


# Office for Rare Conditions Summer Newsletter 2024


The Office for Rare Conditions (OfRC)Glasgow, based at the Royal Hospital for Children and the Queen Elizabeth University Hospital, aims to raise awareness of rare, low prevalence and undiagnosed conditions, enhance the quality of care provided and promote participation in research.

It is funded through Glasgow Children's Hospital Charity.

Summer 2024

Office for Rare Conditions, Glasgow

## In This Issue

- **What's Happening in Rare Conditions**
- **Coming Up**
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## *Rare Disease Day 2024*


*1 - Left to right: Clinical Lead Office for Rare Conditions & Consultant Neonatologist **Dr Martina Rodie,** Genetic Alliance Head of Policy and Engagement Manager, Scotland, **Natalie Frankish**, Scottish Government Deputy Chief Medical Officer **Professor Marion Bain.***

This year ***Rare Disease Day*** fell on the rarest of days, the **29th** day in February , which happens only once every 4 years, a rare day indeed!

The Office for Rare Conditions organised a week long stand in the Royal Hospital for Children to reach out to as many patients and families as possible. Each day we were joined by a different patient support organisation and on February 29th we were delighted to be visited by the **Scottish Government's Deputy Chief Medical Officer, Professor Marion Bain**.

Professor Bain took time to meet with the team, including Arlene Smyth, Chairperson of the OfRC Patient Advisory Group, to discuss current issues and challenges faced by patients and families affected by rare conditions.

“Thank you to the teams for hosting me today and explaining in detail the necessity of the important work that they do. I commend their work in improving the management of patients and families with rare conditions in addition to help drive research, with their partners, to help manage these conditions. We look forward to seeing what’s next as they continue to help Glasgow and Greater Clyde patients,”

*2 - The new artwork in theatre reception. Up Down Around the Town by Laura Darling & Mark Faulkner*

On Rare Disease Day we also celebrated the new reception artwork which is part of the redesign of the Royal Hospital for Children's theatre. Work to ensure the theatres are more empathic, inclusive and welcoming for children who have to visit for surgery has been led by Dr Aly Walker, consultant anaesthetist. Dr Walker asked the illustrator to include a representation of people with rare conditions and, as the zebra has been adopted as a rare disease mascot, it was delightful to see a few zebras going about their business in the busy urban scene. You will often see the Office for Rare Conditions' zebra mascot when you visit our stand. This year we ran a competition to name the Zebra and his chosen name is Jax!

Thank you to all the families that visited to share their thoughts and experiences, helping to bring together the rare condition community.

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*A massive **Thank You** to everyone who took part in supporting **Rare Disease Day 2024!***

**Patients, Families/Carers Health Care Professionals, Genetic Alliance UK, Family Fund, Contact, PIP UK, TSSS UK, Glasgow Life Sport & Physical Activity**

*and to **Glasgow Children's Hospital Charity** for their continuous help and support. We couldn't have done it without them!*

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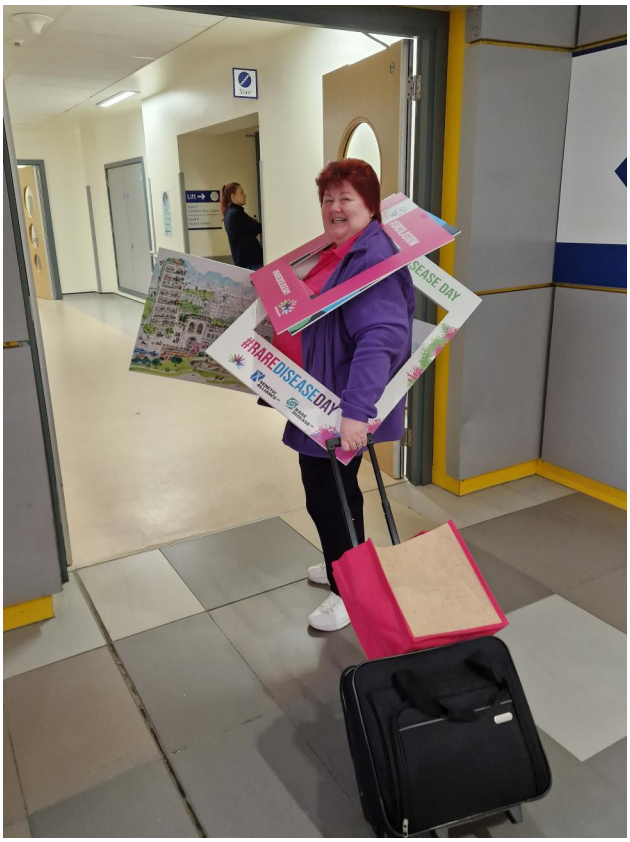
I SUPPORT
#RAREDISEASEDAY
#ORCGLASGOW
RAREDISEASEDAY.ORG


RARE
DISEASE DAY
Atrium

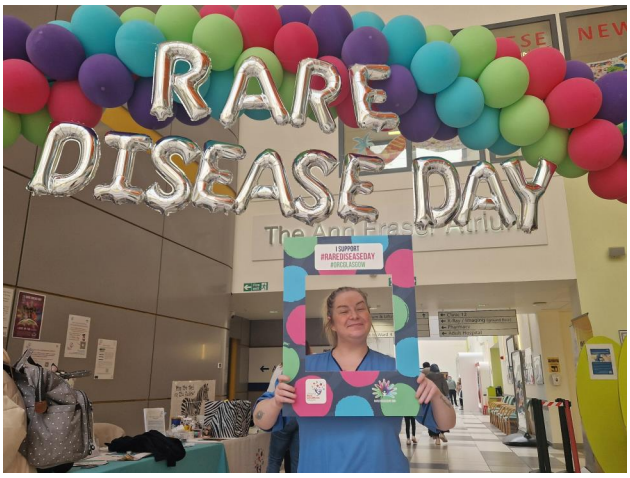

#RAREDISEASEDAY


RARE
DISEASE DAY
The Ann Fraser Atrium
Outpatient
Reception
Adult Hospital


RARE
I SUPPORT
#RAREDISEASEDAY


Atrium
Fire exit
I SUPPORT
#RAREDISEASEDAY
#ORCGLASGOW
NHS


#RAREDISEASEDAY

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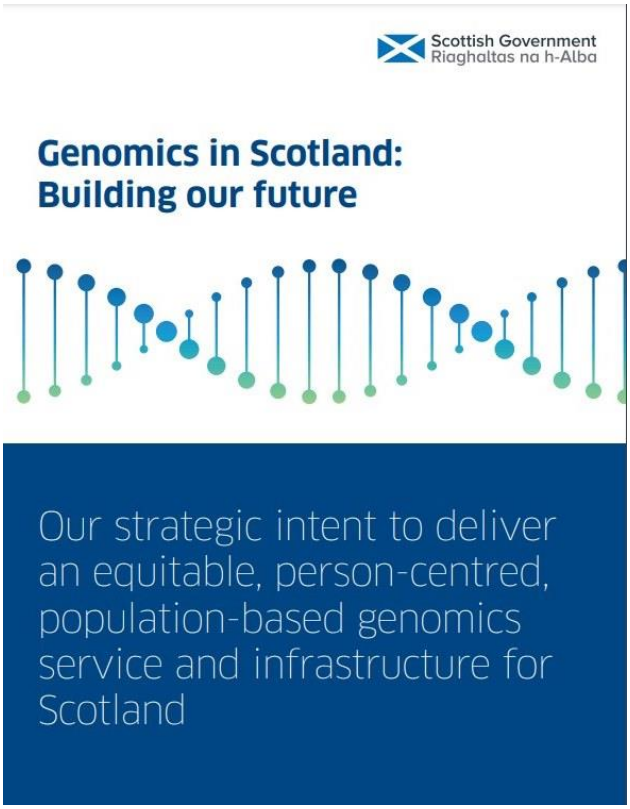
## *What's happening in rare conditions?*

## Genomics in Scotland: Building our Future




Genomic medicine is the use of genetic information (the instructions within our body's cells that shape a person's health, growth and development) to diagnose disease, guide the use of

different treatments or predict the risk of disease.  The Scottish Government is working to establish a genomic medicine service that improves the lives of people in Scotland. The Scottish Strategic Network for Genomic Medicine (SSNGM) will facilitate clinical and other improvements in genomic medicine through delivery of objectives set by the National Specialist Services Committee (NSSC) for NHS Boards and Scottish Government Health & Social Care Directorate (SGHSCD) within the national commissioning process.

The Office For Rare Conditions is an active participant in the SSNGM's  Patient Involvement Advisory Group (PIAG). This group will work to ensure that there are opportunities for patients, families, service users and the wider public to feed into the activities of the network. Their role is to help identify areas of work that need patient and public input and engagement.

You can find more information on the work of the SSNGM here https://www.gov.scot/publications/genomics-scotland-building-future/documents/.

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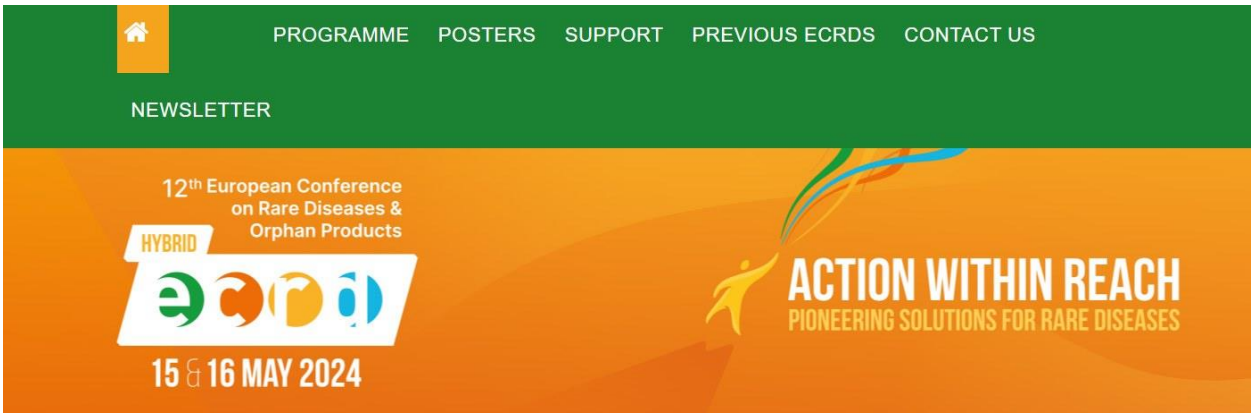
## European Conference on Rare Disease




The European Conference on Rare Disease is the **largest, patient-led, rare disease policy-shaping event** held in Europe.  This year over 700 people attended  with the OfRCs' Dr Martina Rodie and Claire Dinning presenting 2 posters:

***Patient Navigator for The Office for Rare Conditions, Glasgow :A progress update*** Claire Dinning

***Evolving Experience of health care at a reference centre as reported by patients and parents with rare conditions over a 6 year period*** Dr Martina Rodie

Dr Rodie's poster was among the highest scoring at the conference.  Look out for its upcoming publication in the Orphanet Journal of Rare Diseases.

# Patient Navigator for The Office for Rare Conditions, Glasgow: A progress update.

**C Dinning, E Dougan, S Mullen, A Kelly, SF Ahmed, Rodie M**
The Office For Rare Conditions, Royal Hospital for Children, University of Glasgow, UK

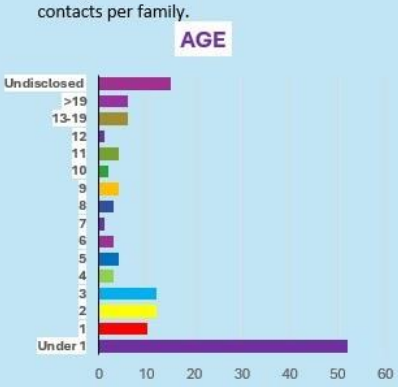
## Introduction

The Patient Navigator role was established in 2023 and is the first of its kind in Scotland. The aims are to raise awareness, offer support and signposting, improve care coordination, provide education and offer a holistic approach to the care of those affected with rare, low prevalence and undiagnosed conditions.

## Results

Contact Measurements from January 2023 to January 2024 Patient Navigator
Families 140
Interactions 369

The method of contact depended on family needs, some were face to face, some by email and phone. The number of times each family had contact ranged from 1 – 14, with an average of 2.5 contacts per family.


## Conditions

98 rare conditions were encountered with the range of incidence from 1 in 2000 to 23 reported cases world-wide.

| | | | |
|---|---|---|---|
| Biotinidase Deficiency | 1: 137,000 | Langerhans Cell Histiocytosis | 4-9 :1,000,000 |
| Cerebro-Costo-Mandibular Syndrome | <1 : 1,000,000 | Nephronophthisis, NPHP3 gene | |
| Chondrodysplasia Punctata Type 2 | 1:400,000 | Noonan syndrome | 1: 1000-2,500 |
| Coffin-Siris Syndrome | <200 worldwide | Oesophageal Atresia | 1:5,000 |
| Congenital Adrenal Hyperplasia | 1:10,000-15,000 | Phenylketonuria (PKU) | 1:15,000 |
| Cornelia De Lange Syndrome (C.D.L.S) | 1:10,000-30,000 | PIK 3cA | |
| Cutaneous Polyarteritis Nodosa Vasculitis | 91: 1,000,000 | Pierre Robin Syndrome | 1: 8,500- 14,000 |
| Cri Du Chat Syndrome | 1:50,000 | Poland Syndrome | 1:20,000 |
| Diaphragmatic Hernia | <5 in 10,000 | Prader Willi Syndrome | 1:10.000 to 30,000 |
| Duchenne Muscular Dystrophy | 1:3,500 | POLR3B | |
| Dyslibrinogenemia | 1 in 100,000 | Primordial Dwarfism | 1:25,000 |
| Ehlers Danlos | 1 in 2,500-5,000 | Spina bifida with Hydrocephalus | 1:7,500-8,000 |
| Exomphalos Major | 2:5,000 | Townes Brocks | 1:9 in 1,000,000 |
| Goldenhar | 1:3,000-5,000 | Trachea-Oesophageal fistula | 1 : 3,500 |
| Gnclo 1 Gene | | 16q12.1 - q21 Chromosome Deletion | 1: 2000 |
| Hirschsprungs | 1:5,000 | Hirschsprung's | 1:5,000 |
| Joubert Syndrome | 1:80,000-100,000 | VACTERL Association | 1 :10,000 – 40,000 |
| Kagami-Ogata Syndrome | 1:1,000,000 | Waardenburgh Shah Syndrome | 1:40,000 |

**Problems managed** included end of life care, obtaining a second opinion, assistance with grant applications, support group information, links to healthcare staff and psychological support and links to other families

**Resources** developed included a holistic needs assessment form, contact log of health care professionals, database for resources for information for families on finance, housing, activities, mental health and wellbeing, refugees, sibling support, treatments, help with healthcare costs and charity contacts.

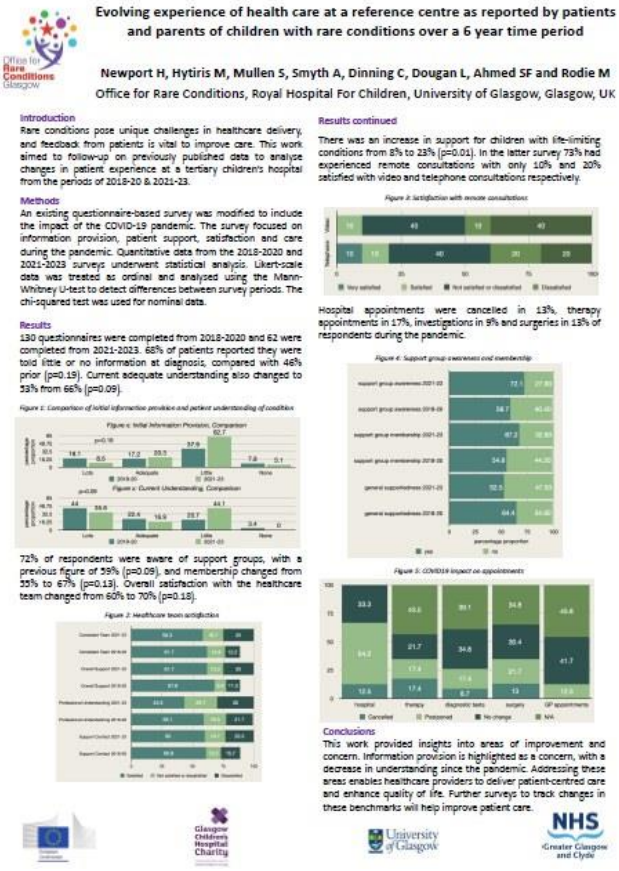



Do you know who to contact if you need further information/support? Yes 100%

## Summary

This novel role has proved a busy and effective one with excellent feedback. The support offered was diverse and families often contacted the Navigator feeling lost and helpless. This role should be considered by other centers caring for those affected by rare conditions.

*3 - E- poster by Patient Navigator Claire Dinning*




*4 - E-poster by Dr Martina Rodie*

The conference closed with the signing of an open letter calling on the next European leaders to:

- Maintain health at the forefront of future policies and programmes.
- Develop a comprehensive European Action Plan for Rare Diseases that bridges diverse policy areas and streamlines existing efforts with clear, measurable objectives.
- Immediately address the most pressing needs of the rare disease population by integrating the actions 'within reach' that have been collectively identified within the work programmes of the next years.

You can read the full Open Letter, and sign it, here.

## Working in Partnership

ENGAGEMENT

The Office for Rare Conditions continues to partner with many 3rd sector organisations, bringing information on available resources directly to patients and families through our information stands within the hospital and at community events.

Contact and Family Fund join our stand in the Royal Hospital for Children on a 2 monthly basis to reach out to patients and families providing invaluable support and information. If you know of an organization that could benefit patients and families and would be a great addition to our stand, please let us know.


*5 - Family Fund, Contact &OfRC*

*6 - Contact & Tumour Support Scotland*


*7 - Neonatal Event*

*8 - Family Information Stand*


*9 - Sickle Cell Awareness day*


*10 - The Hope project*

**Raising Awareness**

To help raise awareness and enhance understanding of rare conditions, the OfRC is collaborating with an increasing number of patient support organisations on their designated awareness days. We provide information on the condition and its management, linking with clinical services whenever possible.  Are you aware of an upcoming awareness day for a rare condition? The OfRC is here to help you raise awareness and support. Get in touch with us for more information.

e-mail info@officeforrareconditions.org


*11 - Turner's Syndrome*


SWAN UK
syndromes without a name
WE SUPPORT
UNDIAGNOSED
CHILDREN'S DAY
geneticalliance.org.uk/swanuk
26 April 2024

Office for Rare Conditions Glasgow

# The Office for Rare Conditions supports

# World Sickle Cell Day

with

## The Hope Project, Scotland

at the Royal Hospital for Children Glasgow

## 19th June 2024

**Sickle cell disease is the name for a group of inherited health conditions that affect the red blood cells. The most serious type is called sickle cell anaemia.**

**Sickle cell disease is particularly common in people with an African or Caribbean family background**

**Sickle cell disease is a serious and lifelong health condition, although treatment can help manage many of the symptoms.**

**Drop by our stand to find out more about sickle cell disease and how the Hope Project and healthcare services can help**

**Find out more here**

**Scottish Paediatric and Adult Haemoglobinopathies Network**

**NHS Inform**

Poland Syndrome Support & Network PIP - UK
SCOTLAND Branch

Office for Rare Conditions Glasgow

# Poland Syndrome Awareness Day
# 30th April 2024

## Join Us at the Royal Hospital for Children!

🌟

*Are you curious about Poland Syndrome and eager to connect with those who understand its challenges firsthand?*

**We invite you to drop by our stand and meet the incredible individuals from PIP UK patient support organization, alongside those with lived experiences of Poland Syndrome.**

📅

**Date: 30th April 2024**

🕒 **Time: 10am - 3pm**

📍 **Location: Atrium, Royal Hospital for Children Glasgow**

# Neurofibromatosis Awareness Day

## Tumour Support Scotland

### will be in the Royal Hospital for Children on 17th May 2024 🌟

**Neurofibromatosis Type 1 (Nf1) is one of the most common but least known genetic conditions, affecting 1 in 3,000 people**
**Tumour Support Scotland supports children and families living with Neurofibromatosis in Scotland**

**We invite you to drop by our stand to find out more about Neurofibromatosis and what information and support Tumour Support Scotland can offer.**

📅

**Date: 17th May 2024**
**🕒 Time: 10am - 3pm**
**📍 Location: Atrium, Royal Hospital for Children Glasgow**

contact *For families with disabled children*

Office for Rare Conditions Glasgow


## Right Decisions for Health and Care


Delivered by Health Improvement Scotland, the **Right Decision Service** is a 'Once for Scotland' source of digital tools that enable people to make safe decisions quickly 'on the go', based on validated evidence. It provides health and social care organisations with tools to build decision-ready guidance, pathways, risk scoring tools, shared decision aids and other decision support resources. These tools are all delivered through this Right Decision Service website and mobile app.

Check it out here [Right Decisions | Right Decisions (scot.nhs.uk)]

## Information & Signposting


To keep everyone updated on new resources, helpful organizations, and the latest guidance, the OfRC releases a quarterly bulletin on its website.

Take a look here.

We would be thrilled to feature your recommended resources in our quarterly bulletin. Please send them to info@officeforrareconditions.org.

Have you explored our patient and parent-inspired resources on our website: www.officeforrareconditions.org?

Alternatively, click this link.

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Phone: 0141 451 5899
Email: info@officeforrareconditions.org
Social Media: @ORCGlasgow

Office for Rare Conditions Glasgow

# CARE SUMMARY

## ESSENTIAL INFORMATION

**Full Name** :
**CHI** :
**Contact person** :
**Diagnosis** :
**Comment:**

## SAFETY ALERT/ALLERGY

**Comment:**

## IMPORTANT INFORMATION ABOUT ME

**Comment:**

## MEDICATIONS AND MEDICAL DEVICES

**Medication names** :

**Direction-Dose and Duration:**

**Any Additional Instructions:**

TURNER SYNDROME TSSS SUPPORT SOCIETY [UK]

# Know your `OLOGISTS`

Office for Rare Conditions Glasgow

**As many of you attend a wide variety of different specialities, we thought it might be helpful to know exactly who the specialists are and what their responsibilities are. This is not an exhaustive list, but it covers the most commonly seen specialists.**

## ANAESTHETIST

Specialise in pain services and managing sedation, and consciousness during surgery

## AUDIOLOGIST

Specialise in hearing tests and hearing aid fitting

## CARDIOLOGIST

Specialise in heart problems (they are the ones who monitor you if you have a problem with your aorta or heart valves)

## DERMATOLOGIST

Specialise in skin, check moles

## OTOLARYNGOLOGIST

(Ear, Nose and Throat (ENT) surgeon) Specialise in ENT conditions that need care and or surgery such as grommets for glue ear, cochlear implants, removal of tonsils and adenoids

## ENDOCRINOLOGIST

Specialise in hormone and growth conditions

## GASTROENTEROLOGIST

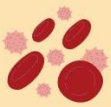
Specialise in digestive, stomach and bowel issues such as Crohn's, ulcerative colitis or coeliac, and may also see you for liver conditions

## GYNAECOLOGIST

Specialise in menstruation (period) and fertility and assisted fertility

## HAEMATOLOGIST

Specialise in blood and bone marrow conditions



## HEPATOLOGIST

Specialise in the liver, fatty liver disease

Glasgow Children's Hospital Charity
Continuing the care of Yorkhill Children's Charity

NHS Greater Glasgow and Clyde

# Have you or your child been diagnosed with a rare condition?

**Are you looking for further support?**
**or want to speak to someone who is going through the same experience?**

Read our welcome letter to the Office for Rare Conditions

*'A Warm Hug'*

by scanning the QR code

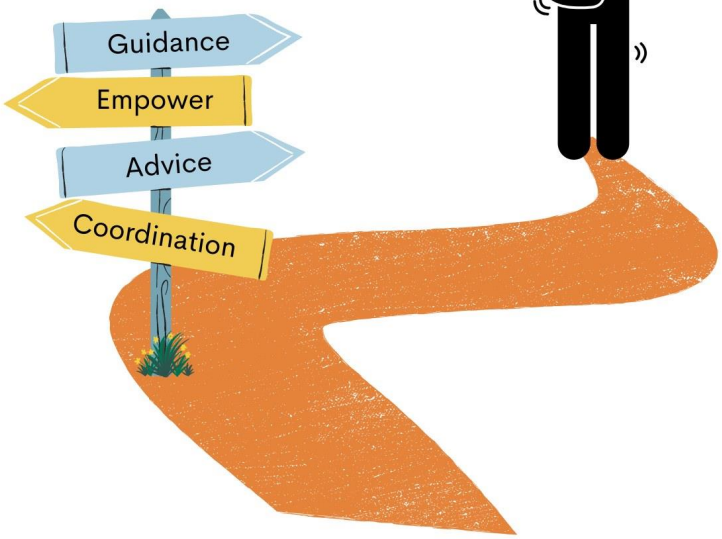
Office for Rare Conditions Glasgow

**Office for Rare Conditions**
**Phone: 0141 451 5899**
**Email: info@officeforrareconditions.org**
**Social media: (FB/Instagram/Twitter): @ORCGlasgow**

**Have you or your child had a diagnosis of a rare condition?**

**Are you still waiting on a diagnosis?**

**Are you looking for further support or someone to speak to?**

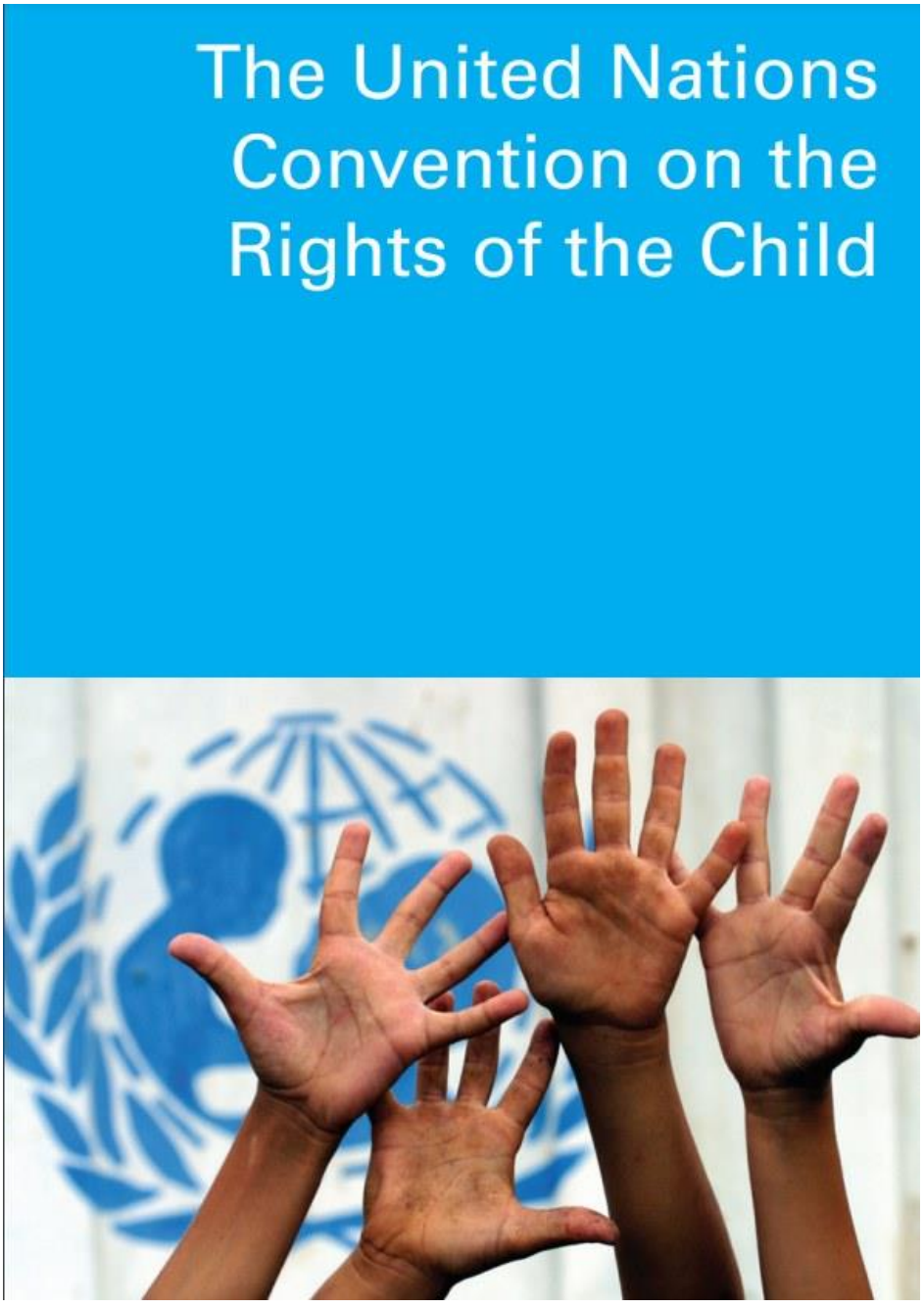

## Why not have a chat with our Patient Navigator, Claire Dinning, at the Office for Rare Conditions!

Claire's main role is to help facilitate the patient journey and improve the coordination of care by providing patients and families with information and support.

***If you feel that you or someone you know may benefit from having a chat with Claire then please get in touch with the Office for Rare Conditions: info@officeforrareconditions.org or 01414515899.***

Office for Rare Conditions Glasgow

## United Nations Convention on the Rights of the Child




On 16 January 2024, the **United Nations Convention on the Rights of the Child** Bill became an Act, receiving royal approval.

The 16 July 2024, was the day when most parts of this historic Act came into effect, highlighting Scotland's commitment to children's rights. Adults must respect and protect the human rights of children and young people when they plan services, make policies and decisions. This incredible milestone celebrates the tireless efforts of Child Health Scotland's Health Rights

Defenders. They have been advocating for years, showcasing Scotland's dedication to its future generations.

**Child Health Scotland** have now launched the My Health, My Rights Charter, crafted by children and young people living with health conditions in Scotland.

This charter presents their 10 most important health rights, listed without order because each one is equally important.

More information on the Act


## Patient Navigator for Rare & Undiagnosed Conditions

The OfRC **Patient Navigator, Claire Dinning,** has been supported by **Glasgow's Children's Charity** for another year. This has allowed Claire to continue supporting, guiding and sign posting families affected by rare and undiagnosed conditions. Claire has supported more than 180 families, providing holistic care.

If you know anyone that would benefit from our patient navigator service, please let us know by e-mailing the OfRC at Info@officeforrareconditions.org. Claire will respond to you directly.

## Patient Advisory Group by Arlene Smyth (Chair)


*"It was just wonderful to be in the hospital atrium for Rare Disease Day this year for a full week. We had a great response from families. We met so many new families with rare conditions. We did have a meeting with the deputy chief medical officer and we were able to explain the length of time a diagnosis can take and what a waste of hospital resources too. If only they would think of rare conditions first such as Turner Syndrome and many others.*

*I would just like to thank all the volunteers, too many to mention individually. Thank you to Liz, Martina and Claire for all their hard work all year round but especially over Rare Disease Day/week. Together we are stronger.*

*Our Patient Advisory Group (PAG) continues to meet on the 3rd Thursday, every two months at 7pm. We have covered a variety of subjects such as the hospital pharmacist, and 111 service and we are looking at repeating some of the talks on mental health and others that were popular. If you have any suggestions please do not hesitate to contact us with suggestions. Do check out the Facebook page for more details on hot topics. We share as much useful information as we can.*

*Our wonderful Patient Navigator Claire has really settled in and done a great job since joining the team. We really do hope we can continue to fund her very valuable work. Many families were saying how beneficial it was to meet Claire and see a friendly face who understands rare conditions.*

*Recently, we conducted a survey to gather members' views on the group's future direction, including meeting times, locations, and frequency. The majority expressed a preference for quarterly meetings, on different days of the week and with a mix of two online sessions and two*

*in-person gatherings that include activities for children. We are delighted to implement this. Additionally, members suggested recording our informational topic talks from each meeting and publishing them on our website for easy access. There is also interest in regular podcasts, with topics to be proposed by the group. We look forward to implementing the new changes.*

*Our next meeting will be in person Tuesday 30th July at Lynn Park Adventure playground. Registration is now open but there are only a few places left*

https://link.webropol.com/ep/summerfunevent2024

*Looking forward to seeing those of you who can join us at our summer family event on the 30th of July.*


*Don't forget to keep track of our social media for up-to-date information. I hope you have a great summer and look forward to seeing everyone on at our next meeting.*

*Kind regards Arlene*

## Patient Reported Experience Measure for Rare & Undiagnosed Conditions

**Help us if you can:** Do you know someone managing a rare condition that could tell us about their journey. Our **Patient Reported Experience Measure** helps us understand what it is like to live with a rare condition, what kind of support works well or not so well, and what services and support might be helpful. We are keen to identify gaps in the provision of services and areas of good practice.

Complete our PREM survey here

https://link.webropolsurveys.com/Participation/Public/780c6c6f-175d-459f-af8d-d4bf552732ba?displayId=Uni2726868

## *Coming Up!*

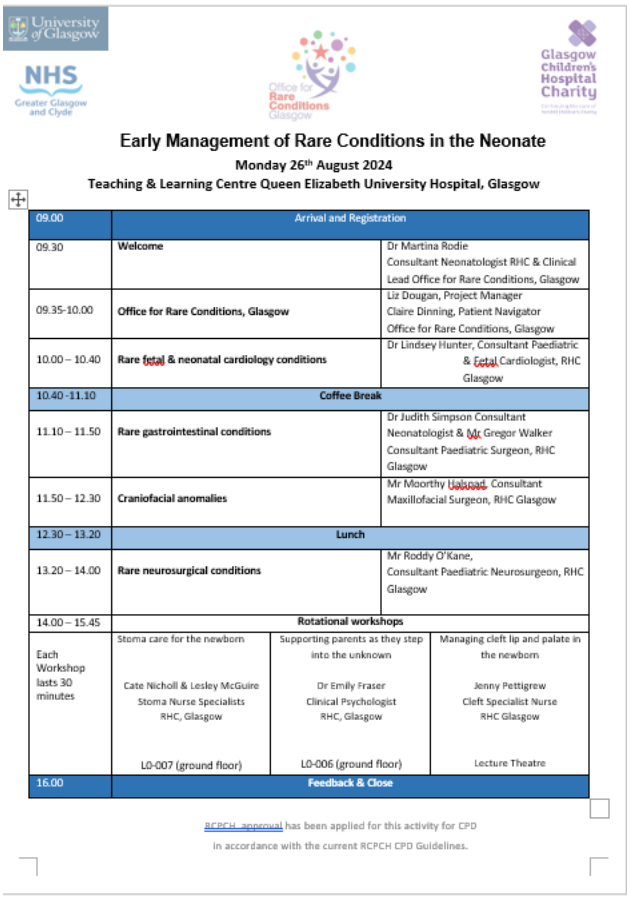
Registration is now open for our **8th Annual Neonatal Study Day** on the **management of rare conditions in the neonatal period**. We are delighted that this year there will be no charge to attend the event!

Talks will not be recorded, so why not come along in person?

**Register now to secure your place**

https://link.webropol.com/ep/earlymanagementofrareconditions2024


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



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

**Monday 26th August 2024**

**Teaching & Learning Centre Queen Elizabeth University Hospital, Glasgow**

| | | | |
|---|---|---|---|
| 09.00 | Arrival and Registration | | |
| 09.30 | **Welcome** | Dr Martina Rodie<br>Consultant Neonatologist RHC & Clinical Lead Office for Rare Conditions, Glasgow | |
| 09.35-10.00 | **Office for Rare Conditions, Glasgow** | Liz Dougan, Project Manager<br>Claire Dinning, Patient Navigator<br>Office for Rare Conditions, Glasgow | |
| 10.00 – 10.40 | **Rare fetal & neonatal cardiology conditions** | Dr Lindsey Hunter, Consultant Paediatric & Fetal Cardiologist, RHC Glasgow | |
| 10.40 -11.10 | **Coffee Break** | | |
| 11.10 – 11.50 | **Rare gastrointestinal conditions** | Dr Judith Simpson Consultant Neonatologist & Mr Gregor Walker Consultant Paediatric Surgeon, RHC Glasgow | |
| 11.50 – 12.30 | **Craniofacial anomalies** | Mr Moorthy Ilahnad, Consultant Maxillofacial Surgeon, RHC Glasgow | |
| 12.30 – 13.20 | **Lunch** | | |
| 13.20 – 14.00 | **Rare neurosurgical conditions** | Mr Roddy O'Kane,<br>Consultant Paediatric Neurosurgeon, RHC Glasgow | |
| 14.00 – 15.45 | **Rotational workshops** | | |
| Each Workshop lasts 30 minutes | Stoma care for the newborn<br><br>Cate Nicholl & Lesley McGuire<br>Stoma Nurse Specialists<br>RHC, Glasgow<br><br>L0-007 (ground floor) | Supporting parents as they step into the unknown<br><br>Dr Emily Fraser<br>Clinical Psychologist<br>RHC, Glasgow<br><br>L0-006 (ground floor) | Managing cleft lip and palate in the newborn<br><br>Jenny Pettigrew<br>Cleft Specialist Nurse<br>RHC Glasgow<br><br>Lecture Theatre |
| 16.00 | **Feedback & Close** | | |

RCPCH approval has been applied for this activity for CPD
in accordance with the current RCPCH CPD Guidelines.

## Follow us on Social Media! @ORC Glasgow


**Office for Rare Conditions**

Office Block  Zone 0.01

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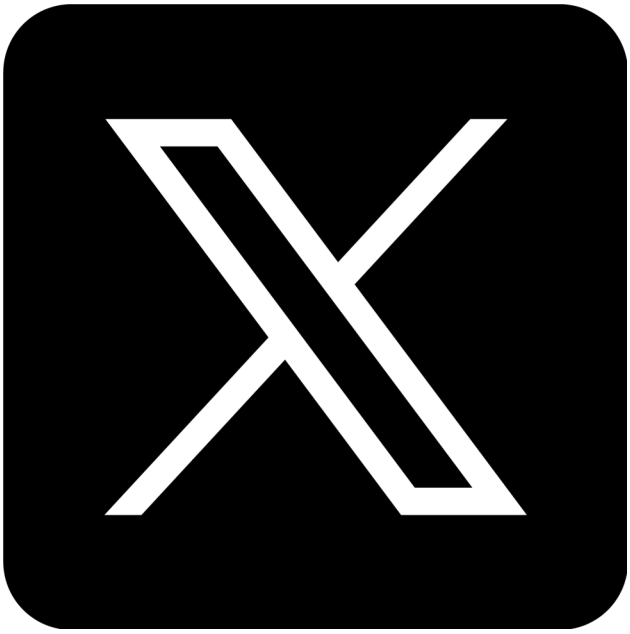
*17 - @orcglasgow*



*18 - @ORCGlasgow*

*19 - @ORCGlasgow*

*20 - www.officeforrareconditions.org*