RARE DISEASE UK PATIENT EMPOWERMENT GROUP

Recommendations for the Rare Disease Action Plans

RARE DISEASE UK

SUMMARY

Following the launch of the England, Northern Ireland and Wales Rare Diseases Action Plans in 2022, the Rare Disease UK Patient Empowerment Group (PEG) has held two meetings to compare the plans and identify areas of learning and collaboration. The discussions in those sessions have been summarised into this set of recommendations.

This paper will be shared with the delivery partners for consideration in the development of future iterations of the action plans and used in our broader advocacy work around the UK Rare Diseases Framework, will from now on be referred to as 'the Framework'. It is noted that the Scotland Rare Disease Action Plan had not been published when this paper was written. We hope that this paper will contribute to the shaping of the plan.

The paper identifies:
- Actions in one action plan which should be adopted in other nations;
- Actions that would benefit from collaboration across borders;
- Actions which require expansion;
- Lastly, it identifies gaps in the action plans

This paper follows a set of recommendations developed by PEG before the launch of the action plans at the end of 2021. The recommendations put forward some actions under each of the priorities which would support the delivery of the Framework.

A common theme throughout the discussions identified that if the nations were to adopt actions from the other nations' action plans, they would be more comprehensive. As such communication, collaboration and learning between the nations is an essential tool to make robust, complimentary, effective plans.

PRIORITY ONE: HELPING PATIENTS GET A FINAL DIAGNOSIS FASTER

Genetic Alliance UK published the Good Diagnosis report in February 2022 which identifies eight guiding principles to a good diagnosis journey and four recommendations which would help to deliver these principles.

The report identified that while a timely and accurate diagnosis is essential to a positive experience, there are six other guiding principles to support a good diagnosis journey. People with rare conditions are also looking for a good diagnosis to be informed, supported, collaborative, coordinated, acknowledged and respected.

Newborn screening

Action one of the England Action Plan aims to establish a Blood Spot Task Group (BSTG). Both Wales, under action 1.6 and Northern Ireland, under action 3, also recognise the importance of working with the UK National Screening Committee to develop newborn screening in their action plans. It is a quirk of the UK political structure that an action that affects the whole of the UK is recorded in the England Action Plan.

PEG were frustrated that the terms of reference had not been published for the BTSG at the time of discussion, and were clear that the activities of the group should be disseminated in detail and that there should be clear routes for the voice of people living with rare conditions to be fed into the work.

- Key priorities for taking forward the debate on newborn screening for rare conditions included:
- Identification of funding routes to address key gaps in evidence for specific screening topics. This is to address the current situation where rare condition support organisations invest significantly in generating this evidence. A situation out of step with the majority of health decision-making in the UK.
- Clear thresholds for evidence requirements for approval of a screening programme for a rare condition. This is to address the current challenge of cycles of refusal with new evidence requirements emerging at each stage.
- Synchronisation with the launch of new therapies for rare conditions, such as gene therapies, to ensure that the full benefit of new treatments can be realised through aligned diagnostic pathways.

Recommendation: the Blood Spot Task Group should expand its membership to include a broader voice of people affected by rare conditions.

PEG welcomed Genomics England's Newborn Genomes Programme research project and are keen to see the outcomes of this work and grateful for the opportunities to feed into the study. Two key concerns were identified, the major one was that potential progress in the development of this tool for screening did not overshadow the debate around expansion or improvements to the traditional newborn screening methodology discussed above. The group were also keen to see participation and learning from the study disseminated among the four nations.

At the request of PEG members, we consulted Medics 4 Rare Diseases (M4RD) regarding education and training for health professionals under priority two, which also extended to diagnosis.

In discussion with M4RD they noted that newborn screening has potential beyond the bloodspot test to identify rare conditions. They suggested a review of the newborn examination conducted by clinicians to ensure every chance of identifying rare conditions are exploited. These types of screening can be improved through better education, training and guidelines for health professionals which will be further addressed under priority two.

Genomic diagnosis

Providing useful tools and resources for health professionals is equally important. Test directories support health professionals when searching for a diagnosis. However, PEG notes that there may be an inequity of tests available across the UK.

PEG felt that greater transparency is needed on how conditions which have ambiguous genetic causes are captured in the diagnostic pathways and in the test directory. Behcet's disease was given as an example in the discussion.

Ring 20 Research and Support UK

Whilst next generation sequencing (NGS), including Whole Genome Sequencing, is becoming more cost-effective and available on the NHS, there is still a need for karyotyping as the only way to reliably detect chromosomal abnormalities with structural changes such as ring chromosomes. It is estimated that only 1% of ring chromosome cases are reported in medical literature worldwide and as in the case of Ring Chromosome 20 Syndrome, the patient group have reported diagnostic decline in the last 5-10 years with the advent of NGS. Worryingly, this trend is likely to persist or even worsen given cytogenetics is being phased out and skills to run a karyotype will be lost. It is therefore imperative that the National Genomics Test Directory signposts appropriately for the use of karyotyping to diagnose chromosomal structural abnormalities in mosaic or non-mosaic form, especially to rule out conditions associated with refractory epilepsies – r(20) syndrome, r(14) and r(17) – where NGS and/or epilepsy gene panel testing return a negative result for epilepsy genes.

Recommendation: the Blood Spot Task Group should expand its membership to include a broader voice of people affected by rare conditions.

Non-genomic diagnosis

Given that more than 20% of rare conditions do not have a genetic cause, PEG is concerned in all the action plans, there is currently no action regarding non-genomic diagnosis. This exclusion of non-genetic conditions is also seen in action four in the England action plan, where Genomics England are responsible for further developing the Genomics England clinical research interface, potentially leaving non-genomic testing behind.

Recommendation: All nations should develop actions which support diagnosis and care for non-genomic conditions.

Undiagnosed conditions

Those affected by conditions which currently do not have a test and who struggle for a diagnosis are equally important. It takes, on average, five years for a person to receive a diagnosis of a rare condition, with some taking much longer. On this journey, the individual should receive coordinated care for symptoms and continued effort to search for a diagnosis.

The Wales action plan is most advanced in addressing the needs of people with undiagnosed conditions through the Wales SWAN Clinic. It is an innovative approach which recognises the challenges of undiagnosed conditions in an unprecedented manner. It presents an opportunity for shared learning with the other nations. PEG were keen to see that learning from the implementation of this service in Wales was reflected in progress on this topic elsewhere in the UK.

Recommendation: England, Scotland and Northern Ireland should take learnings from the Wales undiagnosed clinic in the development of their own clinics to ensure a uniform approach across the UK.

PRIORITY TWO: INCREASING AWARENESS OF RARE CONDITIONS AMONG HEALTHCARE PROFESSIONALS

Raising awareness

Raising awareness of rare conditions amongst health professionals is key to improving care for rare conditions. Signalling their importance to healthcare professionals will assist in boosting recognition of and respect for rare conditions.

Both Wales, under action 2.5 and Northern Ireland, under action 6, have identified celebrating Rare Disease Day as a tool for raising awareness. PEG welcomed plans specifically to celebrate Rare Disease Day in primary and secondary care settings as a method of raising awareness with healthcare professionals. This appears to be a rapid and cost-effective way of making progress on Priority two.

Unique

The GMS provided Unique (the rare chromosome and gene disorder support group) huge support around the awareness day in June earlier this year. Andy Stewart, Senior Communications Manager for Genomics, pro-actively contacted Unique ahead of the day to offer to help us raise awareness and highlight our event. The GMS and Unique co-produced content for the week which included short videos by Unique's CEO, two Unique families and Professor Dame Sue Hill (Chief Scientific Officer) together with some simple infographics. Ahead of the awareness day, the event was listed in the GMS newsletter, and on the day itself the GMS also shared Unique-produced content on their social media channels. It was hugely successful with great engagement and a record number of new families joining Unique!

Recommendation: All nations should celebrate Rare Disease Day in the NHS to help raise awareness of rare conditions.

Having a clinical lead will also raise the profile of rare conditions. Wales appointed a Clinical Lead and Clinical Champion for rare conditions in April 2022 and Northern Ireland has noted an action to explore the scope for a Northern Ireland Rare Disease Champion with responsibility of 'advocacy, policy influencing and acting as a central point of contact'. PEG recognises the positive impact that the Wales Rare Disease Champion has had on the delivery of the action plan in his short time in office. Again, this is a relatively cheap, pragmatic and cost-effective way of delivering priority two.

In England, rare conditions are currently grouped under Genomic Education England which raises concerns that non-genomic aspects of rare conditions will not be recognised. PEG believes that a dedicated clinical lead will be useful to raise the profile of rare conditions as a distinct healthcare issue.

Recommendation: All nations should have a clinical lead for rare conditions.

Wales also notes, under action 2.6, that they will continue to develop relationships with patient advocacy groups. PEG supports this action as these groups are one of the main routes to the voice of the rare condition community which is essential when developing the action plans so that they can accurately capture the needs of the community. Additionally, the support organisations have multiple projects which can provide best practice examples that can feed into the development of actions.

Recommendation: England, Scotland and Northern Ireland should recognise the need to continue to develop relationships with patient advocacy groups when raising awareness of rare conditions.

Education and training

Medics for Rare Diseases

M4RD welcomes the opportunity to work with the national implementation groups to roll out consistent rare disease education for the healthcare workforce. While nations will naturally have differing needs, the UK shares a training and qualified workforce across borders. Therefore a UK wide national approach to Priority 2 would likely benefit all involved. Currently the level of support M4RD has been able to provide has depended mainly on the idiosyncrasies of how each nation or implementation group works.

M4RD promotes the importance of making healthcare education patient and community inspired in order to produce tangible benefits for the community. This is in contrast to the more traditional biomedicine, system or clinician inspired education that still dominates.

A major risk to Priority 2 is the continued lack of recognition of 'Rare Disease' by educators, employers, advisory bodies and governing bodies of healthcare professionals. A major coordinated effort is needed from organisations to achieve the realisation of rare conditions in healthcare providers.
Having Rare Disease (as a group of conditions) resources available to health professionals provided by health public bodies will also indicate the relevance of rare conditions to the NHS. M4RD recommends that key health public bodies host webpages for 'Rare Disease'.
For example, by hosting a rare conditions webpage with the basic facts about rare disease as a specialty area on NICE and NHS websites, it legitimises rare conditions as a healthcare issue and acts as a first point of call when there is suspicion of a rare diagnosis or uncertainty about basic management. Currently this doesn't exist and is a huge missed opportunity for education and for improving diagnosis, care coordination and access to specialist care.

Read further in a three part blog by Dr Emma Huskinson following the publication of the England Action Plan.

PRIORITY THREE: BETTER COORDINATION OF CARE

PEG has serious concerns regarding the lack of content under priority three under the England action plan. The rare condition community has repeatedly expressed the importance of care coordination for an individual's experience when seeking care. Many rare conditions require care from a multidisciplinary team throughout their care journey, from presentation of symptoms through to management of the condition, and this can be spread between departments, hospitals and even countries. Having the support to ensure that people are receiving care in a timely manner without putting the burden on the individual is essential. PEG therefore recommends that England look to Wales and Northern Ireland for actions as well as considering important reports such as the CoOrdiNated Care of Rare Diseases (CONCORD) report, the ARDEnt report, and the recently published report exploring the transition process for young people living with rare conditions that provide recommendations to address uncoordinated care.

Genetic Alliance UK will be publishing a report for Rare Disease Day focusing on care coordination.

Transition

During discussions, transition from paediatric to adult care was recognised as an influential step in the care journey for people with rare conditions yet it is undervalued and often undersupported. Members of PEG shared that their communities often find a deterioration in their care at the point of transition to adult services. They have to re establish support and relationships which were previously in place. As life expectancy increases for people with rare conditions, more people are reaching adulthood and experiencing these negative outcomes of an unsupported transition in care.

Wales, under action 3.1, aims to 'ensure implementation of transition guidance with all paediatric patients transitioning to adult services should have a named worker and digital care plan linked to a patient passport'. Under action eight, Northern Ireland plans to provide 'more patient information on all available services, including transition from paediatric to adult care' as part of plans to develop care pathways.

The report developed by Costello Medical, Cambridge Rare Disease Network and Beacon provides insight into the importance of transition for the rare condition community through a series of focus groups in which a series of solutions have been identified to improve the transition process.

England and Scotland should look to the other action plans and these reports when developing actions regarding transition. It is important to recall that transition came thirteenth in the list of priorities for inclusion from the National Conversation on Rare Diseases survey that formed the basis of the Framework.

Alstrom Syndrome UK

Transition - Knowledge And Skills in Healthcare (T-KASH) are FREE resources that have been created with young people affected by Alström Syndrome, an ultra-rare and complex genetic condition.

Transition in health covers wide and varied aspects of young people's lives, which supports them with their medical condition/s as they mature and enter into adulthood.

To give transition more definition and meaning, the young people came up with 12 logos that cover important areas in their lives. These are the starting points for conversations with young people who are developing the knowledge and skills to manage their own healthcare. T-KASH provides a platform to work alongside transition assessments that are developmentally appropriate for the young person.

T-KASH also aims to:

- Prepare young people, families, and carers to identify and cope with change at key points in the young person's life
- Support young people to plan for their future and cope with their transfer and setting into adult services
- Support health, education, and social care teams to have wider discussions about areas such as vocational choices, confidentiality or how to develop life skills
- Through conversations with young people, T-KASH can identify training needs of professionals, so that they are skilled and confident to discuss broader aspects of the young person's life, e.g., parenting, capacity, and disclosure

Recommendation: England and Scotland should develop actions which support the transition process from paediatric to adult care.

Mental health

Wales, under action 3.6, and Northern Ireland, under action 9, address the need for mental health support. Mental health has become a growing concern for the rare condition community. The covid-19 pandemic highlighted the isolating effect of having a rare condition and the impact it can have on an individual's mental health.

PEG strongly believes that mental health actions are necessary in the implementation of the Framework in all four nations. The Genetic Alliance UK 2018 mental health report gives a series of recommendations which support delivery of mental health services for people with rare conditions.

Recommendation: England and Scotland should include an action which ensures that the mental health needs of people with rare conditions and carers are considered as part of the overall mental health strategy.

Alert cards and patient passports

Alert cards are recognised as an essential tool for the rare condition community when communicating with both health professionals and the wider public about their rare condition. The Implementation Plan for the UK Strategy for Rare Diseases identified alert cards as one of its actions to improve care coordination. Despite engagement from the community, this was not completed by the deadline of the strategy. The rare condition community continues to advocate for the development of alert cards and many support organisations have taken the initiative to develop their own versions of alert cards. The main challenge facing them is the recognition of these alert cards by health professionals as valid sources of information due to the lack of endorsement from the NHS.

Under action 3.5, Wales has committed to develop an easily used 'app' to enable a 'patient passport' for people with rare conditions which also works alongside other actions as an access point for care plans. Similarly, Northern Ireland, under action ten, has committed to a patient portal 'where a patient can view information, with access to all communications, healthcare professionals, summary care record and emergency care record.'
PEG believes that pragmatic actions such as Wales' app are valuable and cost-effective means of coordinating unplanned care in the UK, and should be considered by all four nations. This would be a good area for examining the success of the Welsh implementation and adopting elsewhere in the UK if appropriate.

Cambridge Rare Disease Network

CRDN created 'This is Me' rare disease passport in collaboration with UNIQUE feet families - a Cambridge-based community group formed for children living with rare conditions - medical professionals and the medical consultancy Costello Medical. The passport included information on the child's condition, needs, communication abilities, preferences etc., by their caregivers, to be used in clinical and non-clinical settings. To understand the need for a rare disease passport in the wider rare disease community, CRDN ran a pilot study which identified four key recommended actions:

1. Wider roll-out of the 'This is Me' rare disease passport
2. Create different rare disease passports customised for the needs of different individuals
3. Educate healthcare providers on the use of rare disease passports
4. Wider system changes to accommodate the needs of rare disease patients

The 'This is Me' rare disease passport demonstrated that there is a clear unmet need for a rare disease passport in the rare disease community. It also showed that a rare disease passport needs to be customisable to the needs of different patients, and part of wider changes to the healthcare system, to accommodate the needs of the rare disease community. Next steps will include a second larger trial phase to recruit some 200 people, over the course of a year, to pilot the passport.

Recommendation: All nations should examine the potential of a patient passport or alert card system, coordinating with each other where possible.

PRIORITY FOUR: IMPROVED ACCESS TO SPECIALIST CARE, TREATMENT AND MEDICINE

Repurposed medicines

Priority four in each of the action plans focuses largely on access to medicines in terms of delivering newly licensed treatments to the NHS in a timely manner. There are broader domains of the access to treatment topic, which include alternative methods of accessing treatments which can be equally effective at serving other groups in the community.

For example, widening monitoring from orphan medicines to include all treatments that might benefit people living with rare conditions would be useful as this allows for off-label and repurposed medicines to also be monitored.

The England action plan recognises the work already being done to support repurposed medicines but fails to include any actions. PEG members have welcomed engagement with the group at NHS England and NHS improvement leading the Medicines Repurposing Programme and continue their dialogue with respect to the evidence threshold used to enter the scheme with a view to understanding whether rare condition treatments are disadvantaged by the evidence requirement.

Recommendation: All nations should build actions which support the use of repurposed and off-label medicines and devices.

Promoting access to specialist treatments

Advances in pharmacogenomics have supported better characterizations of rare conditions and have the potential to improve treatment methods by tailoring drug treatments. They are identified in Genome UK, under pillar one, but are not addressed in any of the action plans.

PEG members felt that some rare conditions offer potential for progress in pharmacogenomics that are not being taken-up due to the focus on common complex conditions.

Recommendation: All nations should introduce actions to support the use of pharmacogenomics.

The action plans have included some existing policies which align with and support the delivery of the priorities in the Framework. This is important to ensure wider policy alignment and to ensure that new policies introduced through the action plans work alongside these existing policies.

The Framework is an opportunity to ensure that these existing policies are delivering. For example, the England action plan identifies schemes facilitating early access to medicines for rare conditions including The Early Access to Medicines Scheme (EAMS) and The Innovative Licensing and Access Pathway (ILAP). Expanding action eleven of the England action plan to monitor the progress and impact of these schemes would ensure their effectiveness.

Recommendation: England should measure the impact of the EAMS and ILAP schemes on access to medicines for people with rare conditions.

Access to advanced therapy medicinal products (cell and gene therapies)

England's is the only action plan that has a specific action which focuses on the need for a strategic approach for gene therapies and other ATMPs. This was felt to be a valuable initiative by PEG, and one that could be broadened to cover the whole of the UK, or replicated in the other three nations.

Recommendation: Wales, Scotland and Northern should consider aligning with the strategic approach in England on advanced therapy medicinal products.

Access to care

Additionally, there is a lack of actions regarding the second part of the priority around access to care. The actions appear very linear and do not consider the holistic view of other methods that are being employed when managing a rare condition. It is important to include actions which improve their wider care experience.

PEG believes that not considering access to care is a missed opportunity to improve access to already existing services. For example, care pathways are essential for rare conditions who often depend on multidisciplinary teams. They facilitate access to already existing services that are essential to the management of a condition. Care pathways are mentioned frequently in the England action plan. Wales, under action 3.2, and Northern Ireland under action 8, have set plans to develop exemple care pathways for rare conditions. PEG supports these actions as a practical plan of work which takes advantage of already existing expertise and that which will be developed through actions under priority two.

Recommendation: England and Scotland should include actions which support the development and recognition of care pathways for rare conditions.

Regarding expertise, the scarcity of specialists for rare conditions often creates inequities in access to care. Wales, under action 4.5, plans to ensure access to appropriate specialist services and notes that some services will need to be provided outside Wales for specific conditions to ensure appropriate expertise and critical mass of patients. Additionally, Northern Ireland, under action twelve, plans to improve access to other specialist teams locally, nationally and internationally. PEG recognises the value of this action but believes that this principle would be better served as a UK wide approach.

Recommendation: All nations should collaborate on identifying specialists and improving access for people with rare conditions.

UNDERPINNING THEMES

Research

PEG recognises the importance of encouraging research for rare conditions. England, Northern Ireland and Wales have identified multiple actions in increasing the allure of the rare condition research landscape. PEG notes that some activities could be performed by one nation and then shared with other nations. England, action fifteen and action four, and Northern Ireland, action thirteen, both plan to map the current research landscape and develop a database containing such information. By collaborating on this project, nations can create a more accurate picture of the landscape which can be shared with other nations and which can be shared with clinicians, people with rare conditions and researchers to improve recruitment and encourage projects where research is lacking.

Additionally, Wales, under action 1.4, plans to 'ensure a consent strategy is developed that enables researchers to securely and safely access routine genomic data generated by AWMSG for translational research purposes'. PEG encourages this type of resource to be shared between nations to: prevent repetition of activities; to improve use of capacity; and ensure consistency between the action plans.

Recommendation: All nations should identify actions which have the potential for collaboration or shared approaches between nations.

To encourage the research in the rare condition space, researchers need to be provided with useful resources, including registries. To facilitate this, the nations should take similar approaches regarding the facilitation of research and take learning from each other to make compatible research environments so that researchers can navigate with ease.

Northern Ireland, as part of action four, has engaged with leads at the other nation's registries, NCARDRS, CARIS and CARDRISS, in the development of their registry, NRADCAR. A coordinated approach to the delivery of registries will encourage their use by researchers and health professionals.

Recommendation: the four nations should collaborate in the development of their registries.

Equality, Diversity and Inclusion

The England action plan identifies health equity as a sixth underpinning theme and addresses health inequalities under action twelve of the action plan. Given the diversity of people affected by rare conditions, PEG welcomed this as an important facet which should be considered in the implementation of all actions in all plans.

Recommendation: All nations should include a message relating to equity, diversity and inclusion which is considered throughout the development and implementation of the action plans.

CONCLUSION

Through this paper, PEG have addressed the first iterations of the England, Wales and Northern Ireland action plans for rare diseases. This paper has shared a set of recommendations produced by the Rare Disease UK Patient Empowerment Group which set out:

- Actions which would benefit from collaboration between the nations
- Actions noted by PEG as high quality which would benefit all people with a rare condition in the UK and which should be adopted by all nations
- Actions which have strong potential and PEG believe should be expanded upon
- Actions which could fill common gaps in the action plans that are essential to improving the lives of people with rare conditions

Throughout this paper, we have made reference to several reports which have been developed by or in collaboration with the rare condition community which are valuable sources of information that PEG encourages the delivery groups to engage with when developing future iterations of the action plans. PEG found that collaboration between nations presented the possibility of expanding the potential of and improving the effectiveness of many of the actions in the nations' action plans. As such, PEG encourages the nations to not only engage with stakeholders, but also other nations on actions which may benefit from cross border collaboration.

PEG hopes that this paper will support the development of the next iterations of each of these plans and hopes to continue engagement with the delivery groups to ensure that the voice of the rare condition community is represented in their work.

SUMMARY OF RECOMMENDATIONS

Priority one: helping patients get a final diagnosis faster

1. The Blood Spot Task Group should expand its membership to include a broader voice of people affected by rare conditions.
2. All nations should align the respective test directories and information hub so that they offer the same test options.
3. All nations should develop actions which support diagnosis and care for non-genomic conditions.
4. England, Scotland and Northern Ireland should take learnings from the Wales undiagnosed clinic in the development of their own clinics to ensure a uniform approach across the UK.

Priority two: increasing awareness of rare conditions among healthcare professionals

5. All nations should celebrate Rare Disease Day in the NHS to help raise awareness of rare conditions.
6. All nations should have a clinical lead for rare conditions.
7. England, Scotland and Northern Ireland should recognise the need to continue to develop relationships with patient advocacy groups when raising awareness of rare conditions.

Priority three: better coordination of care

8. England and Scotland should develop actions which support the transition process from paediatric to adult care.
9. England and Scotland should include an action which ensures that the mental health needs of people with rare conditions and carers are considered as part of the overall mental health strategy.
10. All nations should examine the potential of a patient passport or alert card system, coordinating with each other where possible.

Priority four: improved access to specialist care, treatment and medicine

11. All nations should build actions which support the use of repurposed and off-label medicines and devices.
12. Recommendation: All nations should introduce actions to support the use of pharmacogenomics.
13. England should measure the impact of the EAMS and ILAP schemes on access to medicines for people with rare conditions.
14. Wales, Scotland and Northern should consider aligning with the strategic approach in England on advanced therapy medicinal products.
15. England and Scotland should include actions which support the development and recognition of care pathways for rare conditions.
16. All nations should collaborate on identifying specialists and improving access for people with rare conditions.

Underpinning themes

17. All nations should identify actions which have the potential for collaboration or shared approaches between nations.
18. All nations should collaborate in the development of their registries.
19. All nations should include a message relating to equity, diversity and inclusion which is considered throughout the development and implementation of the action plans.