Office for Rare Conditions

Newsletter

September 2017

Approximately 10,000 children with rare or undiagnosed conditions attend the Royal Hospital for Children in Glasgow each year. The Office for Rare Conditions, based at the Children's Hospital 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.

Our Team

Since launching in January 2017, the office team has expanded to include Jillian Bryce (International Liaison and Registries), Sam Okure (Software Engineer) and Hannah Van Hove (Administrator). They join Project Lead Professor Faisal Ahmed and Project Manager Liz Dougan.

Upcoming Events

- **DiGeorge Syndrome Family Day**

Saturday 30th September 2017
Royal Hospital for Children

A programme of the day includes talks from healthcare professionals in the morning and informal workshops in the afternoon.

- **Rare Disease UK Children and Young People Event**

Thursday 19th October 2017
Royal Hospital for Children

Rare Disease UK are coordinating a project that aims to find out more about children and young people's experiences of living with a rare, genetic or undiagnosed condition. Families with children (aged between 5-10) affected by rare conditions, including siblings, are invited to take part in a range of flexible and fun activities to understand their experience of living with a rare, genetic or undiagnosed condition.

- **Von Hippel-Lindau Syndrome Patient Support Group Day**

4th November 2017
Royal Hospital for Children

Connect with other patients and families and hear talks from healthcare professionals on the condition and its management.

Please get in touch if you would like to find out more about any of our upcoming events.

Past Events

- **Early Management of Rare Conditions in the Neonate**

129 people attended this free study day on the 4th September 2017 aimed at all healthcare practitioners caring for newborn babies including doctors, nurses, midwives and other allied health care professionals.

- **"Muscle-Bone Deficits In Chronic Neuromuscular Conditions" symposium**

92 delegates convened in Glasgow on the 15th June 2017 at this symposium which brought specialists and scientists from neuromuscular and bone backgrounds together to discuss recent understandings of bone health in Duchenne Muscular Dystrophy and the path forward.

Scottish Government Cross-Party Group on Rare, Genetic and Undiagnosed Conditions

Liz Dougan represents the Office for Rare Conditions at the recently re-established Cross Party Group on Rare Genetic and Undiagnosed Conditions. The group (consisting of Members of Scottish Parliament, outside organisations and members of the public) meets three times a year with the purpose of looking at the challenges which cross cut the large number of rare, genetic and undiagnosed conditions. At the last meeting on 5th September the group discussed the recent review of Access to New Medicines and Care coordination and Specialist Nursing for rare conditions.

The next meeting will take place at the Scottish Parliament on the 5th December 2017.

Patient Advisory Group

The Office for Rare Conditions Patient Advisory Group brings together patients, families and carers to connect and integrate them into the Office and the decisions that influence its work.
Led by Arlene Smyth, the group meets regularly to discuss patient and personal experiences of living with a rare condition. If you would like to discuss patient-related concerns or share your experience, please get in touch.

Research News

The Queen Elizabeth University Hospital and Royal Hospital for Children in Glasgow are approved Health Care Providers in 3 European Reference Networks (or 'ERNs') for rare conditions: Endo–ERN, BOND and EpiCARE. The ERNs aim to improve clinical care for people with rare conditions across Europe as well as foster international research. These activities are supported by the Office for Rare Conditions.

The Office also co-ordinates 2 international registries (I-DSD and I-CAH) which support international research studies in these rare conditions affecting sex development.

Rare Condition Spotlight

Chromosome 22q11 Deletion Syndrome - a not so rare, rare condition.

Chromosome 22q11 Deletion Syndrome (a term which covers DiGeorge Syndrome) is the commonest chromosome deletion syndrome affecting approximately 1 in 3000 new-borns each year. Each person with Chromosome 22q11 Deletion Syndrome is unique as symptoms are variable in type and severity, depending on what organ system is affected. The condition can cause a range of lifelong problems including heart defects, frequent infections, delayed growth and development and cleft palate.

If suspected, diagnosis is often made by a specific blood test. Treatment usually requires a number of specialists from different fields of medicine and a multidisciplinary community team.

Max Appeal is the main UK patient support organisation. Their website has a wide range of excellent resources for patients, parents, health and education professionals (www.maxappeal.org.uk).

Thank you!

Michelle, Colin and Elsie How raised £500 for the Office for Rare Conditions at a race night on Saturday 9th September in Cambuslang. Thank you very much! This money will be used towards helping children and families with rare conditions.

Michelle, Colin and Elsie How, together with Dr Andreas Brunklaus from the Children's Hospital and Liz Dougan from the Office for Rare Conditions.

Twitter Fact

Wyburn-Mason Syndrome affects affects approximately 1 in 70 million people. It is a non-hereditary condition that affects the vascular system, causing arteriovenous malformation (AVM) of the eye and brain.

The Office for Rare Conditions is on Facebook and Twitter! Follow us and like our page to keep up-to-date with the latest information regarding Rare Conditions, support groups, meetings, events, and much more.

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Facebook.com/ORCGlasgow

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