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Shrewsbury family reveal Ruby's story to help others

Jane and Greg Cotton's little girl is one in 10 million – for eight-year-old Ruby has been diagnosed with an extremely rare genetic condition known as Ring 14 Mosaicism.

From left, mum Jane Cotton, Ruby and childminder Kym Crippin

So rare is the condition that the youngster is only one of 14 people diagnosed with it in the UK and 106 worldwide.

Now her parents want to share Ruby's story to raise awareness about the syndrome and help other families in similar situations.

Ruby and friends enjoy a tea party in the youngster's honour

Ruby's diagnosis at 10 months old came as a huge shock to Jane and Greg, of Shrewsbury.

Jane had had a normal pregnancy and birth, and Ruby was born on May 16, 2005, weighing 6lbs 9.5ozs. But there were soon signs all was not right.

“During the early weeks she was very unsettled and most nights would cry for eight or nine hours in apparent discomfort,” said Jane.

They were told Ruby had colic and was prescribed medication.

“As the months passed Ruby continued to be an extremely fretful, irritable baby. She would cry if anyone other than Greg or I entered the room, which was upsetting for family members and friends,” said Jane.

“Ruby would frequently refuse to feed. Despite this she gained weight steadily and gave the health visitors no worries."

When she was 10 months old, Ruby suffered a seizure and was rushed to hospital. Tests came back normal but Ruby continued to suffer from fits.

She was moved to Birmingham Children’s Hospital where she was diagnosed with epilepsy. Tests later revealed she had Ring 14 Syndrome – a genetic condition affecting the 14th chromosome.

Jane said: “We were devastated, heartbroken about the uncertainty of her future. We did not know if she would walk, talk or have any quality of life. It is so rare that neither we, family nor friends, had even heard of it.”

Jane and Greg used the internet to try and find as much information as they could about Ruby’s condition and finally discovered a support group for rare chromosome disorders.

“Since then we have been the ones to explain what it is to every doctor we have met on our journey,” said Jane.

Ruby suffers from difficult to control epilepsy, severe learning difficulties, has very little speech and has severe curvature of the spine. She needs one-to-one constant care and her behaviour can be unpredictable. The couple now want to share Ruby’s story to raise awareness about her condition.

“The more people that know about it the more hope there is for the families,” Jane said. “Despite all her difficulties, Ruby is a cheeky eight-year-old who loves music and being outdoors.

Ruby faces challenges each day but every small achievement is huge to us. We’re so proud of her and love her dearly. The name Ruby means “precious jewel” and to us that is what she is.”

Ring 14 research is solely funded by private donations. To make a donation which will go to directly fund critical research visit www.ring14.org/eng