Office for Rare Conditions Newsletter February 2019

The Office for Rare Conditions, based at the Royal Hospital for Children and the Queen Elizabeth University Hospital in Glasgow, aims to raise awareness of rare conditions, enhance the quality of care provided and promote participation in multi-centre research.

Upcoming Events

Fun Event for Children with Rare Conditions on **Saturday 2nd March,** we will hold our second **Family Fun Event** for families with rare and undiagnosed conditions at TouchBase in Kinning Park, Glasgow.

The **Office for Rare Conditions** new **Monthly Seminar Series** is now running and our next seminar will be held on **19th February, Teaching & Learning Centre, QEUH,** presented by Dr Martina Rodie, titled: **Care Pathways for Common Rare Conditions**. For more information on these seminars contact: info@officeforrareconditions.org.

Family Information Event
Saturday 23rd March 2019

As part of our Rare Disease Day events, we are hosting an information day for patients with rare conditions, their families/carers as well as professionals working with rare conditions. This event will also launch the new Genetic Alliance Toolkit. For more information register your interest here

Celebrate Rare Disease Day 2019!

Rare Condition Family Fun Event

Sat 2nd March: 11am-2pm
TouchBase, Kinning Park

If you have a child with a rare condition then come along to our #rarediseaseday fun event!
There will be lots of fun activities for children and a chance to meet with other families and children living with rare and undiagnosed conditions.

Spotlight on: Rare Disease Day – what's it all about?

Rare Disease Day began on 29th February 2008 across Europe and Canada and was organized by EURORDIS (European Organisation for Rare Diseases). In 2009 it expanded thanks to NORD (National Organization for Rare Disorders) and China, Australia, and the US (to name only a few) took part. Why 29th February? Because the day itself is a "rare day". Rare Disease Day is now a global annual event held on the last day of February and the aim is to raise awareness of the rare conditions that people live with. It would be fair to argue that the day aims to improve on accessibility to services for those living with a rare condition and for their families.

To raise awareness of rare conditions, the Office will also be present in the atrium of the Royal Hospital for Children and QEUH on **Rare Disease Day: Thursday 28th February.** Come along and speak to us! It would be great to see you..

Fundraising

Money donated to the Office through Glasgow Children's Hospital Charity is used to help raise awareness of rare conditions and to provide support to people of all ages as well as their families in a variety of ways. If you would like to raise funds for the Office for Rare Conditions please get in touch.

We would like to extend our gratitude to everyone who donates to the Office.

If you would like to raise money for the Office for Rare Conditions please get in touch with us

A special thank you!

Our thanks to all who have donated through Glasgow Children's Hospital Charity; in particular, we would like to thank the **Darius Najafian Memorial Fund**. Darius sadly passed away in 2016 just 2 weeks before his 2nd birthday from an undiagnosed condition. His family and friends want to provide support to children and families living with the uncertainty of a rare condition.

Thank you also to **Another Star in the Sky – Ahmar Javed.** Ahmar was only 13 years old when he passed away suddenly in 2017 due to a rare condition that affects the connections between blood vessels and is called arteriovenous malformation (AVM). His family, friends and schoolmates continue to fundraise to increase the awareness of rare conditions and to promote research in rare conditions such as AVM.

Cross Party Group on Rare, Genetic & Undiagnosed

The Cross Party Group (CPG) on Rare, Genetic and Undiagnosed Conditions meets regularly in the Scottish Parliament with the aim of raising awareness of these conditions and to discuss relevant government policy. On the 5 February 2018, the CPG met to discuss European Reference Networks (ERNs). ERNs are virtual networks involving Reference Centers across Europe ec.europa.eu/health/ern_en. They aim to tackle complex or rare conditions that require highly specialised treatment and a concentration of knowledge and resources. Services in Glasgow are approved by the EU in the field of rare endocrine (Endo-ERN), bone (ERN-BOND) and epilepsy (EpiCare). At the CPG meeting, Scottish clinicians explained that they may not be able to participate in ERNs after Brexit. The UK would also be unable to take advantage of research collaboration and unable to contribute or benefit from innovation. The CPG agreed to undertake a programme of work to raise awareness of this issue and to call for the UK's involvement in ERNs to be protected (protect-erns.eu). If you are interested in the work of the CPG and would like to find out more, please contact Natalie Frankish Policy and Engagement Manager at Genetic Alliance UK: natalie@geneticalliance.org.uk

Patent/Parent Reported Experience Measures (PREMs)

The Office for Rare Conditions has enlisted the help of UoG Rare Diseases Society facebook.com/gurdsociety/. Students from this society will be assisting the Office in collecting information on PREMs from families with rare conditions who visit the Royal Hospital for Children.

Spotlight On Rare Conditions Affecting Sex Development

International Disorders of Sex Development (www.i-dsd.org) is a global registry for a wide group of rare conditions affecting sex development and its activities are organized by the Office for Rare Conditions. A symposium is organised every 2 years to provide a platform for research in DSD and the 7th I-DSD Symposium will be held in Sao Paulo, Brazil from 4-6th July 2019 – the first time the symposium has been held outside of Europe. Further details are available at I-DSD.

The Scottish DSD Network, a Managed Clinical Network will hold its annual professional education event on the 8th March 2019. For further information visit: https://www.sdsd.scot.nhs.uk/sdsd-events/

An information event will be held on the QEUH campus for Klinefelter Syndrome on the 7th September 2019 for affected people and their families.

The UK Government's Equalities Office has launched a call for evidence to understand the experiences and needs of all people who may have any condition that may affect their sex development. The call is also open for parents, health care professionals or other service providers. For further information visit https://www.gov.uk/government/consultations/variations-in-sex-characteristics-call-for-evidence

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Neonatal Working Group

The Office for Rare Conditions Working Group care pathway appraisal tool is now approved and in use. The group have begun the appraisal process for 10 rare conditions which are seen regularly in the Neonatal Units in Glasgow. These are gastroschisis, exomphalos, neural tube defect, hydrocephalus, tracheoesophageal fistula, congenital diaphragmatic hernia, Hirschsprung's disease, imperforate anus, duodenal atresia and malrotation with volvulus. Once the appraisal process is completed the results will be available on the Office website. The purpose of the care pathway appraisal will be to encourage the development of robust care pathways, which are fully accessible to parents and professionals.

Patient Advisory Group: Update

The Patient Advisory Group (PAG) met on 10th February. The Office has approached the University of Strathclyde, Department of Digital Health, to assist us with the development of a patient held care summary for rare conditions. PAG met with Michael Connolly, MPhil student, who will be working with us on this project. The group discussed upcoming events to celebrate Rare Disease Day. Members helped plan events and are organising some of their own to raise awareness of rare conditions in their own communities. The group welcomed Wendy Meek as the new deputy Chair of PAG.