Summary of Recommendations

Recommendation 1
Guidelines be developed on coding and recording of rare diseases within relevant Irish health data systems that are consistent at European and global level. The Health and Information Quality Authority (HIQA) will have a role in this, given its functions regarding information standards, including coding standards.

Progress to date
The EU Commission Expert Group on Rare Diseases recommendation is that member states should consider adding Orpha codes to their country's health information system and explore the feasibility and resources needed to do so. ICD-11 coding is expected to commence in 2018, but it is not yet known to what degree this will enhance recognition of rare diseases. An Electronic Patient Record is being piloted with LauraLynn, a children’s palliative care service and other work is being piloted on the Epilepsy Electronic Patient Record System. Orphacodes have been applied on a pilot basis to health information systems in other European countries.

An e-Health Strategy was launched in December 2013 and an allied Knowledge and Information Strategy in May 2015. The e-health unit is progressing a number of strategic e-health programmes: Individual Health Identifier (see details under rec. 2 below); Electronic Health Record; National Children’s Hospital; Primary care IT; Cancer care e-health.

An area for Orpha Codes was created within the Proof-Of-Concept (phase 1) of the National Data Dictionary and Data Model. The link to this is here: http://hive-worx.com/demo/hwmdt/hw_objects.php?modelkey=5&key=6&title=Healthcare.

Following review and trialling of it by stakeholders, the business case for a solution was approved and procured by the HSE. As the HSE progresses with this, Orpha Code Catalogues will be included in the Data Dictionary as a Health Care Classification Standard along with others like ICD-10, Loinc, SNOMED Ct etc.

Over the coming years the data dictionary will assist in a stepped approach in developing capabilities around terminology, standards and interoperability. The HSE currently uses the Australian Modification of ICD10 (called ICD-10-AM) in its Hospital In-Patient Enquiry system with possible scope of engaging with Orphanet on rare diseases.

The EU Commission division DG SANTE has now entrusted the EU Joint Research Centre (JRC) with the development and maintenance of the European Platform on Rare Diseases Registration. Its principal goal is to enable sharing and use of rare diseases’ patient data across Europe, among the multitude of existing patients registries within and across the EU. This should facilitate basis and translational research, epidemiological and clinical trials,
other studies, along with policy guidance and support. The JRC envisages a number of endpoints of the Platform including generation centres of tools and support to promote interoperability between Rare Disease patients’ registries; a registry of European rare diseases registries; and a hub to provide access to European Rare Disease data collections.

**Recommendation 2**
The publication of the Health Identifier Bill and the forthcoming Health Information Bill.

**Progress to date**
The Health Identifiers Bill was published in 2013 and enacted in 2014. The Individual Health Identifier part of the project is now being implemented by the HSE. A revised and much expanded General Scheme of a Health Information and Patient Safety Bill was approved by the government in November 2015 and published on the Department of Health’s website. It is currently with the Attorney-General’s Office for formal drafting and with the Oireachtas Committee on Health for Pre-Legislative Scrutiny.

**Recommendation 3**
The Department of Health and the Health Service Executive (HSE) put in place over 5 years a coherent system to conduct broad epidemiological surveillance of rare disease in Ireland. This epidemiological surveillance should include profiling of rare diseases among high-risk cultural and ethnic minority groups for the purposes of appropriate neonatal screening and improving diagnosis and outcomes.

**Progress to date**
Surveillance of rare diseases in Ireland is somewhat contingent on the development of coding for the recording of each individual rare disease. ICD 11 is being developed and tested; but in the interim, the Commission Expert Group on Rare Diseases has recommended that Orpha codes be considered for use alongside existing health information codes. This therefore refers back to recommendation No 1 above and the work of the HSE in giving consideration to use of orpha codes alongside existing coding infrastructure. A review of data in patient registries in Ireland has been completed by the National Rare Disease Office.

**Recommendation 4**
A periodic national report on the epidemiology of rare diseases in Ireland be published by the Department of Health, similar to that prepared for the European EUROPLAN report, and that reporting on rare diseases be integrated into the existing HSE reporting on health and disability services.

**Progress to date**
Progress on this is contingent on tracking rare disease in health information systems. The National Rare Disease Office has now been established and funded by the HSE with funding provided by the EU Commission. In time, it's envisaged that the remit of the office may include assisting with and supporting research and population studies on rare diseases with various public partners.

**Recommendation 5**
All existing databases to be mobilised. Systems be put in place to enhance the utility of data held in relevant health service-based information systems, including hospital record, laboratory cytogenetic and molecular genetics data.
Progress to date
The establishment of the National Rare Disease Office will in time, assist with work on this recommendation. This is also linked to the recommendation on coding for rare diseases referred to above.

Recommendation 6
Irish data on Orphanet be reviewed and a plan for its development agreed, including an assessment of its relocation to an Irish centre if appropriate. This function should be supported by a National Office for Rare Diseases (further information on the role of this proposed new office is provided in Chapter 4).

Progress to date
The development of data from Ireland on Orphanet is being progressed through the National Rare Disease Office.

- A Business Plan was prepared by the Clinical Programme 2014–2015 to develop this function which was subsequently supported by the HSE. The HSE is a co-applicant on the (June 2016 – May 2019) 3rd EC Rare Diseases Joint Action (beneficiary 16). The Joint Action provides 3 years part (joint) funding for one Orphanet Information Scientist to be housed within the National Rare Disease Office (NRDO). Prof. E Treacy is the country co-ordinator for this function and national Orphanet Validator.

- Manchester ceased supporting the Irish arm of their Orphanet operation in September 2014. The Irish Orphanet function was officially launched on June 2015 at the National Rare Diseases Office (MMUH) with the Senior Information Scientist/Project Manager.

- Irish Orphanet data is in the process of being verified and expanded, according to Orphanet quality standards and quality assessment review, under the guidance of the Orphanet Ireland Country Coordinator (Prof E Treacy) and the Orphanet International coordination team in Paris.

The current position on the creation of a network on genetics and genomic medicine is that:
- The establishment of a National Genetic & Genomic Medicine Network (NGGMN) would bring together all parties within the field of genomic medicine;
- NGGMN should operate on hub and spoke basis with dedicated outreach clinics;
- The creation of a National Genetic and Genomic Medicine Network would provide a modern integrated genetic medicine service, engaged in patient care, education and translational research.

Recommendation 7
Appropriate support be given for the on-going involvement of Irish registries in relevant European collaborations, including the RARECARE and EUROCAT registries.

Progress to date
The EU Commission through its Joint Research Centre (JRC) is also active in the area of rare disease registries. It has begun an exercise of identifying registries referred to in the national plans of member states. The NRDO has performed a preliminary situation analysis of the existing Rare Disease registries this year and is in the process of assigning these known registries on our national orphanet site. The JRC is working towards a European Platform on
rare diseases registration. The EU Commission division DG SANTE has now entrusted the EU Joint Research Centre (JRC) with the development and maintenance of the European Platform on Rare Diseases Registration. Its principal goal is to enable sharing and use of rare diseases' patient data across Europe, among the multitude of existing patients registries within and across the EU. This should facilitate basis and translational research, epidemiological and clinical trials, other studies, along with policy guidance and support. The JRC envisages a number of endpoints of the Platform including generation centres of tools and support to promote interoperability between RD patients’ registries; a registry of European rare diseases registries; and a hub to provide access to European RD data collections. The National Cancer Registry of Ireland continues to be involved in the RARECARE European collaboration. An Irish representative is included on the European Joint Action on Rare Cancers. Ireland remains a partner in the EUROCAT registry.

**Recommendation 8**

An All-Ireland Network of Rare Disease Registries, covering the island of Ireland, be developed and that this network work towards enhancing and standardising rare disease registries in line with HIQA draft guidelines, data protection legislation and international best practice. This function should be supported by the new National Office for Rare Diseases.

**Progress to date**

The Programme for Government, *A Programme for Partnership Government* provides at paragraph 5.3.23 for the development of National Patient Disease Registries. The Health Information and Patient Safety Bill will support this. A revised and much expanded General Scheme of a Health Information and Patient Safety Bill was approved by the government in November 2015 and published on the Department of Health’s website. It is currently with the Attorney-General’s Office for formal drafting and with the Oireachtas Committee on Health for Pre-Legislative Scrutiny. The NRDO staff reviewed existing Irish rare disease registries in summer 2015. The Orphanet entries of the 15 active Rare Disease registries in Ireland have been updated. There is no communication between these registries or common data sets in use.

It was announced in early 2016 that Northern Ireland is collaborating with the UK Congenital Anomalies and Rare Disease Registry. As there are an estimated 280,000 Irish citizens with rare diseases, a South of Ireland rare disease registry will require the appropriate commissioning and national funding. Also support from eHealth and implementation of the Unique Patient Identifiers will be required.

The EU Commission through its Joint Research Centre (JRC) is also active in the area of rare disease registries. It has begun an exercise of identifying registries referred to in the national plans of member states. The JRC is working towards a European Platform on rare diseases registration. Note that compliance of registries with the Epic-Rare minimal dataset remains unclear.

**Recommendation 9**

The development of any future information systems provide for a rare disease code in a patient record in order that all people with rare diseases may be easily identified. The development of a Rare Disease ID Card that could be linked to a person’s PPSN should also be explored once the provisions of the proposed Information Bill have been enacted and promulgated.
Progress to date
The recommendation of the Commission Expert Group on Rare Diseases on member states giving consideration to using Orpha codes alongside existing codes in health information systems is directly applicable here. This refers back to earlier recommendations.

Codification of data within the National Rare Disease office for any spreadsheets or databases uses Orphacodes. Preliminary contact has been made by NRDO staff with the eHealth informatics designers regarding the wish to have an Orphacodes box on eHealth forms to provide the necessary data links to Orphanet files regarding correspondence of ICD codes to Orphacodes.

Recommendation 10
A rare disease research network be developed to:
- enhance the quality and relevance of rare disease research on the island of Ireland in a strategic manner in line with the priorities of this National Rare Disease Plan;
- support the integration of rare disease research within relevant forthcoming Government research policy and legislation;
- develop a clearly identifiable online presence, which would act to attract international interest and research partnerships;
- actively pursue potential international research partners; signpost new and established researchers to relevant resources and contacts;
- facilitate greater international collaboration with relevant registries, organisations and consortia, including the International Rare Disease Research Consortium;
- make proposals to the Department of Health with regard to Irish involvement in international networks such as E-Rare and engage in the rare diseases aspects of BBMRI-ERIC, ECRIN-ERIC and other EU infrastructures.

Progress to date
The HRB made 13 awards for rare disease projects in 2014/15. This includes 7 awards jointly with the MRCG partners and 6 awards through HRB only schemes (HRA and HPF). Irish data on Orphanet will benefit from an update in terms of the full scope of rare disease research underway nationally.

In November 2014, the Minister for Health announced €850,000 for investment into charity-led research priorities, which particularly benefits rare disease research. Five charities were to provide matching funding bringing total investment to €1.9M. They were to share €850,000 in State funding to take part in international research into rare medical conditions, including respiratory infections and retinal blindness. It is a practical example of collaboration between the State and the charity sector with the intention of benefiting Irish people affected by rare disease. The five charities taking part are Alpha One, Cystinosis Ireland, Fighting Blindness, the Irish Thoracic Society, and the Royal Victoria Eye and Ear Hospital Research Foundation. Only rare diseases that have patient organisations, who are members of the MRCG (and who can afford to fund research) have the potential to access this funding. This funding scheme is not just for rare diseases but for all disease areas represented by MRCG charities. In 2016, the next cohort of projects was funded by the State with €1.686M, matched by charity funding of €1.224M. The total funding of €2.91M is shared between 11 charities, including Cystinosis Ireland, Fighting Blindness, Muscular Dystrophy Ireland, Irish Nephrology Society and others. Six of the 15 projects with a total value of €1.1M address rare diseases. The next round of this joint funding initiative will open in autumn 2017.
The HRB is funding researchers internationally where no research capacity exists in Ireland, through the Joint Funding Scheme. In addition, a number of Irish-based clinical trials on rare diseases are registered on Orphanet. The information on the full scope of clinical trials is incomplete at present. These have mostly occurred in the context of Cancer Trials Ireland (formerly ICORG) conducting multi-site trials on cancer treatments.

In May 2016 a meeting took place between the Department of Health, Genomics Medicine Ireland and the National Treasury Management Agency, at which the Department was advised that an Irish genome sequencing project is to be carried out by Genomics Medicine Ireland. It was advised that this project is to run over three years and will sequence 45,000 genomes. It was also advised that €40m in funding had already been secured for the project as part of the initial investment of €150m required for the project. Some funding of the project will be provided by the NTMA.

There is currently a call under from the European Commission Horizon 2020, which relates to - Diagnostic Characterisation of Rare Diseases: Apply genomics and/or other -genomics for molecular characterisation of rare diseases. New Therapies for Rare Diseases: includes appropriate plans to engage with patient organisations; support clinical trials on substances where orphan designation has been given by the EC.

The HRB submitted a business case to join ECRIN-ERIC and BBMRI-ERIC to the DOH; the Department is currently considering this proposal. There are currently no plans for Ireland to join E-rare. The likely benefits and opportunity cost of such partnerships needs careful consideration in terms of the ability of Ireland to compete for European funding grants.

The current position on the creation of a network on genetics and genomic medicine is that:
- The establishment of a National Genetic & Genomic Medicine Network (NGGMN) would bring together all parties within the field of genomic medicine;
- NGGMN should operate on hub and spoke basis with dedicated outreach clinics;
- The creation of a National Genetic and Genomic Medicine Network would provide a modern integrated genetic medicine service, engaged in patient care, education and translational research.

**Recommendation 11**
Research on rare disease in Ireland adhere to the EURORDIS guiding principles for conducting rare disease research.

**Progress to date**
The HRB is increasingly integrating PPI (Public and Patient Involvement) in its work and this is reflected in the new strategy. The MRCG and IPPOSI are leaders in demonstrating the value of PPI. It will benefit from wider acceptance, particularly at the level of the health research funding agencies and within policy groups relating to health research.

**Recommendation 12**
The role of the designated Centres of Expertise in Ireland should include research relevant to rare disease, in particular with regard to registries, health service and translational research.

**Progress to date**
(A full summary of the participation of Irish Centres of Expertise in proposed ERNs and the support provided to them is available under recommendation 21.) The Clinical Lead and
NRDO have supported the active development of CoE’s to participate in ERNs. European reference networks (ERNs) for rare diseases will serve as research and knowledge centres, updating and contributing to the latest evidence. The EU has stipulated criteria that all ERN members must meet with regard to, among others, research and training capacity, information systems and e-health tools. The establishment of ERNs should thus facilitate Irish clinicians to undertake research and recognition of the value of undertaking research, in terms of career progression, are essential to improve the scale of rare disease research currently being undertaken.

**Recommendation 13**
Ireland becomes a member of ECRIN-ERIC in due course and that the capacity of Ireland’s five clinical research facilities to engage in rare disease research nationally or in collaboration with international collaborative research be enhanced.

**Progress to date**
The HRB submitted a business case to join ECRIN to the Department of Health; the Department is currently considering this proposal.;

**Recommendation 14**
The potential for industry collaboration in research relevant to rare diseases is explored, particularly with regard to research relevant to the diagnosis, treatment and management of rare disease.

**Progress to date**
Both the Rare Disease Taskforce and IPPOSI facilitate collaboration between rare disease patient groups and industry which is useful in this regard. National initiatives (primarily led by SFI) have also sought to increase the engagement between academia and industry. Irish involvement in IRDIRC may be considered as a way to facilitate more industry collaboration.

**Recommendation 15**
The forthcoming national biobanking plan provides national coordination and quality standards for biobanking and embraces all opportunities for rare disease research and Ireland becomes a full member of BBMRI-ERIC when the national coordination of biobanking has been established.

**Progress to date**
The development of a co-ordinated structure relating to biobanking is a prerequisite to Irish membership of BBMRI-ERIC. The HRB continues to represent Ireland in the context of development of international standards for biobanking.

**Recommendation 16**
With respect to pregnancy:

a. where family members are known to be at risk of being carriers of genes for rare diseases, they have appropriate access to pre-conception genetic testing and counselling, which can inform them about the risks involved in becoming pregnant;

**Progress to date**
Genetic counselling and pre-conceptual advice for families with rare diseases are available at the Centre for Medical Genetics at Our Lady’s Hospital for Sick Children, Crumlin. Due to increasing demand as a result of additional referrals to the service, among other things,
waiting times for these services have increased. The case for funding of additional professional staff at the centre has been submitted to the HSE.

b. making evidence-based, high-quality pre-conceptual care available to women at higher risk of having babies with rare congenital anomalies (e.g. women with diabetes or epilepsy);

**Progress to date**

Ireland’s first National Maternity Strategy – Creating a Better Future Together – which is a roadmap for the improvement of services over the next ten years, was launched by the then Minister for Health in January 2016. The strategy sets out a vision of maternity services that is about safety, quality and choice, and that places women very firmly at the centre of the service. The strategy describes a range of actions to be taken including that a dietetic service be available in each maternity network, so that the needs of women with type 1, type 2 and gestational diabetes, as well as those with other nutritional issues be addressed. Another action is that each maternity network scopes the necessity for the development of enhanced services at network level including dietetics, perinatal psychiatry, psychology, perinatal pathology, endocrinology, drugs liaison, physiotherapy and medical social work. The strategy also lists an action that any review of the maternity and infant care scheme considers the feasibility of extending coverage to include a preconception consultation and postnatal check at three to four months and/or additional postnatal GP visits where further pregnancy-related needs have been identified. These actions are directly applicable to this recommendation.

c. women are supported regarding preparation for a healthy pregnancy, including healthy diets and lifestyles, folic acid supplementation and good maternal ante-natal care, which can have a role in the prevention of a small number of rare conditions.

**Progress to date**

The DOH has established a Working Group on Folic Acid chaired by Prof Michael Turner. Its Terms of Reference are:

1. To develop a FA Policy which will include the following elements:
   - Development of population guidelines for peri-conceptual FA in pregnancy including at risk groups
   - Make recommendations for appropriate information campaigns for the general public and healthcare professionals
   - Consider the requirements for food fortification with FA.
   - Consider the requirements for surveillance of NTDs and dietary FA intakes
   - Consider research requirements
   - To take account of EU, WHO and other relevant international developments

2. The Group will be inclusive of FSAI, Safefood, HSE, ICGP, Institute of Obstetricians and Gynaecologists and representatives of Department of Health.

3. In conducting its work the Group will take account of the ongoing developments as outlined above, consider short, medium and long term measures and prioritise actions to ensure adequate folate intakes in at risk groups.

4. The Group may seek expert advice on issues as deemed necessary.

5. The Group will submit its’ recommendations to the Minister for Health for his consideration and approval.
**Recommendation 17**
The HSE Governance Committee/Group on Newborn Screening within the Integrated Services Directorate be expanded to include a patients’ advocate. The Committee should consider the population benefits of newborn screening, including whether programmes need to be expanded or modified, and the need for carrier screening. The Department of Health should also provide a policy framework for population-based screening programmes;

**Progress to date**
The Department of Health is considering a policy framework for population-based screening programmes.

**Recommendation 18**
The Department of Health consider addressing the need for a review of legislation that indirectly impinges on the Newborn Bloodspot Screening Programme.

**Progress to date**
The Department is currently undertaking a review of the policy in relation to the NSC Archive. Furthermore, the Department held a forum with key stakeholders to engage in deliberative dialogue in relation to the retention, storage and use of Newborn Screening Cards for research purposes. The outcomes from this will inform policy options.

**Recommendation 19**
Governance arrangements for ‘send out’ genetic tests be strengthened. This should include clinical guidelines for ‘send out’ tests and yearly audits of the quality and diagnostic yield of tests sent out from non-hospital sources in order to minimise wastage. A national funding perspective is required to maximise quality and cost-efficiency. Centres involved in testing should develop and use guidelines regarding the most commonly tested conditions.

**Progress to date**
The European Commission Expert Group on Rare Diseases adopted recommendations on Cross Border Genetic Testing of Rare Diseases in the EU. This covers, among others, the themes of access to appropriate testing and expert clinical genetic counselling; sharing of expertise; improving/ensuring the quality of laboratories conducting genetic testing. The recommendation recognised that cross border genetic testing (CBGT) for RD will remain necessary in the foreseeable future, due to differences in the national/regional testing offer. On this basis, specific recommendations were written for the European Commission and the Member States in their reflections or policy developments on how to ensure timely and accurate genetic diagnostics for Rare Diseases. One recommendation provided that whether genetic testing is provided on the national/regional level or on a cross-border basis, expertise should be shared at the EU (or global) level; and that the organization of the collaboration between expert laboratories should be set within the context of European Reference Networks (ERNs), as per Directive 2011/24/EU by integrating expert laboratories in the different thematic networks linked to their area of expertise. The potential for ERNs to support the process of CBGT for Rare Diseases should be explored. (This further refers to recommendation 23 below.)

**Recommendation 20**
The National Clinical Programme for Rare Disease through a National Office for Rare Diseases develop the clinical and organisational governance framework that will underpin care pathways and access to treatment for rare disease patients, particularly in the context of the transition from paediatric care to adult care.
Progress to date
A Rare Disease National Office was launched in June 2015. Annual ongoing funding of €200k is being provided and once-off funding for minor capital costs have been provided.

The overall Model of Care for rare diseases is in development by the National Clinical Programme for Rare Diseases. The Model of Care for Transition has been completed and currently is in the public consultation stage.

According to the proposed model of care, disease-specific clinical pathways published in peer-reviewed journals by Irish specialists, or international/European clinical pathways which have been reviewed by Irish rare disease specialists are linked in the public domain on the Orphanet Ireland webpage (http://www.orpha.net/national/IE-EN/index/clinical-care-pathways/).

The NCPRD has developed a Transition Model of Care document which is currently in review. A part time (2 year) temporary nurse has commenced at the NRDO to develop this rare disease transition model of care.

Recommendation 21
National Centres of Expertise (CoEs) in Ireland be identified for groupings of rare conditions, based on clinical need and built on foundations already established. There is an urgent requirement for the HSE to map out CoEs and healthcare pathways, and to acknowledge the different role and competencies of CoEs and centres providing care at local level, such mapping to be aligned with the re-organisation of Irish hospitals into hospital groupings. It is also important that broader clinical guidelines take account of the requirements of rare diseases. The potential for cooperation on an all-Ireland basis should be realised. The designation of CoEs should be in accordance with the EUCERD quality criteria for CoEs.

Progress to date
The National Clinical Programme for Rare Diseases has supported the HSE in the development of a process for the designation of CoEs in rare diseases; these would then be considered for membership of European Reference Networks. The process for designation of CoEs developed by the HSE includes a self-assessment by specialised services/centres/specialist networks based on a number of EUCERD criteria. These include:

- Capacity to provide management of rare diseases
- Evidence of expertise including relevant publications and international recognition
- Availability of multi-disciplinary care including health and social care services
- Capacity to produce and adhere to good practice guidelines for diagnosis and care
- Evidence of the development, measurement and improvement initiatives in quality of care, including patient satisfaction and quality control
- Evidence of teaching and training activities
- Evidence of involvement in research and clinical trials if appropriate
- Capacity to provide expert advice remotely/e-Health solutions
- Development of transition pathways as patients from paediatric to adults services
- Out-reach/shared care services
- Links and collaborations with patient organisations
- Arrangements for cross border care and referrals
Call for Interest

In August 2015, the Acute Hospital Division of the HSE made a call for interest for applications for self-assessment to rare disease health care providers nationally. The completed self-assessment templates were reviewed by experts and successful centres or networks were invited to submit applications for national designation as CoEs. The applications were reviewed by the NCPRD Clinical Advisory Group and Acute Hospitals Division. At the time, seven candidate centres were identified and submitted to the HSE Acute Hospitals Division for review.

The National Clinical Programme for Rare Diseases and the Department of Health encouraged designated centres of expertise to participate in European Reference Networks (ERNs) for Rare Diseases in line with the national plan. Any centres applying for membership of an ERN must have strategies in place to ensure that care is patient-centred; that patients' rights and preferences are respected; and must show a research component to their work. Hence, the recommendations in the national plan that related to empowering and protecting patients and carers, and research on rare diseases will be fulfilled in part through this process. It is expected that ERNs will have a major structuring effect by linking thematic expert centres across the EU.

With the encouragement of the HSE National Clinical Programme and the Department of Health, five centres of expertise were designated in June 2016 during the first round of calls from the European Commission for participation in European Reference Networks; three of these designated centres applied for membership of ERNs and two were approved.

This process has also involved Orphanet. This is an EU sponsored encyclopedic resource on rare diseases which also has a process to register CoEs. Professor Eileen Treacy is Ireland’s national coordinator for Orphanet. In addition, rare diseases have already been tabled on the agenda for North-South meetings. Future work to deepen cooperation between both jurisdictions is anticipated.

**Recommendation 22**

The HSE integrate CoEs into national funding planning, with provision for adequate staffing for multidisciplinary care, as well as sustainable research infrastructure for clinical investigation in addition to competitive research.

**Progress to date**

Formal bidding for funding that is structured within a rare disease framework has already featured in the annual round of the Estimates process that commenced in 2015. The result is that the HSE National Service Plan 2016 provided to 1) continue to develop the National Rare Diseases Office that will act as a national point of reference for enquiries relating to services, diagnostics and clinical trials and linked to recognised online information databases. The office will be supported to assist with potential Centres of Expertise to join ERNs as 2) continue to develop the adult metabolic service in the Mater Misericordiae University Hospital for the transition of adolescents from paediatric services.

The information provided at recommendation (10) on funding for rare disease research project is also applicable. In November 2014, the Minister for Health announced €850,000 for investment into charity-led research priorities, which particularly benefits rare disease research. Five charities were to provide matching funding bringing total investment to €1.9M. They were to share €850,000 in State funding to take part in international research into rare
medical conditions, including respiratory infections and retinal blindness. It is a practical example of collaboration between the State and the charity sector with the intention of benefiting Irish people affected by rare disease. The five charities taking part are Alpha One, Cystinosis Ireland, Fighting Blindness, the Irish Thoracic Society, and the Royal Victoria Eye and Ear Hospital Research Foundation. Only rare diseases that have patient organisations, who are members of the MRCG (and who can afford to fund research) have the potential to access this funding. This funding scheme is not just for rare diseases but for all disease areas represented by MRCG charities. In 2016, the next cohort of projects was funded by the State with €1.686M, matched by charity funding of €1.224M. The total funding of €2.91M is shared between 11 charities, including Cystinosis Ireland, Fighting Blindness, Muscular Dystrophy Ireland, Irish Nephrology Society and others. Six of the 15 projects with a total value of €1.1M address rare diseases. The next round of this joint funding initiative will open in autumn 2017.

**Recommendation 23**

The Department of Health and the Health Service Executive (HSE) encourage and support the national Centres of Expertise (when so designated) to seek recognition as EU designated Centres of Expertise or associated national centres in European Reference Networks for Rare Diseases (RD ERNs) according to the timeframe, framework and standards currently being developed at European level through the complementary work of EUCERD and the EU Cross-Border Healthcare Directive 2011/24/EU.

**Progress to date**

The National Clinical Programme for Rare Diseases has supported the HSE in the development of a process for the designation of CoEs in rare diseases; these would then be considered for membership of European Reference Networks. The process for designation of CoEs developed by the HSE includes a self-assessment by specialised services/centres/specialist networks based on a number of EUCERD criteria. These include:

- Capacity to provide management of rare diseases
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- Evidence of the development, measurement and improvement initiatives in quality of care, including patient satisfaction and quality control
- Evidence of teaching and training activities
- Evidence of involvement in research and clinical trials if appropriate
- Capacity to provide expert advice remotely/e-Health solutions
- Development of transition pathways as patients from paediatric to adults services
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membership of an ERN must have strategies in place to ensure that care is patient-centred; that patients’ rights and preferences are respected; and must show a research component to their work. Hence, the recommendations in the national plan that related to empowering and protecting patients and carers, and research on rare diseases will be fulfilled in part through this process. It is expected that ERNs will have a major structuring effect by linking thematic expert centres across the EU.

With the encouragement of the HSE National Clinical Programme and the Department of Health, five centres of expertise were designated in June 2015 during the first round of calls from the European Commission for participation in European Reference Networks; three of these designated centres applied for membership of ERNs and two were approved.

This process has also involved Orphanet. This is an EU sponsored encyclopedic resource on rare diseases which also has a process to register CoEs. Professor Eileen Treacy is Ireland’s national coordinator for Orphanet.

**Recommendation 24**

Residential respite care be available for children with rare diseases.

**Progress to date**

Respite care for children and adults with disabilities is provided by the HSE directly in some instances, or by agencies funded by the HSE to provide services on its behalf. In many instances respite services are part of the overall suite of services provided by voluntary service providers to people with disabilities under their service level agreements with the HSE. The HSE remains committed to working with all voluntary disability service providers to ensure that all of the resources available for specialist disability services, including respite services, are used in the most efficient and effective manner possible.

The HSE’s Social Care Operational Plan for 2017 aims to provide 182,000 overnight stays in centre-based respite services, in addition to 41,000 day respite sessions.

**Recommendation 25**

With respect to palliative care:

a. access is provided to appropriate palliative care for people with rare life-limiting conditions;

**Progress to date**

The HSE’s Eligibility Criteria guidance document helps ensure that each individual has access to palliative care based on need. Access is not condition / disease specific and therefore people with rare diseases are triaged and prioritised based on need.

b. guidelines are developed in palliative care provision to address the complex and multisystemic nature of many rare life-limiting conditions;

**Progress to date**

Two NCCE guidelines for adult palliative care services have so far been developed by the adult National Clinical Palliative Care Programme: one on cancer pain, and one on constipation, and a third guideline is planned for end of life care. There are no plans to develop specific guidelines for rare life-limiting conditions. In practical terms, where a
patient has a rare life-limiting condition with clinical manifestations that palliative care teams have not encountered before, they would be guided by the patient’s consultant / paediatrician and/or GP. Palliative care services also work in close co-operation with families and carers.

c. the National Development Committee for Children’s Palliative Care, chaired by the HSE, take account of the particular needs of children with rare disease in its ongoing programme of work;

**Progress to date**
The National Development Committee for Children's Palliative Care takes forward the recommendations of the 2009 children's palliative care policy. While the policy itself recognises the four different ACT categories of children with life limiting conditions, the recommendations do not distinguish between conditions and have been implemented for all children with life limiting conditions, whether their diseases are rare diseases or not.

d. the next National Cancer Strategy could elaborate further on how best to manage rare cancers, especially in the context of this National Rare Disease Plan, where there is a shared objective to detect and treat early patients with rare cancers.

**Progress to date**
The management and treatment of Rare and Less Common Cancers will be considered in the development of the next Cancer Strategy. The focus is on the potential for further centralisation of the management and treatment of rare cancers given the need to ensure that patients are seen by clinicians with sufficient experience and expertise in these cancers. Some work has already been undertaken by the National Cancer Control Programme in recent years in this area.

**Recommendation 26**
Appropriate modules relating to rare disease feature within all undergraduate and postgraduate programmes of both medical professional and carer disciplines. In addition to developing competency requirements and training programmes for medical professionals and carers engaged with rare conditions, practical experience and exposure to patients with rare conditions is advantageous.

**Progress to date**
Letters to the various colleges responsible for training in this area have issued. The bodies concerned are the Irish College of General Practitioners (ICGP), Royal College of Surgeons in Ireland and Royal College of Physicians in Ireland (RCPI). The letter from the Dept. of Health requested the bodies/colleges to consider this recommendation in the context of the organisation of training for health professionals.

The RCSI reported that its medical school creates awareness for the student of the several thousand different diseases leading to ill-health. It referred to students being equipped with all the competencies to recognise and establish a diagnosis for rare disease processes – though it advised that due to the rare nature of these conditions, clinical exposure can be low.

The RCPI reported to the Department that it facilitates training on rare diseases via a number of specialty study days for doctors on its Higher Specialist Training Programmes. It added that while it is difficult to comprehensively cover all rare diseases in its curriculum, the
College ensures that doctors on its training programmes know how to assess the relevant information and resources available for the area.

**Recommendation 27**
A system of training in rare diseases for healthcare professionals be addressed through their professional bodies with the support of all stakeholder groups, including patients and their families. Action in this area should build on initiatives already underway or in progress (as outlined in Recommendation 26 above).

**Progress to date**
Letters to the various colleges responsible for training in this area have issued. The bodies concerned are the Irish College of General Practitioners (ICGP); Royal College of Surgeons in Ireland; Royal College of Physicians in Ireland. The letter from the Dept. of Health requested the bodies/colleges to consider this recommendation in the context of the organisation of training for health professionals.

The RCSI reported that within each postgraduate discipline the management of rare conditions would be mastered by individuals within their subspecialty. Meanwhile, the RCPI replied that it had included a session on rare diseases in its 2016 Masterclass Series. The session, entitled *Clinical Update: Rare Diseases* was to take place in April 2016 delivered by the NCPRD. The college also referred to the difficulty to comprehensively cover all rare diseases in the curriculum; but that it ensures that doctors on its training programme know how to access the relevant information and resources available.

The NRDO has developed a GP/ AHP module for Rare Diseases to be made available on the NRDO website in the near future.

**Recommendation 28**
The establishment of a National Clinical Programme for Rare Diseases. A key role for this clinical programme will be the mapping, development and implementation of care pathways for rare diseases.

**Progress to date**
The National Clinical Programme for Rare Disease was established in 2014. The National Clinical Programme for Rare Diseases builds upon the work done through the public consultation processes in recent years. A multi-disciplinary working group delivers the work of the programme and is made up of range of healthcare professionals working in the area of patient care and rare diseases. The work of the programme is overseen by a Clinical Advisory Group, a committee of the Royal College of Physicians of Ireland, made up of consultant specialists from a broad range of disease specialities and both paediatric and adult services. The programme's objectives are as follows:

- **Access**: Patients with rare diseases and their families should have access to quality information and support, to enable accurate and timely diagnosis and access to appropriate specialist care.
- **Quality**: Clinical expertise for rare diseases should be provided through a network of national Centres of Excellence/Health Care Providers or at designated centres abroad.
- **Value**: Timely access to appropriate diagnosis and care should result in decreased mortality, morbidity and disability and be cost-effective. The scope of the National Clinical Programme for Rare Diseases includes the following disease categories:
  - Single gene disorders
> Chromosomal disorders
> Hereditary metabolic disorders
> Haemophilia and hereditary coagulation disorders
> Rare congenital disorders
> Rare endocrine disorders
> Neurological disorders and neuro-metabolic disorders
> Rare skin disorders
> Rare kidney diseases
> Rare eye disease
> Rare connective tissue/skeletal/autoimmune disorders
> Rare lung disorders including alpha-1-antitrypsin disorder and excluding Cystic Fibrosis.

**Recommendation 29**

The establishment of a National Office for Rare Diseases to facilitate the coordination and timely access to Centres of Expertise nationally and internationally, and to provide up-to-date information regarding new treatments and management options, including clinical trials.

**Progress to date**

A Rare Disease National Office was launched in June 2015. Annual ongoing funding of €200k is being provided and once-off funding for minor capital costs have been provided. The functions of the office include:

- Centralisation of up-to-date Irish rare disease information through Orphanet Ireland (www.orpha.net)
- The establishment of a rare disease information help line to provide patients, families and health care providers with information and support relating to rare diseases
- A website with information and links to relevant rare disease services and organisations around Ireland and Europe
- Development of national rare disease care pathways and in time, the development of rare disease registries.

The National Rare Diseases Office was launched by Minister Varadkar on June 4th 2015. It will be housed within the Mater Misericordiae Hospital until a location is available at the National Paediatric Hospital.
The NRDO provides information for patients, families and health professionals as outlined above. Information on Irish rare disease resources including: Centres of Expertise, Patient Organizations, Clinical Trials, Research Projects, Registries and Biobanks and Diagnostic Laboratories is available on Orphanet (www.orpha.net).

An information phone line and rare diseases email contact was opened in September 2015 and a 1800 number in February 2016.

The NRDO website launched in December 2015, is hosted on the HSE website. It contains information for patients and their families as well as for health care professionals. It has a dedicated page describing the Irish process for ERN enrolment.

The National Rare Diseases Information Line is currently developing services under the best practice guidelines of the European Rare Diseases Helpline Forum.

The National Rare Diseases Information Line serves as a reference for the Irish Cross-Border Directive and Treatment Abroad Scheme teams.

The National Rare Diseases Office also provides education and awareness of rare disease through dissemination activities (update as of May 2016, below)
Recommendation 30
The HSE develop a Working Group to bring forward appropriate decision criteria for the reimbursement of orphan medicines and technologies. The approach should include an assessment system similar to that for cancer therapies established under the National Cancer Control Programme and link with the CAVOMP at European level.

Recommendation 31
The HSE undertake a preliminary economic evaluation of current activity and costs for orphan medicine and technologies for rare disease patients across all hospitals settings.

Recommendation 32
Applications for the use of orphan medicines and technologies in hospitals be dealt with in the context of a national budget, rather than through individual hospital budgets, and that the HSE take account of this.

Recommendation 33
The HSE develop a publicly available annual report documenting the use of both existing and new-to-market orphan medicines and technologies in Ireland and a summary of applications received and decisions relating to those applications.

Recommendation 34
The existing horizon scanning between pharmaceutical companies and the HSE, including clinical value assessment authorities, continue and be enhanced so as to improve information available regarding orphan medicines in the pipeline and the future needs for these medicines.

Recommendation 35
The capacity to prescribe all orphan medicines and technologies for ultra-rare conditions be limited to specialist teams designated through the Centres of Expertise.
Recommendation 36
The HSE apply a set of guidelines on the prescribing of orphan medicines and technologies in Ireland. The HSE should evaluate clinical outcomes regarding use of orphan medicines.

Recommendation 37
Clinicians should provide data necessary to the monitoring of prescription patterns and pharmacovigilance, so as to ensure patient safety and high-quality healthcare.

Recommendation 38
Early dialogue between the HSE and companies who are running clinical trials in Ireland with Irish patients where license approval is imminent.

Progress to date
The HSE Acute Hospitals Division is developing the terms of reference, required membership and reporting relationship for this committee. This Committee will also be supported by the NCPRD Clinical Advisory Group. The terms of reference for the proposed Rare Diseases Technology Review Group are at final draft stage. The question of elaborating the Terms of Reference further in order to reflect some of the recommendations in the National Plan in this area has been discussed.

A chairperson has been provisionally identified subject to provision of expert pharmacologic support to advise the candidate. The rare disease drugs to be considered by the Rare Diseases Technology Review Group have not been determined yet.

The membership of the group will include:
- A minimum of three members who are consultants in rare or highly specialised diseases, recommended by the relevant professional society, faculty or college, who have content experience in the specific discipline and are approved by the Clinical Lead for the National Clinical Programme for Rare Diseases. In addition the Clinical Lead for the National Clinical Programme for Rare Diseases will be a member;
- Chief Pharmacist and an additional Pharmacist, a minimum of one member with Health Economical, Pharmacoeconomics or statistics and epidemiology expertise;
- A minimum of one invited participant from a related designated centre of expertise, recommended by the Clinical Advisory Group for the National Clinical Programme for Rare Diseases, as required, according to the speciality area;
- One representative appointed by HIQA;
- Patient Group representatives;
- Primary Care Reimbursement Services representative;
- Up to three additional members may be appointed.

The group will report to the HSE Committee for Optimising Pharmaceutical Value. Patient interest groups recently met with the National Director for Acute Hospitals and were asked for their comments on the final draft document.

The National Rare Disease Office has done some work on establishing a database of Orphan Drugs (excluding cancer and Cystic Fibrosis) that are reimbursed; the purpose of this work is to record such information on the Irish Rare Diseases Office Orphanet site. The recommendation relating to limiting the authority to prescribe to a specialised physician applies in the case of adult and children’s metabolic centres.
**Recommendation 39**

Sponsors could be offered an incentive to run trials in Ireland increasing access to innovation for Irish patients.

**Progress to date**

Progress has been made in recent years to advance the capability and quality in Ireland in relation to the design, conduct, analysis and reporting of clinical trials. The state (via agencies such as the HRB, HPRA) as well as patients, researchers, industry and clinician representatives have worked together for this. Ireland has the potential to do much more with respect to participation in European trials.

**Recommendation 40**

The principles of patients’ empowerment be integral to all aspects of this National Rare Disease Plan for Ireland, both now and in the future, in recognition of the fact that patients and their carers require significant clinical and non-clinical support.

**Progress to date**

This recommendation is partly encompassed by firstly the recommendation on ERNs by the Commission Expert Group on Rare Diseases in relation to European Reference Networks (ERNs) and the operational criteria for the assessment of Networks and Health Care Providers (HCP) to be used by the EU Board of Member States. The latter provides that each HCP which applies for membership of an ERN must have strategies in place to ensure that care is patient-centred and that patients' rights and preferences are respected; provides patients with clear and transparent information about the complaints procedures and remedies and forms of redress available for both domestic and foreign patients; regularly collects information on patient care experience within the Network's area of expertise and uses this information to make ongoing improvements; maintains transparency, including providing information to patients and the general public about clinical outcomes, treatment options, and quality and safety standards that are in place.

**Recommendation 41**

Arrangements be put in place to support the integration of the experience and expertise of rare disease patients’ organisations in the implementation and review of this first National Rare Disease Plan for Ireland.

**Progress to date**

NGOs and patient representatives in the area of rare diseases participated in the National Review Group that wrote the first National Rare Disease Plan for Ireland. A National Oversight Group was established in 2015; it was charged with monitoring the implementation of Ireland’s National Rare Disease Plan. The group is comprised of, among others, representatives from the rare disease NGO community and patients’ representatives.

The HSE’s National Clinical Programme for Rare Diseases also has patient representatives on its National Working Group for Rare Diseases. The working group is charged with carrying out the work of the programme under the auspices of the HSE.

**Recommendation 42**

Patients’ rights to appropriate assessment and treatment be realised through a recognised national Centre of Expertise or by linkage through the patient’s healthcare provider to recognised European Reference Networks (ERNs) and in the context of the EU Cross-Border Healthcare Directive 2011/24/EU.
Progress to date
(This refers to recommendation no 23.)

The National Clinical Programme for Rare Diseases has supported the HSE in the development of a process for the designation of CoEs in rare diseases; these would then be considered for membership of European Reference Networks. The process for designation of CoEs developed by the HSE includes a self-assessment by specialised services/centres/specialist networks based on a number of EUCERD criteria. These include:

- Capacity to provide management of rare diseases
- Evidence of expertise including relevant publications and international recognition
- Availability of multi-disciplinary care including health and social care services
- Capacity to produce and adhere to good practice guidelines for diagnosis and care
- Evidence of the development, measurement and improvement initiatives in quality of care, including patient satisfaction and quality control
- Evidence of teaching and training activities
- Evidence of involvement in research and clinical trials if appropriate
- Capacity to provide expert advice remotely/e-Health solutions
- Development of transition pathways as patients from paediatric to adults services
- Out-reach/shared care services
- Links and collaborations with patient organisations
- Arrangements for cross border care and referrals

In August 2015, the Acute Hospital Division of the HSE made a call for interest for applications for self-assessment to rare disease health care providers nationally. The completed self-assessment templates were reviewed by experts and successful centres or networks were invited to submit applications for national designation as CoEs.

The National Clinical Programme for Rare Diseases and the Department of Health encouraged designated centres of expertise to participate in European Reference Networks (ERNs) for Rare Diseases in line with the national plan. Any centres applying for membership of an ERN must have strategies in place to ensure that care is patient-centred; that patients' rights and preferences are respected; and must show a research component to their work. Hence, the recommendations in the national plan that related to empowering and protecting patients and carers, and research on rare diseases will be fulfilled in part through this process. It is expected that ERNs will have a major structuring effect by linking thematic expert centres across the EU.

With the encouragement of the HSE National Clinical Programme and the Department of Health, five centres of expertise were designated in June 2015 during the first round of calls from the European Commission for participation in European Reference Networks; three of these designated centres applied for membership of ERNs and two were approved.

This process has also involved Orphanet. This is an EU sponsored encyclopedic resource on rare diseases which also has a process to register CoEs. Professor Eileen Treacy is Ireland’s national coordinator for Orphanet. In addition, rare diseases have already been tabled on the agenda for North-South meetings. Future work to deepen cooperation between both jurisdictions is anticipated.
**Recommendation 43**

The proposed National Office for Rare Diseases provide support and information to patients.

**Progress to date**

(This recommendation refers also to recommendation (29))

A Rare Disease National Office was launched in June 2015. Annual ongoing funding of €200k is being provided and once-off funding for minor capital costs have been provided. The functions of the office include:

- Centralisation of up-to-date Irish rare disease information through Orphanet Ireland ([www.orpha.net](http://www.orpha.net))
- The establishment of a rare disease information help line to provide patients, families and health care providers with information and support relating to rare diseases.
- A website with information and links to relevant rare disease services and organisations around Ireland and Europe
- Development of national rare disease care pathways and in time, the development of rare disease registries.

- The National Rare Diseases Office was launched by Minister Varadkar on June 4th 2015. It is located within the Mater Misericordiae Hospital until a location is available at the National Paediatric Hospital.

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**Contact Us**

You can reach us at:

**Rare Diseases Information Line:**
Freephone 1800 240365 or 01 854 5065
(Mon-Thurs inclusive 9.30am-1.30pm)

Website: [www.rarediseases.ie](http://www.rarediseases.ie)

Email: rarediseases@mater.ie

General office queries: 01 854 5065

We are based at the Mater Misericordiae University Hospital, Dublin 7.

Please note that we do not have a public office.

**WHAT IS A RARE DISEASE?**

A ‘rare disease’ is defined in Europe as a life-threatening or chronically debilitating disease affecting no more than 5 people per 10,000. There are an estimated 6-8,000 known rare diseases affecting up to 6% of the total EU population and perhaps up to 300,000 Irish people during their lives. Approximately 70-80% of rare diseases are genetic and are present throughout the person’s entire life.

**WHO WE ARE**

The National Rare Diseases Office (NRDO) was established in June 2015 by the Health Services Executive (HSE). It is staffed by information scientists who have significant experience working with individuals and families affected by rare disorders.

**WHAT WE DO**

We provide current and reliable information about all rare diseases to people with rare diseases and their families as well as health care providers and researchers. Information includes:
- Specific disease information and clinical expertise
- Social care supports
- Patient support groups
- Rare Disease (RD) research and clinical trials, in Ireland and across Europe
- Policy information on rare diseases
- Non-directive information on the availability of rare disease specialists

**WHAT WE CANNOT DO**

We do not diagnose, treat or co-ordinate care for people with rare diseases – our role is limited to help people access reliable rare disease information. We do not offer the following services:
- Operate a rare diseases clinic
- Provide clinical advice or make a diagnosis
- Provide care management for individual cases
- Deal with complaints about specific individuals or services

**RARE DISEASE INFORMATION LINE**

We realise it can be difficult to find Rare Disease (RD) information. If our website does not have the details you are looking for please contact us via email or at our Rare Disease Information line (details below) and we will help you find the RD information you seek. We want to give the most up to date and complete information therefore it may take a few days to answer your RD information query.
• The NRDO provides information for patients, families and health professionals as outlined above. Information on Irish rare disease resources including: Centres of Expertise, Patient Organizations, Clinical Trials, Research Projects, Registries and Biobanks and Diagnostic Laboratories is available on Orphanet (www.orpha.net)

• An information phone line and rare diseases email contact was opened in September 2015 and a 1800 number in February 2016.

• The NRDO website launched in December 2015, is hosted on the HSE website. It contains information for patients and their families as well as for health care professionals. It has a dedicated page describing the Irish process for ERN enrolment.

• The National Rare Diseases Information Line is currently developing services under the best practice guidelines of the European Rare Diseases Helpline Forum

• The National Rare Diseases Information Line serves as a reference for the Irish Cross-Border Directive and Treatment Abroad Scheme teams

• The National Rare Diseases Office also provides education and awareness of rare disease through dissemination activities (update as of May 2016, below)
Recommendation 44
The National Rare Disease Plan for Ireland encompass a holistic and person-centred view of the lives of rare disease patients and their families, one that goes beyond healthcare issues.

Progress to date
All of the recommendations in the National Plan were written in the context of making awareness, research and services better for people with rare diseases. Progress with a number of recommendations points to advances that go beyond the health care setting for people with rare diseases. It is emphasised that services be person-centred. For example the National Rare Diseases Office was set up to provide support and information to people with rare diseases. Its functions includes the establishment of a rare disease information help line to provide patients, families and health care providers with information and support relating to rare diseases.

It is also pertinent that the National Clinical Programme for Rare Diseases and the Department of Health encouraged designated centres of expertise to participate in European Reference Networks (ERNs) for Rare Diseases in line with the national plan. Any centres applying for membership of an ERN must have strategies in place to ensure that care is patient-centred and that patients' rights and preferences are respected. In effect, these centres must embrace a person-centred ethos as part of the qualifying criteria to join ERNs.

Recommendation 45
The HSE and non-governmental organisations (NGOs) provide ongoing support for people living with rare diseases and that they cooperate and promote awareness of rare diseases.

Progress to date
Funding for rare diseases has formed part of the annual Estimates process that determined health allocations for expenditure in 2016. Similarly, the HSE has committed in its 2017 National Service Plan to support the designated Centres of Expertise, especially in the context of their involvement with European Networks for Rare Diseases. In addition, as referred to earlier, the National Rare Disease Office has been established for the purposes of among others, providing information on and promoting awareness of rare diseases.
Recommendation 46
The HSE and NGOs avail of the opportunity to promote awareness of and information on rare diseases on Rare Disease Day.

Progress to date
A Rare Disease National Office was launched in June 2015. Annual ongoing funding of €200k will be provided and once-off funding for minor capital costs have been provided. One of its functions will be to act as an information source on and raise awareness of rare diseases. In addition, Orphanet, which is funded by the EU Commission, provides comprehensive information on rare diseases that is directed at patients, clinicians and others in the rare disease community.

Recommendation 47
An Oversight Implementation Group of relevant stakeholders, including patients’ groups, led by the HSE be established to oversee and monitor implementation of the National Rare Disease Plan’s recommendations and associated key outputs. The HSE will report to the Department of Health using key performance indicators (KPIs) on a periodic basis in accordance with reporting requirements under the National Service Plan. It should be noted that the European Union has mandated EUCERD’s KPIs and that Ireland will have to report on these (see Appendix 5).

Progress to date
The Programme for Government, A Programme for Partnership Government provides at paragraph 5.3.26 to implement the National Rare Diseases Plan.

An Oversight Group to monitor the implementation of the National Rare Disease Plan was established in 2015. The group is largely comprised of members from the original steering group that led the drafting of the national plan. There have been a number of meetings of the group with a full agenda for each meeting, for which a progress report on the implementation of each recommendation in the national plan featured prominently. The members of the Oversight Group are as follows:

Dr. John Devlin (Chair)  Department of Health
Prof. Eileen Treacy  Health Service Executive
Dr. Anne Cody  Health Research Board
Mr. Tony Heffernan  Patient Representative (Bee for Battens)
Mr. John McCormack  Medical Research Charities Group
Ms. Avril Daly  Genetics Rare Disorders Organisation (GRDO)
Dr. Geraldine O’Dea  The Health Products Regulatory Authority
Mr. Derrick Mitchell  Irish Platform for Patients’ Organisations
Dr. Helen McAvoy  Institute of Public Health in Ireland
Ms. Helen Byrne  Health Service Executive
Mr. Philip Watt  Cystic Fibrosis Ireland
Mr. Liam McCormack  Department of Health
Ms. Caitriona Connolly  Department of Health
**Recommendation 48**
There should be an overall review of the National Rare Disease Plan prior to development of the next plan in 2019.

**Progress to date**
This interim report represents a detailed review of the implementation of the national plan. It is partly based on the interim progress reports provided to the National Oversight Group on the implementation of Ireland’s National Rare Disease Plan.