

Welcome to the Norrie Disease Foundation's Review of the Year!

What a first year it has been for the Norrie Disease Foundation! It is hard to believe we have only been operational for six months and we have achieved so much in such a short space of time. Whilst the charity became official in January, it was not until May that we formally launched at the Institute of Child Health in London. Since then, we have been very busy!

Research

We are really excited that Professor Maria Bitner-Glindzicz's **research group** at Great Ormond Street Hospital, which has a long history of working on genetic hearing, has been successful in her application for funding for a **research grant into the Norrie hearing loss from Newlife and from Sparks, The Children's Medical Charity**. The research is a laboratory study on hearing loss in Norrie disease and whether this can be treated.

Amina Al-Yassin, a paediatric trainee, and will be working with Professor Bitner-Glindzicz to try and understand more about the characteristics of Norrie disease in families. Amina hopes to talk to, and hopefully meet, as many people and families as possible so that the team at Great Ormond Street Hospital can try to build up a really good idea of the health issues experienced by people with Norrie as well as their journeys in the healthcare system.

One of the reasons for this research, is that if treatments for some of the complications of Norrie become available in the future, researchers need to know who to treat (everyone or just some people?), when to treat (before too much damage has been done) and how to tell if treatment has worked or not (the person might have fewer complications or a less severe medical problem than might be expected for a person of their age with **Norrie**). **This is called a 'natural history study' which means a study of what normally happens over time in a particular medical condition and we need to have this information if we want to offer new treatments.**

You can keep up to date with the research progress on the [NDF website](#).

Norrie Community Network

One of the main aims of the Foundation is to bring together families and the Norrie community to share experiences and provide peer support through the Norrie network. These are wonderful opportunities for the families to get to know each other and share their experiences of living with Norrie disease, and what has and **hasn't worked for them**. **NDF organised three meetings and we have big plans for 2018 so watch this space!**

Fundraising

Our fundraising efforts are currently focused on ensuring the community meetings can take place on a regular occurrence.

We have had some wonderful fundraisers this year, including a concert, football match and bootcamp sessions! If you would like to fundraise for NDF [please let us know](#) - we have written a fundraising document which hopefully gives lots of inspiration, information and advice!

'Chase Your Dreams' by XiJaro & Pitch and Type 41 is now on sale - with all proceeds going to the Norrie Disease Foundation. Anthony Pitch Reyers, who is blind, composed the tune and hopes this will inspire all people, regardless of disability and limitations, or people facing any situations, to chase their dreams no matter what life throws at them. 'Chase Your Dreams' was premiered on Armin van Buuren's A State Of Trance 844! You can download the track at: www.beatport.com/release/chase-your-dreams/2160533

You can also help raise funds through EasyFundraising - it's quick, easy, and doesn't cost you a penny! All you need to do is register with [EasyFundraising](#), select Norrie Disease Foundation as your chosen cause, and for every purchase you make through the site, The Norrie Disease Foundation will receive a donation.

Raising Awareness

BBC Children in Need Big Life Fix featured Josh, who has Norrie Disease and a team of designers and engineers who devised a way for Josh to play with his friends in the playground at school. Josh and his mum Wendy were also guests on BBC Breakfast to promote the programme.

Trustee Kelly Leggett featured in The Sun's Fabulous magazine, which ran an article featuring four women sharing their experiences of giving birth to a child with a rare condition. You can read the article here: www.thesun.co.uk/fabulous/4434018/inspirational-mums-children-genetic-disorders-jeans-for-genes

Taylor and Jackie Adams were invited to speak about Norrie disease at the Sparks Charity Winter Ball hosted by Emma Willis and Paddy McGuinness.

We have also been liaising with Contact (formerly Contact a Family) to ensure its information about Norrie disease and the Foundation and our support network is up to date so that it can be passed onto families. This information will be available in the new year.

NDF was invited to talk at the GOSH charity review meeting in November, which discussed the new Sight and Sound Hospital currently being built at GOSH. It is brilliant to see Norrie disease being mentioned as one of the rare diseases that they are supporting through appointments, treatment and research.

NDF Ambassador

We are thrilled that author and presenter, Simon Reeve, has become our first Ambassador. Simon worked on the Big Life Fix with Josh, who has Norrie disease, where he got to understand the difficulties that visually impaired children experience in everyday life.

With Simon's support, we hope to raise awareness of the challenges faced by children with Norrie disease and highlight the important role play facilities have on ensuring visually impaired children are able to socialise with their peers.

Simon said: "Meeting Josh through The Big Life Fix drove home how difficult life is for children affected by Norrie disease. There are huge obstacles in the way of many of those everyday things we take for granted for our children. Having an organisation like the Norrie Disease Foundation to raise awareness of these challenges and to provide vital support can make all the difference to the children and families affected."

We would like to thank the Norrie community and our NDF friends for all of your support in 2017 and look forward to having an even more productive 2018!

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