Office for Rare Conditions

Newsletter

February 2018

The Office celebrates its 1st birthday!

The Office for Rare Conditions in Glasgow has had a very busy and productive first year. In Glasgow, the Office organised several education events for patients, parents and professionals, patient surveys requested by clinical teams and developed a local patient advisory group. I am extremely grateful to Arlene Smyth from the Turner Syndrome Support Group for chairing this group and for representing the interests of patients and parents on the Office's Steering Committee.

At an international level, the Office has been heavily involved in supporting Glasgow's activities in the European Reference Networks for rare conditions. I am also pleased to report that the Office was successful in securing EU funding to lead on a new project that will see the development of a core registry that will run from Glasgow and link over 71 expert centres across Europe. I would like to thank the Reid-Timoney Foundation, the Glasgow Children's Hospital Charity, the University of Glasgow, Greater Glasgow & Clyde NHS Health Board and Kyowa Kirin for their continuing support to this initiative.

Professor Faisal Ahmed,
Project Lead

Rare Disease Day 2018

Every year, the last day in February is global Rare Disease Day, a day in which people and organisations all over the world draw attention to rare conditions and their impact on patients' lives. The Office is organizing a number of events to mark Rare Disease Day 2018 – join us and help raise awareness of rare conditions!

Upcoming Events

Rare Disease Day Family Fun Event

Wednesday 28th February 2018
4-7pm, Partick Burgh Hall

Come and join us on Rare Disease Day and help raise awareness of rare conditions! There will be lots of fun activities for children and a chance to meet with other families and children living with rare conditions. All ages welcome! Register here.

Rare Condition Information Day

Saturday 3rd March 2018
11am-4pm, Royal Hospital for Children

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. Register here.

To raise awareness of rare conditions, the Office will also be speaking at professional events at the Queen Elizabeth University Hospital and the Royal Hospital for Children during Rare Disease Day week.

Stickler Syndrome (Family) Day

Saturday 17th March 2018
11am-4pm, Royal Hospital for Children

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

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

Patient Advisory Group

The Office for Rare Conditions Patient Advisory Group [PAG] has been set up and two meetings have taken place so far to discuss experiences of care in Glasgow. Arlene Smyth (chair), states: 'It has been inspiring to meet the amazing parents who have come along to the meetings and shared their views and experiences. A report of these meetings will be given to the steering group for consideration. I would like to say a big thank you to all who have given time and energy to join PAG and I look forward to continue working with everyone to enhance the quality of care provided to patients with rare conditions.'

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

Fiona McQueen, Scotland's Chief Nursing Officer, attended the most recent meeting of the Cross-Party Group on 5th December to discuss the recently published report on the Specialist Nursing and Care Fund. Points agreed on included: the value of specialist nurses for support and information, the need for succession planning in established specialist nurse posts and improved communication between regional health boards. The next meeting will take place on 20th March 2018

On Tuesday 20th February 2018, Professor Ahmed will be speaking at the annual Rare Disease Day reception at the Scottish Parliament Building, organised by Rare Disease UK. The event will feature an update on progress being made to implement the Scottish Rare Disease Plan. Other speakers include: Bob Doris MSP, Convener of the Cross Party Parliamentary Group on Rare, Genetic and Undiagnosed Conditions and Dr Jayne Spink, Chair of Rare Disease UK.

Projects Beyond Glasgow

The Office for Rare Conditions supports a number of international initiatives: QEUH and RHC are approved Health Care Providers in 3 European Reference Networks for rare conditions: Endo–ERN (endocrine), BOND (bone) and EpiCARE (neurology). From February the Office will be co-ordinating a new European project, EuRRECa, which will develop a core registry for all rare endocrine conditions in Endo-ERN. The Office also co-ordinates 2 international registries (I-DSD and I-CAH) which support research in rare conditions affecting sex development. The Office also coordinates UK-wide audit projects such as the BSPED audit of GH use.

What are European Reference Networks (ERNs)?

No centre alone has the knowledge and capacity to treat all rare and complex conditions, but by cooperating and exchanging life-saving knowledge at European level through ERNs, patients across the EU will have access to the best expertise available. European Reference Networks are virtual networks involving Reference Centers across Europe. They aim to tackle complex or rare conditions that require highly specialised treatment and concentrated knowledge and resources. The first ERNs were launched in March 2017, involving more than 900 highly-specialised healthcare units from over 300 hospitals in 26 Member States. 24 ERNs are working on a range of thematic issues.

The Reid-Timoney Foundation

University of Glasgow NHS Greater Glasgow and Clyde Glasgow Children's Hospital Charity KYOWA KIRIN

Share Your Views!

Are you a patient with a rare condition? 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.

Are you a healthcare professional? Take part in this survey by Health Improvement Scotland on information resources for the diagnosis and management of rare conditions.

Spotlight on Stickler Syndrome

Stickler Syndrome is a group of genetic conditions that affect collagen, a protein in the body that is important in many structures. Possible problems include:

- **Eyes:** Early onset short sightedness, retinal detachment, cataracts, glaucoma.
- **Mouth/Face:** cleft palate, small lower jaw, flat face with small nose or flat nasal bridge.
- **Joints & Bones**: hypermobile joints which may later become stiff, osteoarthritis in later life.
- **Hearing**: hearing loss, glue ear.
- **Other**: spine and heart problems.

Approximately **1 in 7,500** newborn babies can be affected. Diagnosis can be difficult as the symptoms and severity of this group of conditions may vary from one person to another. Some people have no signs or symptoms whilst others have all the above features. When suspected, the diagnosis can confirmed by a genetic test. For more information and support contact Stickler Syndrome UK.

@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
Email:
info@officeforrareconditions.org