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RARE DISEASE POLICY NEWSLETTER

The Quarterly Newsletter of the UK Rare
Diseases Forum



THE LATEST FROM RARE DISEASE POLICY

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ABOUT THE NEWSLETTER

Welcome to the fourth edition of the UK Rare Diseases Forum Newsletter. This is a chance to provide you – the rare disease community – with the updates on rare disease policy from each of the four UK nations. Additionally, we'll highlight relevant news stories and upcoming events that you can get involved in.

This newsletter is part of the UK Rare Diseases Forum which is our primary form of engagement with individuals across the rare disease community. The online forum platform offers an active discussion space for community members and a repository for the upload of governance structure papers.

This newsletter covers all four nations of the UK and is published to members of the Forum online platform. We welcome contributions and comments from members on any of the content posted here. For more information on the newsletter or the Forum, please contact gset@dhsc.gov.uk.



NEWS

UK Rare Disease Research Platform (MRC-NIHR)

The UK Medical Research Council and National Institute for Health and Care Research (NIHR) have recently made a significant announcement of a call for proposals to form the UK Rare Disease Research Platform, with an investment of around £12 million over five years. The Platform will support the coordination of UK rare disease research and address tractable research challenges, with the ultimate aim of a step change in the mechanistic understanding, diagnosis and therapy of rare diseases. The Platform will fully engage with the full range of crucial stakeholders in rare disease research, including patients and families, academic, clinical and industry researchers, policy makers and regulators.

Full information is available on [the Funding Finder webpage for this call](#), and any queries can be directed to rarediseaseresearch@mrc.ukri.org.

New UK Rare Disease Research into Pregnancy Associated Osteoporosis Study

The Pregnancy Associated Osteoporosis Study (PAO) opened to recruits in June 2022. This ground-breaking study, which is led by Professor Stuart H. Ralston at the University of Edinburgh aims to find out more about the underlying causes of PAO as well as the impact which the condition has on the quality of life of those affected, both at the time of the original diagnosis and later in life. It is a very rare condition with an estimated prevalence of 4-8 women affected per million births, although the exact number of women affected during pregnancy is not known. The usual presentation of PAO is with severe back pain caused by fractures of the spine towards the end of pregnancy or shortly after childbirth. Attempts to find out more about the condition have been hampered by the fact that it is so rare and that there isn't even a diagnostic code for the disorder in GP medical record systems.

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This ground-breaking new UK research is rare disease patient centric and focussed. The study is open to any woman who has experienced the onset of osteoporosis during pregnancy. The research team are using a two-pronged approach to enrol women who have experienced the disease. The first is to encourage women who have been affected to self-enrol into the study and complete a questionnaire giving details of how their condition presented, potential risk factors and how their physical and mental health was affected. More details can be found at the [University of Edinburgh website](#). Secondly the research team are contacting specialists in bone disease across the UK to gather information on patients currently being treated for the disease. Participants who volunteer will have the opportunity of providing a blood sample for genetic and epigenetic studies as well as studies on biochemical markers of the disease.



The long-term aim is to develop a scoring system for the risk of PAO and recurrence during further pregnancies, through a combination of clinical risk factors, genetic, epigenetic and biochemical profiling. This could allow high risk women to be offered enhanced surveillance and would inform the design of a future clinical trial for secondary prevention of fractures in PAO. If further funding can be obtained, there are additional aspects of this disease about which little is known and where additional research would be beneficial, not least regarding the children of the women diagnosed with PAO.

Research Project on adult patients living with Inherited Retinal Degenerations

Belfast Trust Ophthalmology Consultants and Medical Genetics colleagues have been working on a research project that has provided information on adult patients living with Inherited Retinal Degenerations. The project which includes an All-Ireland collaboration with researchers in Dublin has provided many benefits in terms of being able to give a firm diagnosis and pattern of inheritance. Through funding from voluntary partners, the Trust research team have been able to identify about 80% + of individuals with Inherited Retinal Degenerations in Northern Ireland, genotyping 50% of these individuals and been able to provide NHS accredited confirmation to around 125 patients. Recently the Trust has been able to secure further funding through partnership with a Pharmaceutical body which will enable genotyping for the remainder of the cohort. The genotyping in the adult population is now helping to provide diagnoses for some of the paediatric population through families known to the Trust and has also facilitated improved discussions between adult and paediatric Ophthalmology and Medical Genetics.

NEWS

SWAN (Syndrome Without A Name) clinic

In working to address the top priorities of the UK Rare Disease Framework, the Welsh government funded a two year pilot programme to establish Syndrome Without A Name (SWAN) clinics. These are provided by Cardiff and Vale University Health Board and work using genetic and other diagnostic approaches and where possible to direct this information to improve treatment. The team also includes a nursing care coordinator to aim to help patients navigate the often multiple health professionals needed for their care.

There are both adult and paediatric SWAN clinics. The adult SWAN clinic is led by Professor Stephen Jolles and Dr Ian Tully and the paediatric SWAN by Dr Jennifer Evans and Dr Jennifer Gardner. The adult SWAN clinic is inviting referral of adults who have the involvement of two or more systems from the following list: cardiology, respiratory, gastroenterology/hepatology, metabolic, endocrinology, nephrology, haematology, rheumatology, immunology, dermatology, growth disturbance, mental health or other; and who are suspected of having a unifying underlying diagnosis. Patients and their families should be informed of the referral, and the patient will remain in the care of the referring lead clinician. Information on referral process has been issued to all health boards in Wales, number of referrals has been low so far.

The goal is to shorten the diagnostic odyssey, improve coordination of care and access to specialist care and increase rare disease awareness among healthcare professionals.



NEWS

Genomic Notes for Clinicians

Health Education England's Genomics Education Programme (GEP) has developed an exciting new flagship resource for healthcare professionals, in collaboration with expert working groups across the NHS. [GeNotes](#) – or genomic notes for clinicians – is being released on a public beta basis, with two packages of resources now available to access. GeNotes offers 'just in time' education on when, why and how to request genomic testing as well as feeding back results to patients, and is based around – and designed to support – the latest [National Genomic Test Directory](#). But that's not all. GeNotes is the first resource of its kind to offer both practical and educational information on genomic testing to NHS clinicians at the point of need, underpinned by supporting education for those keen to extend their knowledge. Organised into two tiers, GeNotes offers '[In the Clinic](#)' scenarios focused on the point of patient care, while the '[Knowledge Hub](#)' acts as an encyclopaedia of educational resources to provide learning opportunities for those keen to extend their knowledge.

Find out more about this fabulous new [resource on the GEP website](#).

The Children, Young People, Families and Adult Learning Third Sector Fund

Third-sector organisations with a presence in Scotland are being encouraged to apply for the new Children, Young People, Families and Adult Learning Third Sector Fund. This will be focused on providing core funding to organisations delivering services and support that are targeted towards positive outcomes for children, young people, families and adult learners.

The Scottish Government policy priorities on which the Fund is focused include Mental and Physical Health and Mental Wellbeing.

We are therefore encouraging Scottish rare disease third-sector organisations to apply and share details of the announcement among their networks. Applications are now open until 3pm on Monday 5 September 2022.

Full details can be found at the [CORRA Foundation website](#).



NEWS

Scientists recommend updating global guidelines to improve rare disease diagnosis

Work led by scientists at Genomics England, The University of Manchester and The University of Oxford has led to the proposal that [global guidelines](#) for clinicians and researchers be expanded to allow them to better leverage the potential of using whole genome sequencing (WGS).

With the cost of WGS falling and its use in clinical practice rising, ever larger numbers of potentially disease-causing variants are being detected. However, the guidance criteria isn't currently in place to assess their impact on rare disease.

The scientists want the new recommendations to sit alongside the existing guidance, allowing researchers to take full advantage of WGS. This will lead to the provision of definitive diagnoses for more patients whilst also uncovering novel mechanisms of disease. Many families remain without a diagnosis for years and work like this is vital to changing that. Genomics England are proud that research participants in the 100,000 Genomes Project have contributed to such a pressing need in the rare disease community.



Northern Ireland All Party Group (APG)

An All-Party Group (APG) on Rare Diseases was officially approved on 25 February. However, due to the Northern Ireland Election on 5 May 2022, the first APG meeting took place via zoom on 22 June and the APG was successfully reconstituted in the new mandate. Once up and running, the cross-party nature of the APG will provide a much-needed voice at the NI Assembly for the rare disease population, ensuring wider policy alignment by raising issues affecting the community and increasing awareness of rare conditions within the NI Assembly, working together to influence positive change for those affected by rare conditions.



POLICY UPDATES

Northern Ireland

At the Northern Ireland Rare Diseases Implementation Group (NIRDIG) meeting on 26 April, members agreed on the formation of 5 working groups and leads to take forward the agreed actions for 2022-23. The majority of the Working Groups are underway, meetings have been convened, work plans agreed and initiated. Implementation plans for each group were presented at the NIRDIG meeting on 23 June 2022. Going forward, working groups will be invited to present updates on progress at the NIRDIG meetings. The next NIRDIG will be taking place on 25th August.

England

The England Rare Diseases Framework Delivery Group is the group responsible for developing and monitoring the delivery of rare diseases action plans for England. Following the publication of the first England Rare Diseases Action Plan in February 2022, the Delivery Group continues to meet regularly at an 8 weekly schedule to report on the progress of commitments from the first action plan, and work towards developing new actions for inclusion in future plans.

At each of the two meetings since the last newsletter, delivery partners have reported on their commitments. All actions which were reported on were on track for delivery, although due to pressures associated with monkeypox NHS England were unable to provide an update on the progress of actions in time for the latest meeting.

In developing actions for the next action plan, the group has been working to address areas of focus including holding 'deep dive' discussions on mental health, and access to advice and resources – with invited experts in each of these areas invited to meetings to share their experience and advice. The delivery group will meet next on 13 September.

The DHSC team continues to engage with the rare diseases community and key stakeholders. Following feedback from the UK Rare Diseases Forum, DHSC is in discussions with Genetic Alliance UK on the development of a group to amplify patient voice in the development of future England Rare Diseases Action Plans.

POLICY UPDATES

Wales

The [Wales Rare Diseases Action Plan](#) was published on the 16 June, supported by newly launched [webpages](#) on the NHS Wales Collaborative website, and a new [Twitter account](#) (@NHSW_RDIG). The action plan was developed by the Wales Rare Diseases Implementation Group (RDIG) sets out a number of key actions based around four priority areas. It is a five year plan that will be reviewed annually, RDIG will be working with all stakeholders including health boards, commissioners, health care professionals and third sector to take forward its implementation and monitor progress.

National Developments

- The [Core Team](#) for RDIG are now in place, with Prof Iolo Doull as Chair, Dr Jamie Duckers as National clinical lead for rare diseases and Rhiannon Edwards as RDIG Coordinator supported by Project support from NHS Wales Collaborative.

Rare Diseases Implementation Group (RDIG) Related Work

- A clinical fellow has been funded, with the advertisement to be published shortly to work between research activities and the SWAN clinic, including supporting the development and validation of PROMS and PREMS within the ongoing evaluation of the clinic.
- The Rare Disease patient passport, supported by RDIG will be shortly piloted in a patient population. With presentations to show our learning arranged in the NHS Wales Confederation Conference in Autumn.

Discussions regarding raising awareness of rare diseases and education opportunities for clinicians continue, including the consideration of an online platform to support both patient and clinical needs.

Scotland

Scotland's Rare Disease Implementation Board last met on 17 June 2022 and discussed a proposal to convene small working groups that will drive implementation of the actions to be set out in our Action Plan. We subsequently discussed with the proposed working group leads what steer and support they may require, and suggestions for group memberships. This will ensure that we have the implementation structures in place when we publish our Action Plan later this year. Scotland's Action Plan will be published in alignment with the Scottish Government's current reassessment of priorities, resulting in a stronger Plan that has a greater impact for people in Scotland living with a rare disease. The next Rare Disease Implementation Board meeting is scheduled for Thursday 22 September.



EVENTS

Community events

- The Multiple System Atrophy Trust is hosting a study day for Health and Care Professionals in Cardiff on Thursday 17 November. With a broad variety of topics covered this is a great chance for professionals to further develop their knowledge of MSA and network with colleagues. Tickets are £25 and are available, along with the programme, at [the MSA trust website](#).
- Alex TLC are hosting an online meeting on 23 September. Alex, The Leukodystrophy charity is a patient support group for individuals and families affected by a genetic Leukodystrophy. Find out more at [the Genetic Alliance UK website](#).
- The Cambridge Rare Disease Network is hosting [RAREfest22](#) on 25 and 26 November. This is a 2-day rare disease inspired festival with interactive exhibits, talks, film and art. It aims to showcase science, technology and organisations which are helping to improve the lives of those affected by rare diseases.
- Beacon for rare diseases is hosting a [Drug repurposing for rare disease](#) conference on the 10 October. The conference, open to all, will bring patient groups, researchers, and industry professionals together to highlight the role that drug repurposing can play in lowering the cost and accelerating the development of rare disease treatments.

Governance events

- Northern Ireland Rare Diseases Implementation Group: 25 August 2022
- Welsh Rare Disease Implementation Group: 13 September 2022
- England Rare Diseases Framework Delivery Group: 13 September 2022
- Scotland Rare Diseases Implementation Board: 22 September 2022
- UK Rare Diseases Framework Board: 8 November 2022
- UK Rare Disease Forum: 18 October 2022