

RehabCare Report Calls for Centre of Excellence for Rare Disorders in Ireland



Pictured above: At the launch of RehabCare's research into rare disorders were (left to right) Professor Hilary Hoey, Head of the Department of Paediatrics, Trinity College Dublin and Consultant Paediatric Endocrinologist with the National Children's Hospital, Tallaght, Angela Kerins, Rehab's Chief Executive, and Sarah Jane Dillon, Rehab's Director of Policy and Co-ordination.



They form a sector of our community that is often overlooked, but a research report published by RehabCare earlier this year has focused attention on the plight of individuals with rare disorders and their families on the island of Ireland. A rare disorder is classified as affecting five or fewer people in every 10,000 and it is estimated that between 5,000 and 7,000 rare disorders exist, the vast majority affecting children. Among the better-known rare disorders are Prader-Willi Syndrome, Cornelia de Lange, Fragile X and Williams Syndrome.

The Health Research Board-funded research, which took place between December 2007 and March 2008 and involved interviews with families affected by rare disorders and a survey of GPs in both Northern Ireland and the Republic, found severe deficits in the information available to health professionals.

With a staggering 60 per cent of GPs forced to access information on rare disorders from the internet, the report highlighted the need for reliable, accessible information to be available to health professionals from an Irish source, in a format suitable for families. A centralised information service would minimise the risk of families who experience rare disorders accessing worst-case scenarios, inaccurate and out-of-date information and feeling overwhelmed as a result.

“Many of the families that were consulted reported negative experiences of receiving information

about rare disorders. Doctors also highlighted the issue of the lack of information available with over three-quarters experiencing difficulties in providing information to families about rare disorders,” said Rehab Chief Executive, Angela Kerins. “A centre of expertise, supported by a website and guided by specialist health professionals and families, is therefore a necessity. The centre would provide social support and information on rare disorders, outreach and community-based services as well as training to healthcare professionals, teachers and other community stakeholders.”

With over 72 per cent of GPs reporting that having a rare disorder gives rise to family problems, including feelings of isolation, additional financial pressures and siblings feeling overlooked, the new centre would also address these needs through the provision of counselling, respite care, peer and sibling support.

Welcoming the research, Professor Hilary Hoey, Head of the Department of Paediatrics, Trinity College Dublin and Consultant Paediatric Endocrinologist with the National Children’s Hospital, Tallaght, said, “This provides an important evidence base as, until now, little research has been done on the social support and information needs of families who experience rare disorders across the island of Ireland. RehabCare’s research not only benchmarks the current provision of information and support services but plays an important step in helping to inform future support service development.” ■

Annette's Story

"He's my forever-baby", says Annette Kinne of her four-year-old son, Alex, who was born with the rare disorder, Mowat Wilson syndrome.

In fact, Alex's condition is so rare, that when he was diagnosed at the age of 15 months, he was the only case of Mowat Wilson syndrome in Ireland. Since then, Annette says that two more cases of the genetic condition have been diagnosed in Ireland.

Annette speaks with great knowledge about her son's condition. "Mowat Wilson Syndrome can affect children differently. However, there are some common traits. For instance, moderate to severe learning and physical disability is usual, as are heart and kidney problems. Epilepsy is also extremely common, as are speech difficulties. Muscle tone among children with the syndrome tends to be badly developed, most will not walk until they are at least four years of age and some will never walk independently at all."

Unfortunately, Annette's knowledge of Mowat Wilson's syndrome is not something that the medical profession as a whole tends to share. "In many respects, we were quite fortunate. A geneticist we attended in Crumlin Childrens' Hospital had been at a conference where Mowat Wilson syndrome was discussed, and he was able to diagnose Alex as soon as he had examined him. However, the geneticist was about the 12th consultant that we had seen over an eight-month period, so the search for a diagnosis certainly wasn't straightforward."

Even after Alex had been diagnosed, the Kinne family still encountered problems with the lack of knowledge about the condition among the medical profession. "Every time we visit the hospital with Alex, we're constantly having to repeat what Mowat Wilson's syndrome is, and how it affects Alex. Doctors then usually have to go off and research the condition, sometimes on the internet, to get further information before they treat Alex.

"A centre of excellence on rare disorders is something that was identified in the RehabCare report, and it is badly needed. At the moment there is no centre where you can go for advice, and sometimes you can feel really on your own.

"The sharing of information is vital for families affected by rare disorders, but so too is research into rare disorders. Peer and sibling support is also important, so that the siblings of those with rare disorders aren't forgotten themselves. They need to have a childhood too."



Pictured above: Annette Kinne with her son, Alex, at the launch of the RehabCare Report on Rare Disorders on the island of Ireland.

“

Every time we visit the hospital with Alex, we're constantly having to repeat what Mowat Wilson's syndrome is, and how it affects Alex.

”