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Media Release

Mum thanks RUH for diagnosing young son's rare genetic disorder

The mother of a three year old boy born with a rare genetic disorder has thanked the Royal United Hospitals Bath NHS Foundation Trust for helping to diagnose his condition.

Magdalena Watling's son, Hugo, has Angelman syndrome, a neuro-genetic condition that affects the nervous system and causes severe physical and learning disabilities.

Magdalena, a former RUH midwife and current Birth Trauma Therapist from Peasedown St John, said she first realised something was wrong a few weeks after Hugo was born.

"When Hugo was born he seemed healthy and there were no complications but after about five weeks we noticed he was having problems feeding," she said. "He was being sick a lot and had lost around 20% of his weight so we were really worried. Nothing we could do seemed to help.

"He also wasn't behaving like you would expect a baby to. He wouldn't let us cuddle him or hold him – he would arch his back and scream. We knew something wasn't right."

Because of his age and condition, it wasn't possible for Hugo to have a successful MRI scan at the RUH until he was nine months old. The scan showed that there were some abnormalities to his brain so Hugo was later given a genetic test that concluded he was missing a piece of chromosome 15, diagnosing Angelman Syndrome.

Magdalena said: "We were very fortunate to meet Dr Tobias Hunt at the RUH who was fantastically patient and understanding and had an awareness of rare genetic conditions, enabling him to diagnose Hugo when he was aged one.

“The support we had from not only Dr Hunt but so many other staff at the RUH over that first year of Hugo’s life was incredible and I’m so grateful. All of the different therapists we came into contact with were also excellent, so helpful.

“Not all families are so fortunate to meet a doctor with such an awareness of rare illnesses and receive such an early diagnosis. Better awareness of rare genetic conditions across society can make a huge difference to all of those affected and in setting goals and expectations for the future.

“That isn’t to say life is easy, but with the help of charities like AngelmanUK and the wonderful skills and knowledge of the staff at the RUH there is now a wealth of information about disability and rare genetic conditions and no one should be feeling alone.”

Magdalena, who is married to husband Daniel with whom she also has an eight year old daughter, Rosamund, said that despite the difficulties Hugo faces, he is a big character and already popular with his friends at pre-school.

“He is a very happy, smiley boy, which is wonderful to see,” she said. “He adores water so loves going swimming and it’s always hard to get him out of the bath at bathtime!

“He’s a real explorer too and is quite nosy – he always wants to know what’s going on and enjoys annoying his sister. He loves being around other children too and has already made some really good school friends.

“He’s a real character, a lovely sweet boy, and we all love him to bits.”

You can find out more information about Angelman syndrome on the NHS website:

<https://www.nhs.uk/conditions/angelman-syndrome/>

There is lots more information and support available on the Angelman UK charity website:

<https://www.angelmanuk.org/>

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Notes to Editor:

At the RUH we’re proud to put people at the heart of what we do, striving to create an environment where everyone matters. Everyone means the people we care for, the people we work with and the people in our community.

We provide a wide range of services including medicine and surgery, services for women and children, accident and emergency services, and diagnostic and clinical support services.

We are also provide specialist services for rheumatology, chronic pain and chronic fatigue syndrome/ME via the Royal National Hospital for Rheumatic Diseases which we acquired in 2015.



Royal United Hospitals Bath

NHS Foundation Trust

In 2021, we acquired Sulis Hospital Bath, an independent hospital that provides care for both private and NHS patients. This has enabled us to provide more care for NHS patients, as well as continuing to provide private care to those who choose it. Any additional income earned through private care is reinvested in services for the benefit of the people we care for at both Sulis and the RUH.

We're currently building a new Cancer Centre at the RUH. The Dyson Cancer Centre, which is set to open in autumn 2023, will help transform the care we provide for patients, families and carers.

We work closely with other health and care organisations as members of the Bath and North East Somerset, Swindon and Wiltshire Integrated Care Board. We strive to improve the health and wellbeing of the people in our community by working together build one of the healthiest places to live and work.

We are rated 'Good' by the Care Quality Commission (CQC).

For more information about the Royal United Hospitals Bath NHS Foundation Trust visit: www.ruh.nhs.uk