

Thursday 29 February 2024

Rare Disease Day Since 2008 the last day of February has been marked as Rare Disease Day. A condition is considered rare if it affects fewer than 1 in 2000 people, but collectively around 300 million people around the world are affected by a rare disease. More than 70% of these diseases are genetic, and 70% of rare diseases start in childhood. To mark this event, we're grateful to Ashleigh Robson, a staff nurse in ANCHOR outpatients, for allowing us to share her story:

"I had a baby girl in March 2022 – Annabelle. She was born with a port wine stain on her leg and at two months old she had a GI bleed and was diagnosed with Klippel Trenaunary syndrome - a rare mosaic genetic disease.

"Following biopsies, Annabelle was then diagnosed with the PIK3CA gene mutation and noted to have PIK3CA related overgrowth syndrome (PROS). As a family, this was a very hard time for us, we felt completely alone. Thankfully we were pointed in the direction of the gopi3ks group, this was a godsend. They are a very small charity who love to help, advise and educate people on PROS. It is run by Mandy Sellars who has PROS herself. Last November, we all went down to London for a family weekend run by the charity. We met with doctors who specialise in this disease and had a full day of presentations to try and understand more. As valuable as this was, meeting other parents and families in the same situation was priceless and just the support we needed.

"I am so grateful to this small group for all the support and advice they have given us and will be doing everything I can to raise awareness of rare diseases."

For more information on Rare Disease Day, please visit their website: [Rare Disease Day 2024](#)

Financial position Thank you to everyone who came along to our finance drop-in sessions. We plan to run more of these sessions in the coming months; for now, if you were unable to attend, the presentation given has been attached to the email used to send out this brief.

ICYMI - Service redesign toolkit The transformation team have been developing a Service Redesign Toolkit to support services with the guidance, resources, and tools required to support in the improvement of referral pathways and vetting processes. The aim is to create an effective, more streamlined process for our patients, so that their waiting time is reduced, and they can be seen more quickly. Please see the attached poster and a link to the toolkit is here: [GRAM Service Redesign Toolkit - Home \(sharepoint.com\)](#). Please note you will need to request access to the toolkit.

Performance Appraisal for Managers and Reviewers. New courses on Performance Appraisal skills are available to book on Turas Learn: [Performance Appraisal Toolkit for Managers and Reviewers | Turas | Learn \(nhs.scot\)](#) (log-in required). A short video on how to navigate Turas Appraisal is also

available on the same page. Please contact the Learning and Development team (gram.learningdevelopment@nhs.scot) for any queries.

NoSCAR NMAHP research in Grampian seminar series The next seminar in this series, on "Writing Abstracts & Preparing Posters for Conferences", will be held via MS Teams on Thursday 7 March from 1.30-2.30pm. No need to book, [just use this link to join the meeting](#).

NES Knowledge Services - value and impact survey The Knowledge Network Scotland is NHS Scotland's national collection of digital library resources e.g., journals, articles, evidence summaries, databases and more. To maintain equity of access on a 'once for Scotland' basis, NHS Education for Scotland colleagues are asking for regular users of these resources to share feedback through this survey: [The Knowledge Network value and impact survey \(office.com\)](#) It takes 5-10 mins to complete and is open until end of March. Your answers will directly influence how much money NES has and what resources and subscriptions are purchased. For more on The Knowledge Network Scotland please contact [NHS Grampian Library & Knowledge Service](#).

Tune of the day It's a bumper selection today! Firstly, we bid farewell to healthcare chaplain Claudia Francis. Grace and friends wish her the best with her studies and dedicate [Hold On To Me](#) by Lauren Daigle to her.

Secondly, Vanessa Muir, a team leader in endoscopy, is moving on to new specialist role. After more than 20 years in the GI team, it's safe to say she will be much missed! Angie and the team have asked for [Best of You](#) by Foo Fighters

Finally, Mike Brown, dental clinical lead for Aberdeenshire, is limbering up for an epic cycling challenge. He and four chums set off from Edinburgh at 6am tomorrow, to cycle all the way to Rome in time for Scotland's 6 Nations clash with Italy and raise money for the My Name's Doddie Foundation. For Mike and his teammates, here's AC/DC and [It's A Long Way To The Top \(If You Wanna Rock 'n' Roll\)](#) (EP)

If you want to request a tune, follow up on items included in this brief, or suggest an item for sharing, drop us an email via gram.communications@nhs.scot