EUROPLAN National Conferences

Template for
CONFERENCE FINAL REPORT

Writing a clear Final Report is as important as the flawless execution of the National Conference on Rare Diseases. Report writing is an essential element to collect the main findings and the outcomes of the debates, so that they can be usefully fed into the work carried out at the national level to develop a RD strategy and into the EUROPLAN Recommendations to draft a National Plan or Strategy on Rare Diseases.

I. General information

Country	**UK**
Date & place of the National Conference	**16th November 2010 - Nowgen Centre, Manchester**
Website	**www.rarediseases.org.uk** **www.geneticalliance.org.uk**
Organisers	**Rare Disease UK and Genetic Alliance UK.**
Members of the Steering Committee	Alastair Kent - Director of Genetic Alliance UK (**Chair of Rare Disease UK**) Steve Potter - Represents the Orphan Diseases Industry Group Partnership Becky Purvis - Policy and Public Affairs Manager - Association of Medical Research Charities Dr Peter Corry - Paediatrician, Bradford Mark Barrett - Chair of the Orphan Disease

	Industry Group Dr Marita Pohlschmidt - Head of Research, Muscular Dystrophy Campaign Laura Gilbert - Freelance Research Consultant Dr Stephen Jolles - Clinical Immunologist, University of Wales Hospital Cardiff (**Advisor for Wales**) Marie McGill - National Lead, Single Gene Complex Need Project (SGCN) (**Advisor for Scotland**) Dr Fiona Stewart - Consultant in Medical Genetics, Belfast City Hospital (**Advisor for Northern Ireland**)
Names and list of workshops	**Standards of Care** **Patient Empowerment** **Research** **Governance** **Codification**
Chairs and Rapporteurs of workshops	Governance of a UK Strategy Chair: Alastair Kent, Chair of Rare Disease UK Rapporter: Stephen Nutt, Rare Disease UK Definition, codification and inventorying of rare disease strategy (information and training) Chair: Dr Mark Taylor, Birmingham Children's Hospital Rapporter: Lauren Limb, Rare Disease UK Standards of Care / Centres of expertise (orphan drugs and provision of treatment) Chair: Professor Kate Bushby, Treat NMD Rapporter: Buddug Williams, Rare Disease UK &

	Genetic Alliance UK Research Chair: Professor Dian Donnai, Executive Director, Nowgen Centre Rapporteur: Lauren Limb, Rare Disease UK Patient Empowerment Chair: Lesley Greene, CLIMB & Eurordis Rapporteur: Stephen Nutt, Rare Disease UK
Attachments (programme, list of participants, etc.)	Agenda List of participants Presentations

II. Main Report

The **Conference Final Report is based on the structure of the National Conference on Rare Diseases (RD), which is common to all EUROPLAN Conferences**. This has been illustrated in the document "CONTENT OUTLINE - Minimum requirements and recommended content for the WORKSHOPS of EUROPLAN National Conferences" (27/11/2009).

Each National Conference is configured in Plenary Session and Workshops. The Workshops will be set up according to specific themes and will deliver concrete proposals for the Plenary. Each Workshop will be devoted to a specific Theme and will report to the plenary on that Theme.

Conference organisers will have to account for how each theme and sub-theme has been dealt with in the National Conference and what practical solutions or proposals emerged in that area. Therefore the **Conference Final Report will be based on the Workshops' reports and cover the proposal emerged for each of the Workshops' Themes**:

Main Themes

Theme 1 - Methodology and Governance of a National Plan / Strategy (NP)

Sub-Themes

1. Mapping exercise before developing a National Plan

General initial discussion

Currently delegates felt that specialised services were commissioned in a rather ad-hoc manner and that there was little structure in this. Even if a condition does have nationally funded clinics there are still issues such as poor links with primary care and a reluctance to consult with experts at the specialised services especially when patients are inpatients. This was noted particularly by metabolic conditions where infections can escalate quickly, becoming serious if expert advice is not adhered to.

Current health services tend to be grouped into areas of speciality, particularly around organs or systems and these specialities don't always communicate with each other. Multidisciplinary clinics are important and they are one of the only ways that services are seen to improve.

There is a clear need for hospitals to have or develop care pathways and/or protocols for dealing with rare diseases, and this could be something that a national strategy is well placed to address.

Patients also felt that it was important for them to have access to their own health record.

Is there an awareness of the situation of RD in your country?

There is not enough awareness of rare diseases or of the complexity involved in caring and treating patients with rare conditions. Mechanisms can be developed and put in place (as we have seen for a selection of rare diseases in the UK) which can make huge differences to patient care but this is currently not done in any systematic way and data is not collected on many of the rare diseases that are present in the UK.

Is an inventory being made or a report?

There is currently no inventory being made of rare diseases but it is certainly something to which all delegates felt there was a great need to have.

Are the unmet needs being evaluated?

Currently, many felt the needs of patients and health professionals were not being met in the field of rare diseases but that this was something that needed to be addressed and could be addressed within a rare disease strategy.

What steps are being taken to establish national strategy?

The UK government have confirmed that they will be developing a national strategy over the summer of 2011; this will last for the duration of the current Coalition Government and possibly beyond. Rare Disease UK will be launching their report "Improving Lives, Optimising Resources – A Vision for the UK Rare Disease Strategy" on Rare Disease Day 2011, which will contain recommendations gathered from the rare disease community on what should included in a UK strategy.

2. Development and structure of a National Plan / Strategy
3. Governance of a national strategy

Structure and governance of a strategy

What general and specific objectives or priority areas can be identified?

It was felt that protocols needed to be set and that in order to do so capturing the knowledge of patients and patient organisations would be key.

Identifying what services are based around similar groups of rare diseases may also be beneficial particularly mapping out what services can be provided at local, district and specialist centres. Developing care pathways - this would help to feed into the diagnosis, continuous expert care that patients and families with rare diseases require.

It was felt extremely important to acknowledge that very often syndromes cross system boundaries, affecting various parts of the body. This means that patients need to be looked at as a whole rather than their individual parts, which is often what happens now.

Some delegates found that disease alert cards for patients can be useful in encouraging local healthcare professionals to contact a specialist when appropriate. However, not everyone likes to carry such a card as it can be seen as a symbol of a disability.

In order for the UK Rare Disease Strategy to be implemented there will need to be certain amounts of organisational and cultural change. At the present time, the NHS is undergoing a major restructure and changes to how it is organised in England and it will be important that those with an interest in this area communicate during this process how rare disease services can be included and improved.

Improvement in diagnosis must be a key objective in a rare disease strategy to ensure that people are identified and don't wait many years for an accurate diagnosis. As well as care pathways to be developed, diagnosis algorithms also need to be created and linked to these plans.

The way in which specialised services are commissioned and how new services are identified needs to be reviewed and a clearer and more transparent structure developed.

Delegates felt that services needed to move away from focusing on specific body parts/systems into a health services that looks at conditions as a whole.

Should a national strategy be developed by UK government?

The overwhelming majority of attendees to the Europlan Conference felt that the UK government should be developing a strategy together with the Devolved Administrations and in collaboration with the rare disease community. It will be important to have the involvement of all the stakeholders in the process.

Should patients participate at all phases of developing strategy?

It was felt that patients were an important partner in the development of a strategy and should be treated as such; this was agreed by all participants involved in the conference. Patients and patient organisations should be included in all phases of the development of a strategy, including areas around use of drugs and reimbursement.

In England, there is a move towards "value based pricing"[1] patient involvement is essential, currently people felt there is disconnect between the value a patient attaches to medicine and the price the health service is willing to pay.

Where training for patients and patient organisations would be useful to ensure active participation in the development of a strategy this should be looked at, it may also be that patients can be engaged in a variety of ways to ensure that everyone can participate. There needs to be a recognition that patients face different challenges to becoming involved, eg cost and time and that this needs to be factored into any participation plans.

[1] **Value based pricing**, or Value optimized pricing is a business strategy. It sets selling prices primarily, but not exclusively, on the perceived value to the customer, rather than on the actual cost of the product, the market price, competitors prices, or the historical price. The goal of value-based pricing is to better align price with value delivered.
http://en.wikipedia.org/wiki/Value-based_pricing (Feb 2011)

4. Monitoring the National Plan

Monitoring a national strategy

How could a strategy be monitored?

Delegates felt that the monitoring of the national strategy as it was implemented in practice would be imperative. It was important that this took place to ensure that the strategy was having an impact on the services and information patients and families with rare disease receive. It would also be an opportunity to identify areas that need more focus or where the national strategy may need to be adjusted slightly.

There needs to be accountability within the UK strategy for rare diseases, if funding for treatment is refused an examination of the alternatives needs to be looked at to ensure it is not a false economy.

Reporting of how the strategy is being implemented in practice needs to be the responsibility of a specific unit within the Department of Health, so that the responsibility falls within someone's remit.

The idea of having a National Clinical Director for Rare Diseases was warmly welcomed as it was felt this would give a focus to rare diseases and help with the implementation of a national strategy. Delegates felt that it was key to involve organisations such as Rare Disease UK and other patient groups in the monitoring of the strategy.

It was hoped that the new NHS Commissioning Board will play a role in the Rare Disease Strategy.

Clarity on budgets and expenditure for the Rare Disease Strategy will be required.

What type of indicators could be used once a strategy has been implemented?

Many indicators were suggested for example the number of patients diagnosed before and after the implementation of the strategy; how many registries have been established and how many patients have been recorded – although you need separate registries necessarily. ICD 11 and the universal adoption of the NHS number is a quicker and easier way to build up records and this should become part of mainstream record keeping as soon as possible within the NHS.

Referral times should be monitored and patient evaluation surveys carried out, identifying how many clusters of pathways have been created and looking ahead how much progress has been made in the development of treatments for rare diseases.

Is the evaluation on a national strategy envisaged to be done externally?

It was felt that both internal and external evaluation will be required, however delegates didn't identify one particular external body that would be suitable for this task but that is possibly something that can be identified by the Department of Health as part of its evaluation strategy. It would be advisable to have patients and patient organisations involved in this evaluation.

5. Sustainability of the National Plan

Sustainability

Sustaining the Rare Disease Strategy will be very important, particularly in terms of embedding the strategy into every day NHS healthcare services. Anecdotally, we understand that patients with rare diseases take up a significant proportion of hospital beds and developing, implementing and sustaining a rare disease strategy will help the NHS to deal with these patients far more appropriately and cost effectively.

It was also felt by many that with the pace of current research it is highly likely that many common conditions will fragment into rarer conditions over time and so the strategy will not be an isolated one but should become imbedded into every day NHS care.

Research and monitoring of rare diseases will not only benefit the 3.5 million people currently affected in the UK but will also increase our understanding of common conditions. Meaning that implementing such a strategy will have wider benefits than is maybe currently expected and will prove to be even more cost effective in the long term.

There are still issues to overcome in relation to access to new therapies, however these debates are already beginning to form and it is wise to include the rare disease aspect at this point to ensure that the balance of cost of access verses no access is properly analysed. Orphan drugs are often seen as a soft target, as in some cases they appear to be expensive but looking at the overall cost of drugs to the NHS it is a tiny proportion as the population use is low and in many cases the benefits are life changing.

Sustainability of the Rare Disease Strategy could well be achieved by spending resources effectively and not wasting resources on interventions that aren't going to help. By diagnosing patients earlier and having care pathways and channels for them to be supported the Rare Disease Strategy is anticipated to be self sufficient.

Is there a budget for a National Strategy?

Currently there is no budget for a Rare Disease Strategy but the NHS does have a budget for specialised services which mainly cover this area.

Theme 2 - Definition, codification and inventorying of RD

Sub-Themes

1. Definition of RD

In the UK, the European definition for a rare disease is used (5 in 10,000) and there is also a term used for very rare conditions, "ultra orphan". The Advisory Group for National Specialised Services (AGNSS) has recently developed a new framework for evaluating "ultra-orphan" drugs (which expanded the

previous remit which was focussed on services). The definition of an ultra-orphan disease is one that affects less than 500 people in England (NB: not the UK).

2. Classification and traceability of RDs in the national health system

Currently in the UK, ICD 10 is used, however ICD 11 will be adopted when it's ready. Although this is a step in the right direction, delegates did highlight concerns for how rare diseases will be grouped and defined in ICD 11. For example, rare chromosomal disorders collectively have a quite high prevalence, but some individual chromosomal defects affect only one or two people. You could therefore group, for example, all chromosome 1 disorders but as each has a different phenotype this may not actually be very appropriate. This is also an issue for some individual conditions which present differently in each patient despite having the same genotype. Planning of services for these conditions would be difficult if it were just based on genotype rather than phenotype as the services needed could vary greatly even if patients have the same classification.

There were also discussions on how the definitions would be defined and how to sub-define different conditions. It is very difficult to sub-define on a genetic basis and grouping around organ systems becomes difficult for multisystem conditions.

It is important that developments in science and medicine are then incorporated into codification systems. The speed of developments in identifying conditions could lead to problems in classification which needs to be addressed in some way. There are also conditions that could be classified under multiple titles e.g. porphyria which could raise issues with the classification.

The natural history of a disease should form the underlying basis of classification. There should be an overall terminology which is then further broken down within the label of that condition.

3. Inventories, registries and lists

Registries provide the data that is needed before the disease can be subdivided down and appropriately coded. Registries can therefore act as the foundation for classification.

It was felt that for registries to be useful that international collaboration may be needed for better classification of the disease. A very rare disease may only affect a few people in the country so to increase the amount of available data, registries will need to be international or collaborate with registries in other countries.

What kind of registries are kept in the UK?

In the UK registries are kept for some individual conditions (e.g. chronic granulomatosis disease) and some groups of conditions (e.g. Radar – a national renal rare disease registry, part of the UK Renal Registry). However, they are not kept on a systematic basis for rare diseases. Registries can be very

useful for coding of rare disease, as they can provide much more detail on the effects of the condition, and how phenotypes may vary between patients. This can therefore help in planning services. Those present at this meeting felt that registries would be very useful to use as a tool to measure the success of a national strategy for rare diseases.

Are there any official lists of rare diseases in the UK?

There is no official list of rare diseases or registries in the UK, but Orphanet is used by some health professionals. A suggestion was made that registries should be listed on Orphanet so that people are aware of what exists and work isn't repeated unnecessarily.

Listing registries on Orphanet would lead to improved research and collaboration and would avoid reinventing the wheel when someone in another country wanted to establish a registry for a condition that already has a registry in the UK.

Those that used Orphanet felt it was extremely useful but that awareness and funding for Orphanet needs to be raised so that it can be used to its full potential.

Listing in Orphanet would be beneficial to the registry holders as not only would it raise awareness but it would prove its credibility and therefore possibly assist in funding applications.

How are registries funded in the UK?

In the UK funding is mostly charitable, the National Institute for Health Research (NIHR) sometimes contributes to registries but not on an on-going basis. Pharmaceutical companies also sometimes contribute, if they are developing a drug to treat that condition.

Some registries, such as the UK Renal Registry, are funded by the data providers in return for clear benefits of the registries – e.g. annual reports and training. The cancer registry covers every cancer diagnosed in the UK and receives NHS money. There is the need for a similar thing for rare diseases. There is a need to say that 'rare diseases are as important as cancer'.

In peoples' experiences registries normally start from the bottom up – i.e. they are often developed by charities or interested groups with charitable funding and then may obtain funding from larger bodies later on.

Some funding bodies in the UK won't generally fund registries as they are of undetermined duration and are not good short term projects. When they are being established it is difficult to predict how long it will be before they are of optimum use and this doesn't appeal to a lot of funders. Once the registry is well-established and there are well defined cohorts of patients, funders are sometimes more willing to fund. This can make it extremely difficult to keep registries running as there is no source of regular funding. It often takes a while for a registry to reach 'critical mass' before it can be used for research and can get research funding.

Long term sustainability of registries

Registries often rely on enthusiasm and funding. Ownership issues can sometimes arise when setting up registries – there is a need to address this issue and others when first establishing the registry so that it is clearly defined. The desired use of the registry should also be clearly set out at the start of the process.

Registries exist to assist in clinical management, research, definition of disease, coding and there is no other way this could be done so well and those attending the Europlan meeting felt that therefore the NHS should provide sustained funding for registries in rare diseases.

It was felt by many that registry data enables the NHS to save money by better management of patients. Having some health economic analysis to quantify this and prove the worth of registries would be extremely useful.

Another issue affecting the long term sustainability of registries is the time it takes to input data. If this is a lengthy process clinicians are likely to lose interest. However, 'if you put rubbish in, you get rubbish out' so the data sets need to be as complete as possible to make the registry worth having. Sometimes patients are able to input their own data, and have been shown to do so very accurately, which reduces the time the clinician needs to spend on it and this option should be considered to alleviate workloads.

There is a need to capture what is already known by doctors. The work is already done by them, there just needs to be a system in place to record it. There is a need to pilot schemes that collect data from clinicians as they see patients. This however is difficult to implement because of coding issues.

Another sustainability issue may lie around patient consent, although the Europlan Conference didn't have time to go into this in any great detail.

2.1. Information and training

4. How to improve information on available care for RDs in general, for different audiences
5. How to improve access to quality information on RDs
6. How to ensure adequate training of healthcare professionals on RDs

Theme 3 - Research on RD

Sub-Themes

1. Mapping of existing research resources, infrastructures and programmes for RDs

Does a specific national rare disease research programme with dedicated funds exist?

Currently no such programme exists. There may be problems with the composition of committees on funding boards and there is a need for recognition of the important work done by rare disease research. Major funding bodies need to recognise that rare diseases have a place and that research into rare diseases helps to understand all complex diseases.

Delegates felt that there was a need for global collaboration for research into very rare diseases. National research is a problem due to the scale of the diseases and a lack of focus in this area by funders. It was also acknowledged that infrastructure funding can be very difficult to obtain, for example when setting up a registry. Infrastructure for rare disease research isn't a priority. There are many examples of good initiatives that have been started but not optimised as the resources aren't available. More consistent steady funding is required. There are also examples of where research has been funded to find the gene involved in a particular rare disease but there hasn't been any continual funding to build on that discovery.

Delegates felt that there is a need for a portal for research in rare diseases where you are able to click on a condition and see what research is being done, and who is funding it. Orphanet is a tool that could be better used for this purpose. There is also a need for better collaboration between government, charities and industry funding research to avoid duplication and to help with the sustainability of initiatives.

Is there scope for a dedicated research programme for rare diseases? How do we envisage it working?

There was overwhelming support for a dedicated research programme for rare diseases. Such a programme should focus on the quality of research being done and the outcomes of such work. There were concerns raised that such a programme with dedicated funds might enable funders to tick off rare disease research and actually detract from it, if the dedicated funds were only small. However the profile of rare diseases gained from this programme may make research more attractive to other funders – 'success breeds success'. The increased profile would lead the field further forward and therefore facilitate and encourage further research by highlighting the good work being done.

This kind of programme would have to be carefully managed and monitored. The criteria applied would be important and the definitions would need to be clear.

It was felt that the resource could come from re-allocation, not new money. There would be a slight reduction of funding from other programmes but people didn't feel it would be detrimental to other areas of research, particularly as many felt that research into rare disease often leads to advances in other more common complex disease so the benefits would have a ripple effect.

The only concern raised was that there may be the issue of the funding call getting too many proposals, resulting in having to prioritise rare diseases dependent on how many people are affected. This would result in more common rare diseases taking priority, and so is a situation that must be considered.

This programme could have an allocated pot of money to use towards funding infrastructure which is notoriously difficult to obtain in terms of rare diseases.

Most rare diseases don't get a good or fair chance at getting funding, this programme could go some way to changing this.

2. Needs and priorities for research in the field of RDs

What are the priorities for basic, clinical and translational research?

There is a need for more basic underlying research to be undertaken into rare diseases to enable clinical and translational research.

All three types of research are important – basic, clinical and translational. There is also the need for research that can lead to the production of rare disease guidelines. These guidelines then need funding to be disseminated effectively.

NICE prioritises its reviews of guidelines based on frequency. There is the need for an alternative assessment system for rare diseases.

What research should be carried out for rare diseases?

This was a very difficult question to address as individuals really felt that it depended on the condition itself and what was already taking place.

There is however a need to understand the basic epidemiology of rare diseases which can lead to clinical research and this could well be a starting point for the many rare diseases where little or no research is currently being undertaken. Having a dedicated funding stream for rare diseases would be very beneficial.

How can rare disease research be coordinated?

The Research Councils in the UK coordinate the work of various funding bodies to coordinate clinical research in the UK. They could be encouraged to put rare diseases on their agenda and set them as a priority. There is the need for UK-wide and EU-wide coordination so as to avoid duplication.

Fostering interest and participation of all interested parties including national laboratories, researchers, patients and patient organisations in rare disease research projects was felt to be a key part of coordinating rare disease research.

Universities and hospitals help to make a link between basic and translational research and in many instances this link is already there. The NIHR has done well in building up the link between different types of research.

Meetings and conferences help to strengthen the exchanges among patient organisations to share their experiences and knowledge within this area. There are also some specific programmes for funding and recruiting young scientists into rare disease research. These include PhD programmes and this is also something the Wellcome Trust does quite well and could be built upon.

3. Sustainability of research on RD

As mentioned in previous points there is a need for sustainability to be built into rare disease funding streams. There are often issues with the continuation of data collection in registries when the initial funding runs out. This was acknowledged by delegates and it was felt that any funding streams developed should include a sustainability strand.

4. EU collaboration on research on RD

Delegates felt this was imperative in the field of rare diseases . For some conditions this was already taking place and there were lots of examples of European Networks of Excellence already in existence but again sustainable funding was raised as a common concern.

Theme 4 - Standards of care for RDs - Centres of Expertise (CoE)/ European Reference Networks (ERN)

Sub-Themes

1. Identification of national or regional CoE all through the national territory by 2013

A collated list or register of the centres of excellence already in existence across the UK, may be viewed as a 'map' of centres of excellence that could define known provision and identify gaps, this could form part of the implementation of a national strategy.

Although not all rare diseases and conditions are genetic, it should be acknowledged that Regional Genetics Centres across the UK have high value in directing and sign-posting patients and colleagues for centres of excellence and specialist services available for various conditions as they themselves are expert centres for the diagnosis of rare conditions.

Many groups and organisations view the situation of establishing a service or clinic as a Centre of Expertise to be 'quite random' and sometimes down to happenstance or luck. What should be recognised as a systematic approach to funding and commissioning such services is not always the case and should be reviewed to ensure the process is more transparent and logical.

Delegates indicated that it is not always clear or understood the process for deciding what is included and detailed on the Specialised Services National Definitions Set and that this should be explored in more detail to ensure the community that uses the services in this definition set understand the process behind it.

2. Sustainability of CoE

It is important to set out the requirements for centres of excellence based on the needs of patients and not dictated by current rationing of resources in the health service, as the financial position may change in a few years.

Large hospitals [tertiary centres], are often well-placed to support many rare diseases because of the presence of specialty services already in place. Therefore existing structures should be maximised and placed in a well-supported area.

The value of multi-disciplinary care is crucial for centres of excellence for many rare diseases. The roles and input of involving and coordinating individual specialist clinicians/healthcare professionals must be recognised and protected as local pressures may not value or recognise their involvement. It was noted that currently many health professionals give up their time to work with rare disease patients because they are interested, however it was felt that time should be built into clinicians jobs to ensure this work is seen as part of their duties and not an added extra.

The role of patient groups in establishing centres of excellence and multi-disciplinary groups within an NHS healthcare provider (e.g. NHS Trust in England) can be vital. The patient organisation's role can be key to the process of applying for funding from NHS Specialised Services, identifying patients' needs and actually sign-posting and directing patients to the new service.

It is difficult to assess the cost of a centre of excellence as many have 'grown organically' over the years, but perhaps others that are now being assessed by AGNSS are getting the services costed and can present and share this information. Patient organisations can provide useful information to other groups.

3. Participation in ERN

There is a perception that there is a great deal of activity at the scientific centre of the disease, but very little understanding at the level of trying to treat the disease. This gap should be closed by disseminating information regarding best practice and research initiatives.

Patient organisations can be drivers of change in improving the research environment and research community.

4. How to shorten the route to diagnosis

Educating doctors to enable them to identify when to refer their patients on for further investigation and diagnostics is crucial.

There needs to be a trigger for the clinician to identify that the patient (in their surgery / clinic) is different to their expectation and may need to be referred to a specialist. There is a need to build the

training and Continued Professional Development (CPD) offered to non-specialists regarding dealing with rare diseases as to often this does not happen.

5. How to offer suitable care and organise adequate healthcare pathways for RD patients

Care pathways for rare diseases are vital. It is important to not only focus on a CoE or a National Centre but crucially also the integration of local services which patients will be accessing on a regular basis.

It is the right of an individual patient to have a care plan for them, with a named key worker and to expect that their pathway is determined according to best practice standards of care (if they exist). These type of care pathways do not exist for many rare diseases at present and need to be developed as part of the national strategy as without them it is very difficult for patients to gain the high quality of services they require, it can also mean that services vary depending on where people live.

The centre of excellence has a coordinating role that can define the patient pathway and therefore bring together the specialised and the local service provision, helping to guide the care that patients receive in their local healthcare to ensure that it is of a high standard and also helping to educate local practitioners to the needs of those living with rare diseases.

6. How to ensure in centres of excellence multidisciplinary approaches and integration between medical and social levels

There is a requirement to have a clinical specialist and a multidisciplinary team working with them, also involving other specialists on various transition services (where appropriate for the condition), social services and translational research.

7. How to evaluate centres of excellence

Evaluating centres of excellence should form part of the UK national strategy. New centres should be introduced in a much more organised and transparent way to ensure that centres are meeting patient need. Currently as many centres are developed due to the passion of one or two clinicians it can be hard to evaluate the service as they all vary considerably.

4.1. Orphan Drugs (OD)

8. Future of OD
9. Access of RD patients to orphan drugs Pricing and Reimbursement
10. Compassionate use and temporary approval of orphan drugs. Off label use

Theme 5 - Patient Empowerment and Specialised Services

Sub-Themes

1. Involvement of patients and their representatives in decision-making processes in the field of RDs

2. Support to the activities performed by patient organisations

3. Specialised social services: Respite Care Services; Therapeutic Recreational Programmes; Services aimed at the integration of patients in daily life

What kinds of programmes exist in the UK to support patients and families with rare diseases and or disabilities in general?

There were not any specific services identified that were either UK wide or covering England, Wales, Scotland or Northern Ireland for patients and families with rare conditions. In the UK we have a system called "Direct Payments" which allows individuals to access the funding that they are due directly so they can choose how to spend it on their care and support needs. Delegates felt this allowed greater independence for families and allowed them to choose an option of care that individuals wanted and needed. This control was felt very important.

However quite a number of concerns were also raised particularly around the lack of funding for social care services, particularly in the current economic climate as there are likely to be cutbacks.
It was felt that more stringent eligibility criteria for incapacity and other health related benefits will be enforced in the coming months, patients and families will not be able to personally afford to fill any gaps resulting from cut backs to social services and this was a concern for many.

Many felt that due to the fact that rare diseases are not currently seen as a priority that they are often "invisible" to social care services and that the services they do have are likely to be seen as targets for cut backs. It was agreed that this issue may benefit from consistent, validated and evidence based 'care pathways' designed to cover a spectrum of conditions and symptoms. It was believed that this would be particularly beneficial for those with a progressive or chronic condition.

The psychosocial issues that patients and families face are often not addressed in care packages. It was suggested that counselling should be an integral part of a patients/family care package as this is something that would be valued highly by those affected by rare diseases.

It was agreed that a strategy for rare diseases in the UK must bring both health and social care together and a point was raised suggesting the importance of integrating social care in centres of excellence. Another key period that is often overlooked is the transition period between paediatric and adult services; it was felt that this should be addressed within the centres of excellence and within the care pathways.

What kind of schemes do exist supporting access to rare disease patients to respite services?

It was agreed that there is currently no co-ordinated scheme providing respite care. It was suggested that local authorities were not interested in spending money on respite services, it was often seen as a low priority, the vast majority of respite services were provided through third sector and charitable

bodies. In addition, it was felt that charitable bodies were the greatest source of information, support, advice and advocacy.

It was suggested that there often seems to be a conflict between health and social services when they should be working in partnership. Health services claim that a patient is 'not ill enough' for respite services whilst social services claim that the patient is 'too ill' for respite services.

A point was raised that occupational health would not step in until the situation was deemed 'bad enough' for intervention. It was highlighted that in doing so, there was little opportunity for prevention of the deterioration of a patient's condition.

What can be done to improve their availability and accessibility of such services? (eg funding? Improved networking, flexibility in service provision?)

As with other sections of the Europlan Workshop 'Care Pathways' were once again suggested as way of improving availability and accessibility of services. It was felt that by coordinating information and outlining what a "good" pathway should look like could help provide a more consistent service across the UK, potentially decreasing the variations that patients and families currently receive. The Dutch example of personal budgets was also highlighted and generally agreed by the group as a beneficial scheme.

It was agreed that patient groups offered significant support and delegates suggested that there could perhaps be a role for patient organisations in providing / improving access to respite care.

There was support for the idea of a clinical liaison nurse specialist – it was suggested that this should be the nurse most appropriate to the particular needs of the person and should also have a sound knowledge of the welfare system so being able to signpost appropriately. A suggestion of a Family Officer was also raised, with knowledge of the welfare system, ability to signpost and support families through the transition from child to adult services.

Signposting was considered to be very important and it was suggested that it may be quite difficult to identify a person to be responsible for this. Contact a Family was cited as a good example of an organisation that effectively signposts. It was agreed that the more central support like this is, the more well known it would become.

How are specialised services financed? What is deemed to be a specialist service?

England:

At present services outlined in the Specialised Services National Definition Set (SSNDS) are funded by NHS Specialised Services, they also fund National Specialised Services.

In future the new NHS Commissioning Board could provide an opportunity for more consistency and reduce postcode lotteries.

Wales:

NHS Wales has recently undergone reorganisation and since April 2010 seven Local Health Boards are now responsible for planning health services for their population. For specialised services, the Welsh Health Specialised Services Committee (WHSSC) is responsible for the joint planning of Specialised and Tertiary Services on behalf of Local Health Boards in Wales.

Scotland:

National Services Division commissions nationally designated specialist services funded by top sliced funding from the Scottish Government Health Directorates.

A service may receive designation if the service need is very low/unpredictable and that there is clinical need for such a service.

Northern Ireland

The Health and Social Care Board along with 5 local commissioning groups commission services in Northern Ireland.

4. Help Lines

What kind of help lines (all diseases) exist in the UK for RD patients and professionals?

It was agreed that there already a number of excellent help lines in the UK provided by a great number of patient organisations. It was suggested that in most cases there are a wide range of specific help lines to specific conditions and few 'centralised' helpline services meaning that many patients with rare diseases may not be able to access help lines with any expertise in their condition.

How to develop or consolidate existing patient run help line services for RD?

It was suggested that an umbrella organisation such as Rare Disease UK may be useful in consolidating helpline services and perhaps acting as a signposting service. However, it was also acknowledged that currently there were not sufficient funds to carry out this work and that this would need to be addressed before any signposting or consolidated patient help line could be developed.

How to improve the service offered? How to improve visibility esp. for patients

It was suggested that support and training be offered to patient organisations to ensure a standard approach to help lines is taken. The importance of ongoing training and support for those operating the help line was also mentioned.

As part of a national strategy for rare diseases a rare disease portal should be created that could help publicise charity help lines.

It was suggested that this information could be given to patients as part of their 'care pathway', by a clinical liaison nurse specialist, genetic counsellor or other clinicians/staff that patients come into contact with.

Conclusion of the Final Report

In addition to the thematic reporting as described above, the Final Report will include:

- *An overall assessment of the usefulness of the EUROPLAN Recommendations and EUROPLAN Indicators for the advancement of a national strategy in the country.*
- *The transferability of the EUROPLAN Recommendations in the country.*

This assessment will result from the collection of the evaluation carried out at the end of each Workshop and included in the Workshop reports.

The Europlan conference was attended by over 80 delegates, including representation from the UK's health departments. The conference provided an opportunity to gather more valuable evidence to inform the recommendations that are being developed by Rare Disease UK to be published on Rare Disease Day 2011.

The EUROPLAN Recommendations and Indicators have helped in informing the work of Rare Disease UK to ensure that all aspects of a rare disease strategy are considered.

III. Document history

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