Office for Rare Conditions

Newsletter

September 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

Wee Scottish Livers Family Day

Saturday 29th September 2018
10am-4pm, Royal Hospital for Children

This family education day will give families and liver teams the chance to share ideas and support service development to improve the care of children with liver conditions in Scotland. Register here or contact www.glasgowchildrenshospitalcharity.org

Undiagnosed & Rare Genetic Conditions Information Day

Tuesday 2nd October 2018
10am-2pm, Royal Hospital for Children

We are supporting SWAN UK's free information event for families that will cover genetic testing, the 100,000 Genomes Project, and what life is like raising a child with a rare or undiagnosed condition. Email SWAN to register or phone them on 020 7831 0883.

Achondroplasia Family Day

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

Connect with other patients and families and hear talks from healthcare staff on this condition and its management. For more information e-mail info@officeforrareconditions.org.

Rare Condition Family Fun Event

On 11th August 2018, we held our first Family Fun Event for families with rare and undiagnosed conditions at TouchBase. We were delighted with the response to the event which was sold out and had a great day filled with fun, games and dancing, as well as Taekwondo and Gymnastics taster sessions, face painting, craft workshops and lots more! Thanks to all the families, entertainers and organisations who made the day such a success!

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.

Rare Resources Project

Genetic Alliance is looking for 100 families in Scotland to join the Rare Resources project. This project has developed a toolkit of information resources for families in Scotland who have received a diagnosis of a rare, genetic condition or who have an undiagnosed condition. To find out more about the project and how to join, click here or search rare resources toolkit at www.geneticalliance.org.uk

Fundraising News

Many thanks to Dr Martina Rodie who ran the Great Women's 10k in June and raised £1110 for the Office for Rare Conditions! If you would like to find out in what way you could help to raise money for the Office, please get in touch or e-mail: info@officeforrareconditions.org

Neonatal Working Group

The Office for Rare Conditions Neonatal Working Group aims to identify and evaluate current care pathways for neonates with rare conditions who attend hospitals in Glasgow. The working group has developed a tool and a process for care pathway appraisal which is currently being piloted. 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

We recently held our 3rd PAG meeting on 7th August 2018 – thank you to everyone who came along. It was an interesting and informative meeting; Prof Faisal Ahmed shared some of the background of the Office for Rare Conditions and its aims and objectives, emphasising the importance of the Patient Advisory Group in making sure that the voices of patients and families are heard and help direct the work of the Office. We also reviewed a draft version of a Rare Condition Care Summary which the Office continues to develop and had some interesting discussions regarding various health apps as well as updates from rare condition community and patient groups. We now have an Office for Rare Conditions Facebook Friendship Group aimed at families with rare or undiagnosed conditions who attend a Glasgow hospital.
Do consider joining this group where we share and exchange information and offer support for those difficult days.
(www.facebook.com/ORCGlasgow search 'friendship group')

Arlene Smyth
Chair of the Patient Advisory Group

Clinical Patient Management System

CPMS is a secure web-based platform to support European Reference Networks (ERNs) in the diagnosis and treatment of rare conditions across national borders; bringing expert specialised care to all patients in Europe.

Any doctor can ask for virtual consultation of ERN reference centres. The platform enables a multidisciplinary team (MDT) to discuss highly specialised patient cases in a secure closed-off environment. Their own treating physician can then directly refer the resulting advice back to the patients.

The Office for Rare Conditions can provide support for any health care professional interested in using the CPMS platform.

Cheerio!

In this edition, we sadly say goodbye to Hannah Van Hove who is leaving us to conduct postdoctoral research in Belgium. Thanks to Hannah for all her hard work helping us establish the Office for Rare Conditions, Glasgow. We will miss you!

Spotlight on: Undiagnosed Conditions

Approximately 6,000 children are born in the UK each year with a syndrome without a name – a genetic condition so rare that it is likely to remain undiagnosed.

Some children affected by a syndrome without a name might be described as having global developmental delay or failure to thrive. They might have learning disabilities and/or physical disabilities. They can sometimes have complex medical needs and epilepsy. Some children may not have any learning disabilities but be physically disabled whilst others are physically fine but have learning disabilities.

SWAN UK (syndromes without a name) is the only dedicated support network for families of children and young adults (0-25) with undiagnosed genetic conditions. It is run by the charity Genetic Alliance UK. SWAN UK enables families to:

- Make contact with other families who understand the unique challenges of raising a child affected by an undiagnosed genetic condition
- Attend free, fun whole-family events and regional day trips
- Access 24/7 information and support through our online forums.

Find out how to join SWAN here or www.undiagnosed.org.uk/join

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

@ORCGlasgow
Facebook.com/ORCGlasgow

OFFICE FOR RARE CONDITIONS
Royal Hospital for Children
Zone 1 Office Block, 1345 Govan Rd
G51 4TF, Glasgow, Scotland, UK
Tel: 0141 451 5899
info@officeforrareconditions.org
www.officeforrareconditions.org