departments

The UK Strategy for Rare Diseases states:

'The UK wants to support the sharing of information, data, knowledge and best practice in treatment nationally, across Europe and further afield'.

There was agreement from all stakeholders that **electronic health records hold great potential,** and developments in each nation have been promising. For example, the Queen Elizabeth Hospital in Birmingham is paperless.

Centres in Northern Ireland have recently introduced **electronic care records**; this has meant that clinicians can now view test results, imaging information, letters and other documents in moments. The new system has helped patients enormously. In Scotland a similar system exists, but attendees heard that small issues can create frustrating problems. For example, firewalls can block the movement of electronic information from primary to tertiary care, which can be particularly disruptive to care for rare conditions.

Participants recognised that data collection can be time consuming and difficult to make sustainable but felt it was important that **all four nations must work together to invest in a standardisation of electronic health records**.

[2] European Reference Networks, http://ec.europa.eu/health/rare_diseases/european_reference_networks/index_en.htm

12 Telemedicine should be explored as a potential means of enhancing healthcare for patients and families affected by rare conditions.

Action by: National health services in each nation with the support of respective Health Departments

Participants agreed that **health tools** such as mobile phone applications and telehealth were potentially important for better **meeting the needs of patients** and enabling more efficient use of a clinician's time.

In Wales, steps have been taken to develop a new application for use with smart phones that enables: **home monitoring**; patients to **update information at home**; and the patient's specialist to have access to the patient and their records. This has reduced the number of patients attending clinic without documentation and **improved patient compliance.**

In Northern Ireland **telehealth has allowed patients to contact specialists** by video link to discuss, for example, lab results. In England, clinicians are using Skype to reduce face-to-face contact where appropriate.

Participants felt that **telehealth for nations that have geographical challenges would be particularly beneficial** e.g. providing healthcare for rural populations in Scotland.

Concerns were raised regarding the commissioning structures (the process of planning, agreeing and monitoring services) of the National Health Services which are not suited to encouraging telehealth. Participants reported that telehealth activities are not always classed as a valuable use of clinicians' time. Attendees and speakers from specialist centres noted that there were lengthy waiting times to get approval for the use of technology in their practice and called for **greater flexibility and innovation**.

Although telehealth is a valuable tool, there was **consensus amongst all stakeholders that telehealth cannot entirely replace physical interaction with a clinician.**

13 Stakeholders should not overlook excellent services that do not have the 'Centre of Expertise' label

Action by: all stakeholders

Delegates felt that it was necessary for National Health Services across the country to determine what services are already available for patients affected by rare conditions. Attendees noted that in many cases **excellent services are being provided** but not under the formal title of a Centre of Expertise.

In some cases patients value access to a Centre of Expertise despite receiving excellent care at their local services. Attendees were keen to ensure that Centres that do not have a formal title are not undervalued.

More information about Centres of Expertise and the relationship with local services can be found in the **Rare Disease UK report 'Centres of Expertise'**.

Country Updates

Each country in the UK has published their initial response the UK Strategy outlining how they will meet the 51 commitments. In Scotland and Wales, country plans can be viewed here:

http://www.scotland.gov.uk/Topics/Health/Services/RareDiseases

http://wales.gov.uk/consultations/healthsocialcare/disease/?lang=en

In England and Northern Ireland, Statements of Intent can be viewed here:

http://www.england.nhs.uk/2014/02/26/stat-rare-diseases/

http://www.dhsspsni.gov.uk/statement-of-intent-rare-diseases

England

NHS England published its **Statement of Intent** on Rare Disease Day 2014. The Statement identified the commitments led by NHS England and identified the commitments where NHS England is the partner to other lead organisations.

The Statement of Intent was signed off by the **Rare Diseases Advisory Group (RDAG)**, which makes recommendations to NHS England and the devolved administrations of the NHS on developing and implementing the UK Strategy for Rare Diseases; the secretariat for the group is provided by NHS England. A sub-group of RDAG has also been formed to **identify solutions to improve early diagnosis in England**.

NHS England is currently in the process of developing its implementation plan which will be integrated within a wider **5 Year Strategy for Specialised Services**. At present the Specialised Services Team in NHS England is separating out which of the 51 commitments NHS England can work on directly and reaching out to partner organisations for the remainder; these include NIHR and the UK National Screening Committee. **NHS England is moving forward on the commitments related to research, data and information, and screening.**

NHS England is establishing priorities through the identification of issues within key services for rare conditions and treatments for small patient populations (which are not always for patients with rare diseases). This is being carried out in the context of a major financial overspend of approximately £900 million in the specialised commissioning budget. A dozen services, which will act as a proxy for all specialised services, will be looked at as part of a Baseline Analysis to see how much money is being spent on services; how many patients are affected; and identifying where there are shortcomings in the service.

The next steps in the Baseline Analysis will be to work with clinical and patient experts to develop **effective solutions for service planning**. NHS England has stated its commitment to ensure that every patient has the right to access their services and expects the final implementation plan to be published later in 2014.

Professor David Walker, Deputy Chief Medical Officer, has been appointed as the National Lead for Rare Diseases in England. His role as a rare disease policy supporter will be to raise awareness of rare diseases.

Wales

Whilst the Welsh Government's health department is linked to the UK system in many ways, it is a separate service that is managed with different structures.

The formal consultation of the Welsh Implementation Plan began on Rare Disease Day 2014 and closed on the 23rd May 2014. 51 responses were received.

'Rare Diseases' are a new policy area for Wales; as a result the **Implementation Plan** has been constructed in a similar way to existing plans for other disease areas but takes into account and **responds to the five elements of the UK Strategy**. This standardised approach enables the Local Health Boards, which exist in every part of Wales, to understand the Plan and help the 150,000 people affected by a rare disease in Wales.

The draft Plan will make the Health Boards look for the first time at who has rare diseases in their area and how they will help them. They will produce an annual report which will describe how the Health Board's clinicians and other staff will: **work better with patient groups** and provide **better information to people with rare diseases**; **improve diagnosis and treatment** for people with rare diseases; and link with the research community.

The Implementation Plan in Wales will not just consider clinical issues but also **social care issues**. The National Institute of Social Care and Health Research (NISCHR) is the part of the Welsh Government that deals with research relating to health and social care. NISCHR will:

- Address how patients in Wales access research. **Help patients influence and benefit from research and involve** rare disease patient organisations in its work to support public and patient engagement in research in Wales
- Take the lead in ensuring research commitments in the Welsh Plan are delivered. These outline the Plan's aim to **increase the amount and quality of research carried out on rare diseases.** The Welsh Government will help make it easy for everyone who can help support relevant, successful research into rare diseases to work together.
- Help **simplify processes for agreeing what research is done** and for making research studies run well. Working with researchers and clinical genetics experts, it will support efforts to highlight the best areas to research.

The **Welsh Implementation Group for Rare Diseases** will work with the NHS to oversee the implementation process and ensure the Plan is 'fresh and relevant'.

Scotland

The Scottish Government aims to have patients at the heart of any rare disease policy and have made good progress in developing patient centred policy including the New Medicines Review and the Rare Conditions Medicines Fund. The next step towards patient-centred rare disease policy is a country specific Implementation Plan.

In July 2014 the Scottish Implementation Plan '**It's Not Rare To Have A Rare Disease**' was launched following a discussion document that was published earlier in the year. The Implementation Plan seeks to help the **300,000 people in Scotland affected by rare diseases**.

The Scottish health system is significantly different from the other UK nations: there are no NHS Trusts and instead 14 unified Territorial NHS Boards deliver services to meet the assessed needs of the population; there is no local or regional commissioning and eight Special Health Boards provide national services; Clinical guidelines are set by the Scottish Intercollegiate Guidelines Network (SIGN) and not NICE; and health and social care integration will take place within Scotland. It is therefore important that Scotland has a country-specific plan.

There are a number of areas that work particularly well for those affected by rare conditions in Scotland; for example there are local, regional and UK-wide rare disease service arrangements, meaning that **everyone is working towards the same goal of improving services**. There is recognition from health officials, however, that Scotland cannot work alone to address the needs of patients with rare diseases. There are areas in which Scotland is keen to collaborate with the rest of the UK. These include: **patient registries, developing undergraduate rare disease training, research into rare diseases and development of expert clinical centres**.

The next steps in Scotland will be to establish the **Implementation Oversight Group**, which will **agree actions, timescales and also monitor progress**.

Northern Ireland

Northern Ireland was the final UK nation to release their initial response to the UK Strategy for Rare Diseases in June 2014. The **Statement of Intent** is a high-level document, which identifies key partners and sets out how the 51 commitments will be implemented in Northern Ireland.

Northern Ireland is the only UK country to share a border with another nation and is geographically separated from Britain. This raises a number of issues and challenges in delivering services for rare disease patients. Due to the smaller size of the nation, Northern Ireland cannot always provide specialist services as they lack the necessary 'critical mass' of patients. Northern Ireland, therefore,

recognises the need to **collaborate with other home nations.** A key priority is to ensure that patients affected by rare conditions have equivalent access to genetic testing services as other patients in the UK.

The Northern Irish Government is actively working with the Republic of Ireland and in recent times have been able to provide more services within their own borders as a result. This is in contrast with previous years where it was usually necessary for patient to be flown over to England for treatment. This can be particularly difficult for families and children with very complex conditions.

It is the Northern Irish Government's intentions that a **comprehensive plan which includes measures and timescales will be published later this year**. This will follow a 12-week consultation period to allow all stakeholders the opportunity to have their say. Officials aim to have a final plan in place by Autumn 2014 but recognise that they are behind schedule.

Conclusion

The EUROPLAN National Conference was a successful day that brought together rare disease stakeholders from across the UK. The conference generated many ideas but the overwhelming theme from the day, with agreement from all stakeholders, was that **all four home nations of the UK must work together**.

Delegates raised a number of concerns; in particular, they felt that the different structures in each nation made it difficult to move forward and at times challenging to work out who has responsibility of taking commitments forward. RDUK will create a tool that will be made available to all stakeholders on the RDUK website which will: identify what bodies are responsible for leading on individual commitments; the status of the commitment in terms of implementation; and how to take action to apply pressure to relevant bodies.

Within the report there are a number of recommendations which, though specific to the UK, can be applied to many other European countries. In the UK, RDUK will work to push forward these recommendations, working with relevant stakeholders to ensure better healthcare and health outcomes for patients affected by rare conditions. We hope that the UK can then share its experiences with a view to achieving improvements in care for patients across Europe.

We were pleased to hear that attendees at the EUROPLAN National Conference found the day informative and a good opportunity to meet others working in the rare disease field. We were delighted also, that the conference has inspired attendees to take action to improve healthcare for rare disease patients.

Annexe I: Programme

EUROPLAN II National Conference

24th June 2014

Society Building, 8 All Saints Street, London N1 9RL

Agenda

9.00 – 10.00	**Registration open & networking opportunity**
10.00 – 10.10	**Welcome** – Alastair Kent OBE, Director of Genetic Alliance UK, Chair of Rare Disease UK
10.10 – 11.30	**Methodology, Governance and Monitoring of the UK Strategy for Rare Diseases** UK Strategy for Rare Diseases - Robin Herbert, Health Science and Bioethics, Rare Disease Team, Department of Health Implementation in England, Dr Felicity Taylor, National Medical Director's Clinical Fellow , Highly Specialised Services, NHS England Wales Implementation Plan for Rare Diseases, Dr Chris Riley, Department of Health and Social Services, Welsh Government

Scottish Implementation Plan for Rare Disease, Elizabeth Porterfield, Head of Strategic Planning and Clinical Priorities, Scottish Government

Implementation in Northern Ireland, Alastair Campbell, Deputy Director for Secondary Care, Department for Health, Social Services & Public Safety

UK Rare Disease Forum - Alastair Kent OBE, Chair of UK Rare Disease Forum

11.30 - 1.00 **Research into Rare Diseases**

Presentations from:

Dr Jonathan Berg, Senior Lecturer and Consultant in Clinical Genetics, University of Dundee

Dr Ian Frayling, Consultant in Genetic Pathology, Laboratory Director, All-Wales Medical Genetics Service, Honorary Senior Clinical Research Fellow, Cardiff University

Dr Amy Mcknight, Senior Lecturer and Director of Postgraduate Education and Training, Queen's University Belfast

Dr Tony Soteriou, Research Infrastructure and Growth Senior Manager, Department of Health, England

Panel Discussion, chaired by Dr Nick Sireau, Chair and CEO of AKU Society

1.00 – 1.45 **Lunch**

1. 45 – 3.15 **Centres of Expertise and European Reference Networks**

Prof. Philip Beales, Professor of Medical Genetics at the Institute of Child Health, University College London

Dr Stephen Jolles, Consultant Clinical Immunologist, Cardiff & Vale NHS Trust

Dr Fiona Stewart, Consultant in Genetic Medicine, Northern Ireland Regional Genetic Service, Belfast City Hospital

Dr Jonathan Berg, Senior Lecturer and Consultant in Clinical Genetics, University of Dundee

Panel discussion, chaired by Prof. Kate Bushby, Professor of Neuromuscular Genetics, Newcastle University

3.15 – 3.30 **Conference close**

Prof. Kate Bushby, Professor of Neuromuscular Genetics, Newcastle University

Annexe II: List of Participants

Name
Aidan Gill
Alan Thomas
Alison Tresidder
Andrew Symes
Andy Soar
Azad Haleem
Barny Foot
Becca Wright
Brian Lovatt
Bruce Faulkner-Dunkley
Cameron Bisset
Charlotte Hermans
Christiane Kellner
Christina Jones
Christoph Bettin
David Cunningham
Deborah Mann
Debra Morgan
Dee Morrison
Diane Thomson
Emma Hughes
Ewan Forbes
Fabiola Martin
George Dickson
Gillian Thomas
Hana Ayoob
Hana Salussolia
Hugh Adams
Humphrey Bowen
Hywel Williams
Ian Stuart
Irfan Khan
Jack Howlett
Jane Cox
Jane Swainson

Janet Bloor
Jayne Bressington
John-Paul Westwood
Julian Trimming
June Carr
Kate Bushby
Kate Eden
Keith Woods
Kerry Leeson-Beevers
Lara Bloom
Len Woodward
Lesley Harrison
Liz Bevins
Lizzie Perdeaux
Louise Arnold
Marcy Vasvary
Margaret Bowler
Marie Scully
Marijcke Veltman
Mark Barrett
Marta De Santis
Monika Preuss
Mustafa Munye
Natalie Fernandes
Nick Sireau
Nigel Banister
Noemi Roy
Noura Petrocco
Patricia Bissessur
Patricia Osborne
Paul Mylchreest
Rachel Austin
Rafael Yanez
Rhiannon Stephens
Rhonda Curran
Rhya Homewood
Richard Lynn
Richard Piper
Rob Hagendijk
Robert Stojnic
Rosalind Trante
Ross Moat
Rupert Purchase
Ruth Abuzaid
Sara Taiyari

Sarah Rollinson
Sarita Workman
Sasha Daly
Saskia Baron
Simon Denegri
Simon Lem
Sophie Dziwinski
Sorsha Roberts
Sue Millman
Suzanne Dobson
Tess Harris
Tonia Hymers
Tymandra Blewett-Silcock
Val Stevenson
Valentina Bottarelli
Victoria Hedley
William Davis
William Owen
Zoe Farrington