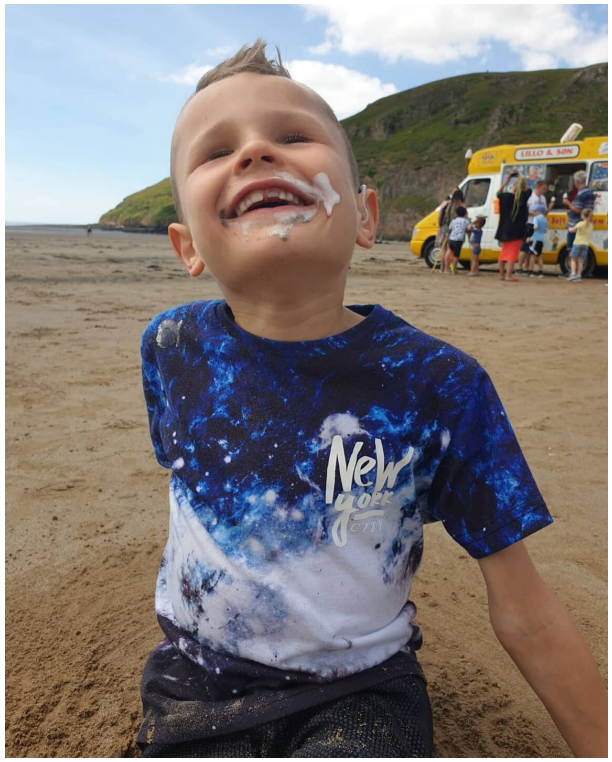
Annual Report 2018

THE NORRIE DISEASE FOUNDATION

Changing lives and making a difference to those living with Norrie disease



Norrie Disease Foundation

Our vision

To see a better supported and informed Norrie community and to find a treatment for the Norrie disease hearing loss.

Our purpose

To share information, resources and contacts; provide funded family days; and to actively support research, starting with treating the hearing loss through gene therapy.

Our beliefs

We believe in empowering the Norrie disease community through support and medical research so that they have the fullest and richest life experiences, and each individual can reach their maximum potential

Celebrating moments

2018 was another brilliant year for the Norrie Disease Foundation.

We gained 3 new Ambassadors, each with Norrie disease who will work alongside our first Ambassador, Simon Reeve. Our driven and dedicated Ambassadors promote our work; raise awareness of Norrie disease and steer the direction of the Board.

The NDF Board created a more challenging fundraising target of £25,000 which was achieved with thanks to our fantastic supporters and a grant from Genetic Disorders UK Jeans for Genes day programme.

In November 2018 we celebrated our first funded family meet - this meant we were able to offer families transport costs and where applicable accommodation costs thanks to the Jeans for Genes Day grant.

The number of Norrie families in touch with the NDF in the UK rose to forty and work started on the creation of a Norrie Disease patient registry at Great Ormond Street Hospital - the aim of the database is to enable global collaboration in the future.

Pioneering research into treating the Norrie disease hearing loss at University College London, Great Ormond Street Institute of Child Health under the late Professor Maria Bitner-Glindzicz's team entered its second year and the NDF Medical Advisory Board held two meetings where they discussed the needs of the Norrie community and how best to meet them. This also included very exciting steps forwards in the creation of a dual sensory clinic at the new Sight and Sound centre opening in 2020.

However, 2018 wasn't all positive and in September 2018 we lost our biggest champion, Professor Maria Bitner-Glindzicz who was tragically killed in a cycling accident. There are no words to describe her loss and how much we miss her.

I would like to finish by saying a very big thank you to all our supporters - without you the NDF wouldn't be where it is today.

Wendy Horrobin
Chair, Norrie Disease Foundation

Remembering Maria

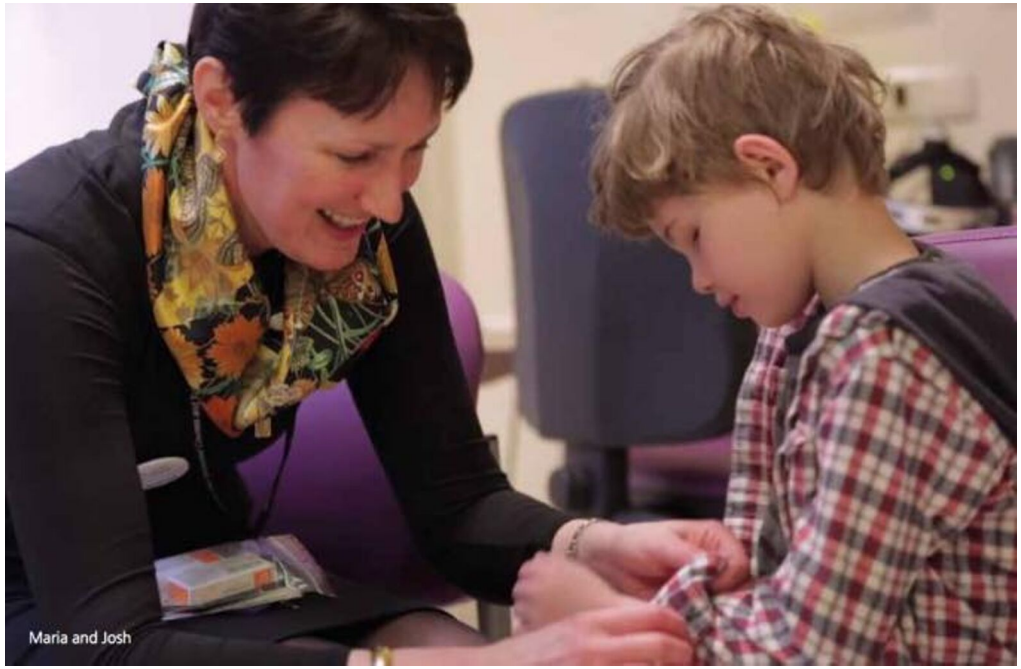
Professor of genetics at UCL Great Ormond St Hospital Institute of Child Health, Maria Bitner-Glindzicz had a special interest in researching and treating rare conditions with dual sensory loss. Tragically Maria was killed in a cycling accident in 2018.

Maria was the kindest, loveliest person you could meet and was the driving force behind the creation of the NDF and its medical advisory board. Her vision was to find a cure for the hearing loss associated with Norrie disease.

With Maria's dedication, expertise and support she steered us through multiple hurdles, including creating a strong UK family support network and championing the pioneering research into finding a therapy to treat the hearing loss.

Maria has left a strong and remarkable legacy, and we are determined to continue her amazing work which is currently in progress due to the dedication and support from her colleagues at University College London Great Ormond Street Institute of Child Health.

The NDF is incredibly grateful to the continued support from Maria's family to ensure her vision is achieved.



Maria and Josh

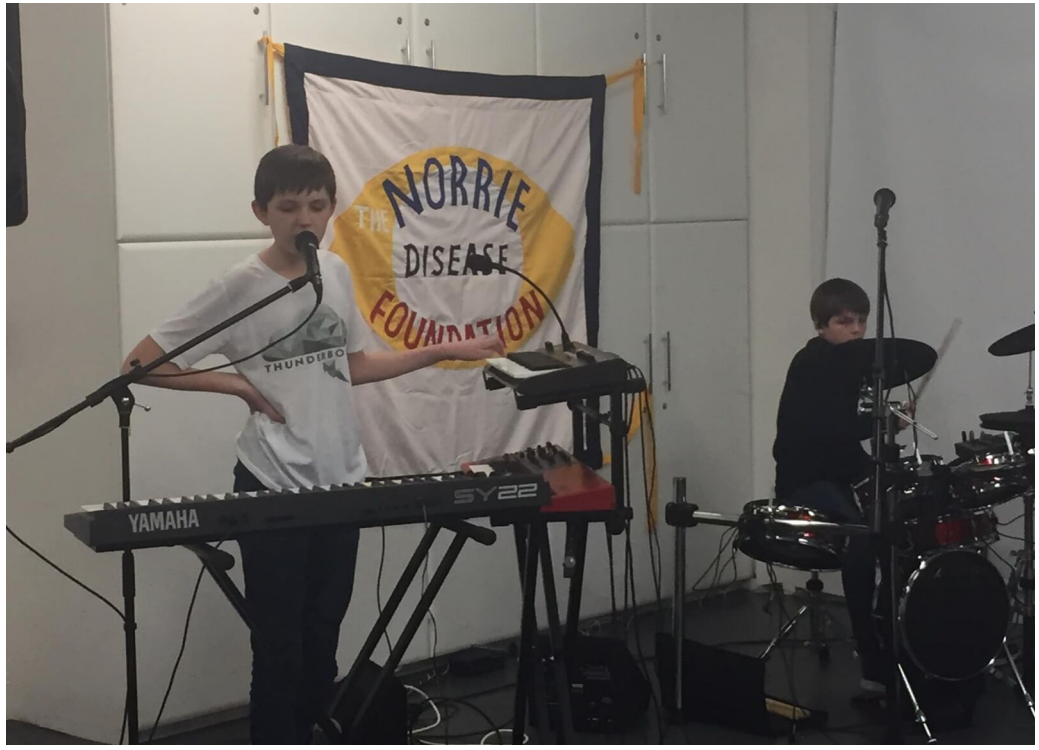
The first official Family Day

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.

In November 2018, the first official Family Day took place, enabling 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. For many this was the first time they had met another person with the same condition.

Members of the NDF Medical Advisory Board were also present, which enabled families to speak to medical professionals in a relaxed and non-clinical setting. We had 17 families attend, and activities included goal ball and an animal sensory session. The Thunderbolts, also gave a performance - one member of the band has Norrie disease.

To coincide with the family day, NDF created the first Norrie Awareness Week. The objective of the awareness week was to raise the profile of Norrie disease, the impact of the diagnosis on families, and highlight challenges families faced. The key aim however was to showcase positive aspects, such as achievements in music and sport, creativity, etc.



The family day was closed by The Thunderbolts who also released a single to raise funds to support days like this. Ethan, who plays the keyboard and is the lead singer, has Norrie disease, and his brother Jasper plays the drums.

Meet Cameron

At just 8 weeks old, Cameron was diagnosed with Norrie Disease.

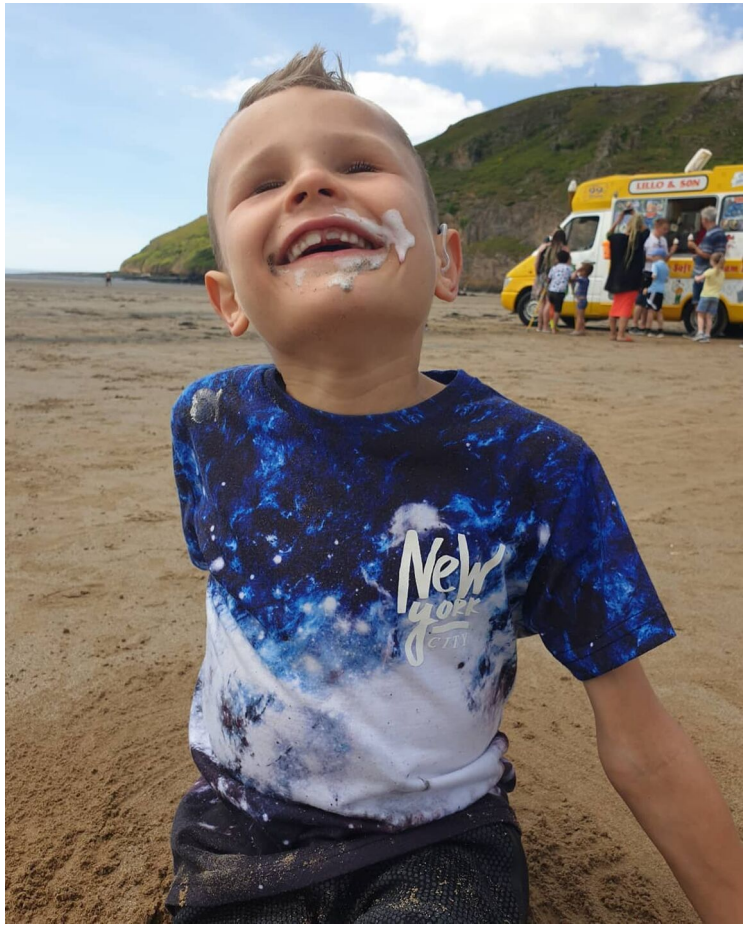
Mum Carla says: “It was very upsetting to be given the diagnosis. We were devastated. We were told Cameron has no vision or light perception at all. Finding out I was a carrier meant I had a sense of guilt that I had passed it on.”

Since the diagnosis, Cameron has had many challenges. In his first year Cameron had three general anaesthetics to remove scar tissue at the back of the retinas. A hearing test at the age of three revealed he had no hearing in his right ear. He could still lose his hearing in his left ear.

"We live in fear that the progressive hearing loss will affect his other ear before he develops the skills to talk. It is a ticking time bomb in the corner of the room, just waiting to happen."

Cameron had a feeding tube fitted in March 2017 as feeding had become a battle and he regularly ended up in hospital with hypoglycaemia. His development has been very delayed. He didn’t stand up until he was two and started to walk at three, taking a year to be able to walk unaided. He walks on tip-toes but needs a wheelchair when out and about. He doesn’t speak, and communication is very difficult. He was diagnosed with autism at the end of 2017.

But his personality shines through all of that, and the most important thing you will remember about Cameron is his smile.



Research

Medical Advisory Board (MAB)

The MAB was formed to guide the trustees in scientific matters, including research efforts and direction, and consists of renowned experts in the field.

The MAB continues to meet every 6 months which facilitates the expanding body of knowledge around Norrie disease so families can be better supported - the trustees are very grateful to the MAB members for their dedication and commitment to helping Norrie families.

The MAB takes the lead in identifying research opportunities.

Research Activities

The 3 year laboratory study at UCL GOSH, which is sponsored by Sparks and Newlife, is now half way through investigating the hearing loss in Norrie to better understand if this can be treated through gene therapy. The NDF researchers involved in this study are currently planning next steps research to reach this goal.

The NDF are working with UCL GOSH to set up a Norrie patient registry with view to collaborating globally.

Chair Wendy Horrobin and Dr Amina Al-Yassin traveled to Boston (USA) to present at the Norrie Disease Association's 4th International Conference.

Fundraising

We have been thrilled by the innovative fundraising of our supporters through concerts, quiz nights, and football matches.

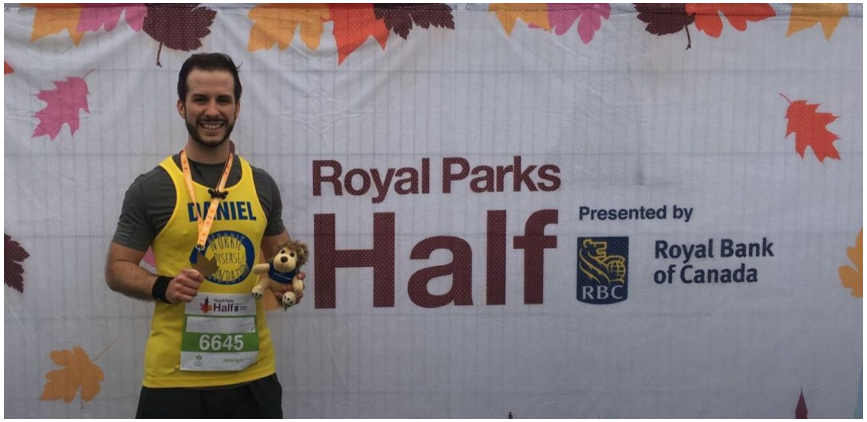
In addition, this year saw NDF secure charity places in the Royal Parks Half Marathon and successfully recruited three people to fundraise and run in aid of NDF. The NDF was also successful in securing a place at the 2019 London Marathon through the charity ballot.

To celebrate turning 40, our treasurer has taken on a year-long challenge to run 40 events in one year to raise funds for the charity.

The trustees would like to thank all of its incredibly generous supporters, particularly from the Norrie community in the UK, who have fundraised and given their time and energy to enable the charity to have such an incredible and successful second year.

We are especially grateful to Genetic Disorders UK for its support and providing grants to enable NDF activities to take place. We would also like to thank the AKO Foundation for its donation as part of its employee give back scheme.

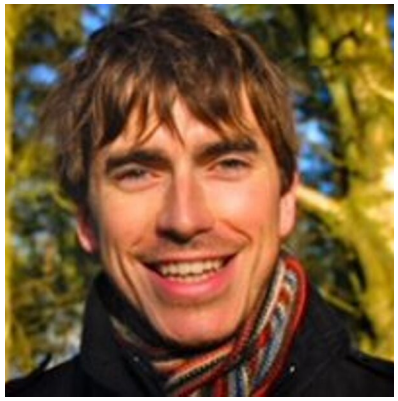
Finally, a huge thank you to John Ball primary school in south east London for selecting NDF as one its chosen charities to benefit from its fundraising 'Heath for Health community fun run'.



Our Ambassadors

The NDF was delighted that British author and television presenter, Simon Reeve agreed to become the charity's first ambassador shortly after the charity launched. In November 2018 three additional ambassadors from the Norrie community were appointed; Anthony Reyers, David Wilkins and Selina Litt all have Norrie disease and are hugely inspirational figures and role models for the Norrie community.

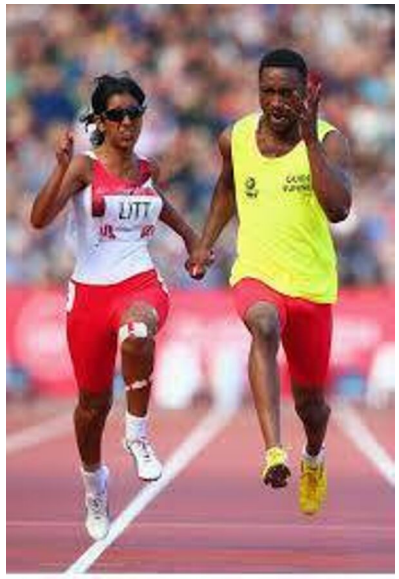
Together the ambassadors strive to raise awareness of Norrie disease and help to create more positive outlooks for our community.



Simon Reeve

While presenting The Big Life Fix for BBC Children in Need, Simon Reeve met Josh, who has Norrie Disease and heard first-hand how difficult Josh finds it to cope with the school playground.

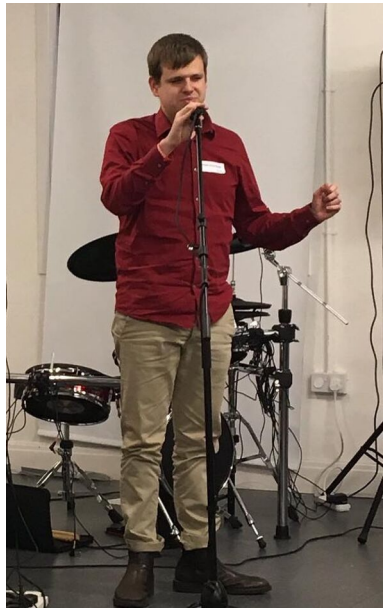
Simon says: "Meeting Josh drove home how difficult life is for children affected by Norrie disease. There are huge obstacles in the way of many of those everyday things we take for granted for our children. Having an organisation like the Norrie Disease Foundation to raise awareness of these challenges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



Selina Litt

Selina was the first female in the world to be diagnosed with Norrie Disease. Her brother and two uncles are also affected by the condition. Selina was born with vision, but gradually lost it over the years and now just has light perception in her left eye.

Selina says "Whilst coping with sight loss is challenging at times, I do my best to live life to the full. My biggest achievements are graduating with a First Class English Language and Literature degree and representing England at the Glasgow 2014 Commonwealth Games in athletics. I am very much looking forward to connecting with the Norrie disease community, sharing my knowledge and experience and helping to spread awareness of Norrie disease."

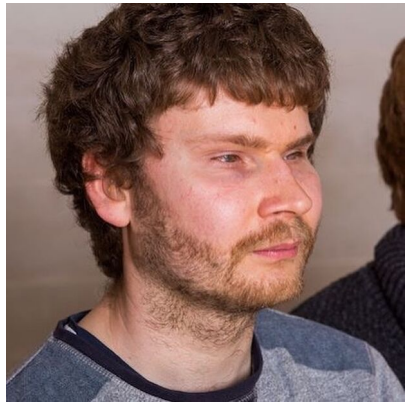


Anthony Pitch-Reyers

When he is not busy studying communication and media studies at KU Leuven university or traveling the world independently, spending time with friends or exploring new cultures, Anthony acts as one half of the melodic dance music DJ and producers of duo XiJaroAndPitch.

Anthony says "It is a true honour to be part of this organisation. No less than a year ago, I didn't know of any other people with Norrie disease, let alone that any research on the condition was being done.

"When I discovered the Norrie Disease Foundation and experienced their mission and shared passion of connecting people and promoting research first hand, that changed drastically. Offering my help to spread the word and reach even more people with our story and message, was for me hence not more than a natural step to follow."



David Wilkins

David was born totally blind and has been partially deaf since the age of 13. David says "The latter was a serious blow because I knew exactly what I was losing. However I've always been encouraged to have adventures, and not to allow my disabilities to define who or what I am. For this reason I've worked in South Africa, Guernsey and in the European Parliament.

"I'm here to promote our organisation and to speak for them wherever I go, locally, nationally and I hope one day internationally too. The foundation is involved in long and short term projects that will lead to the benefit of people affected by Norrie disease, a condition which I'm delighted to promote and raise money for as well. I'm also here to speak to parents and families who are wondering how to deal with this condition."

Our finances

Statement of Financial Actitivies

For the year ended 31 December 2018

	Unrestricted funds	2017
	£	£
Income and endowments from:		
Donations and legacies	23,344	8,529
Charitable activities	1,661	-
Total	**25,005**	**8,529**
Expenditure on:		
Raising funds	(2,699)	(3,734)
Charitable activities	(4,652)	-
Total	**(7,351)**	**(3,734)**
Net income	**17,654**	**4,795**
Reconciliation of funds		
Total funds brought forward	4,795	-
Total funds carried forward	**22,449**	**4,795**

The Norrie Disease Foundation
PO Box 12476
Colchester CO1 9RB

www.norriedisease.org.uk

Registered with FUNDRAISING REGULATOR

Charity Registration Number: 1171274

THE NORRIE DISEASE FOUNDATION