Office for Rare Conditions

Newsletter

June 2018

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

Turner Syndrome Family Information Day

Saturday 9th June 2018
11am-4pm, Royal Hospital for Children

Health care staff will deliver informal talks followed by a Q & A session. There will be practical information on education, podiatry, employment and much more. Register here.

Hypospadias Information Day

Saturday 16th June 2018
11am-3pm, Royal Hospital for Children

This day is for parents of children with Hypospadias under 4 years of age. The programme includes advice on clinical care of boys with this condition. There will be an opportunity to talk to other families as well as healthcare staff. Register here.

Rare Condition Family Fun Event

Saturday 11th August 2018
11am-2pm, Touchbase
Sense Scotland, Kinning Park

The Family Fun Event we had planned for Rare Disease Day 2018 couldn't take place due to the #BeastFromTheEast, but we are rescheduling this event for Summer 2018! Register here.

Tuberous Sclerosis Association Annual Scottish Meeting

Saturday 25th August 2018
Royal Hospital for Children

The Tuberous Sclerosis Association Annual Scottish Meeting will include talks from healthcare staff and information on support available.

'Rare Conditions in the Neonate' Study Day

Monday 3rd September 2018
9-4.30pm, Teaching & Learning Centre

This study day, which was a great success last year, is aimed at all healthcare staff caring for newborn babies including doctors, nurses, midwives and allied health care professionals. Register here.

Achondroplasia Family Information Day

Saturday 3rd November 2018
11am-3pm, Royal Hospital for Children

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

Stickler Syndrome Information Day

Saturday 10th November 2018
11am-4pm, Royal Hospital for Children

This day aimed at people and families with this condition will include talks from healthcare staff in the morning and informal workshops in the afternoon. Register here.

Patient Advisory Group: Update

It was nice to meet some of the new members at our recent PAG meeting on 3rd May 2018 - thank you to everyone who came along or joined us via conference call. We had an interesting meeting, with lots of discussions and feedback from the Office for Rare Conditions Steering Committee meeting. Topics discussed included the essential information required when families with rare conditions first present to a new clinical service or when faced with an emergency. We also heard from Natalie Frankish, from Genetic Alliance UK, about the development of the Fresh Steps Toolkit, a new initiative that will help families navigate their way through the various support services. We also agreed to create a Facebook friendship group, which anyone can join. This Facebook group will help inform the PAG of patient and families' experiences of living with a rare condition and the care received when attending hospitals in Glasgow. Please do consider joining – you can do so here. We are delighted with the response to our PAG and thank everyone who has joined us and helps influence our work.

Arlene Smyth
Chair of the Patient Advisory Group

Research News

The 236th European Neuromuscular Centre (ENMC) workshop, led by Dr Jarod Wong, was held from 1-3 June 2018 in the Netherlands to discuss osteoporosis in people with Duchenne Muscular Dystrophy (DMD). A total of 21 clinicians and scientists convened to discuss the development of clinical trials of osteoporosis in DMD. The workshop, which is also supported by Action Duchenne, Duchenne Parent Project and Parent Project Muscular Dystrophy, further aims to set up an international consortium to develop research in the area of metabolic consequences in neuromuscular conditions.

E-Reporting of Rare Conditions across Europe

The Office is providing support for the EuRRECa project (European Registries for Rare Endocrine Conditions) which aims to create a core registry for rare endocrine conditions covered by Endo-ERN (www.eurreca.net). The project is one of five to receive funding from the European Union's Health Programme. In March 2018 the project launched a pilot e-reporting programme (e-REC) which will monitor new presentations of rare endocrine conditions on a monthly basis for the duration of the project. After the pilot phase, e-REC will be rolled out to all 71 Endo-ERN reference centres and other interested health care providers across Europe.

Neonatal Working Group

The Office for Rare Conditions Neonatal Working Group aims to identify and evaluate care pathways which currently exist for neonates with rare conditions attending the hospitals in Glasgow. The Office will collate and evaluate these pathways and provide access through its website.

Care pathways are tools which can guide healthcare professionals in managing, monitoring and recording a child's care and these clinical pathways are especially important for children with rare conditions. These children may regularly meet clinicians who have not previously encountered their specific rare condition. A good care pathway should describe the patient journey, contain information about the key elements of care based on a solid evidence base, contain information about the roles within the multidisciplinary team and should also identify appropriate resources.

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

The last cross party group meeting was held on 17th April. This meeting was the group's AGM and featured an overview of the CPG's work to date and an update on the campaign for a specialist nursing service for rare, genetic and undiagnosed conditions. On the 19th June 2018, the CPG will meet to celebrate Scotland's year of young people with an 'end of term' celebration. Visit the dedicated Scottish Parliament webpage for more information about the group.

Are you a patient with a rare condition who attends a hospital in Glasgow? Care for someone who is? Take part in this survey and help direct priorities in the future work of the Office for Rare Conditions, Glasgow.

Spotlight on: Scottish Genomes Partnership

The Scottish Genomes Partnership (SGP) is using whole genome sequencing, health information and data analysis to try to understand what causes rare genetic conditions. More than 1600 samples have been provided and analysed for SGP by Edinburgh Genomics.

Studies currently underway are investigating the genetic causes of eye malformation, microcephaly, extreme short stature, disorders of sexual development and motor neurone disease.

Working with the NHS, SGP is recruiting 1,000 participants (individuals diagnosed with rare conditions and their close family members) from across Scotland for the Genomics England 100,000 Genomes Project.

Recruitment to the study is almost complete, with genome data already produced for almost 500 people. By the end of May 2018, 326 people who attend clinical services in Glasgow had consented into the study.

The first clinical results are eagerly anticipated in Scotland. It is hoped that these studies will pave the way for more diagnoses and better lives for patients with rare genetic conditions. Visit the SGP website for more info.

The Office for Rare Conditions is on Facebook and Twitter – follow us for the latest updates!

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