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What is Kabuki Syndrome

Kabuki Syndrome is a very rare genetic condition which affects approximately 1 in 32,000 births worldwide.

Kabuki Syndrome is complicated and has been underdiagnosed due to a lack of awareness amongst the medical profession.

People with Kabuki Syndrome often have congenital heart defects, hypertonia, feeding difficulties, cleft palate, speech and language delay, hearing problems, learning difficulties and kidney abnormalities, to name just a few.

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What is Kabuki UK

Kabuki UK is a newly established charity which has been set up by a group of parents who all have children with Kabuki Syndrome.

Each of us has been through the confusion, pain and heartache of a Kabuki Syndrome diagnosis.

We have all experienced the isolation that comes from having a child with such a rare and complicated condition.

Until now there was no UK-based organisation to support people with Kabuki Syndrome and their families. Although there are wonderful organisations in other countries their advice and experiences are often different to what we find here.

Kabuki UK has been set up to address this problem. The website (www.kabukiuk.org.uk) has a wealth of information on the health, educational and developmental aspects of Kabuki Syndrome but is written in a reassuring way. There are lots of photographs of children with Kabuki Syndrome having fun with their friends and siblings.

We are all now committed to establishing a support system for newly diagnosed families in the UK. Our network of regional contacts will provide a friendly support by phone, email and through family get togethers.

We know that this charity will make a big difference because it already is.

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What are our aims?

To raise awareness of Kabuki Syndrome by distributing information to local hospitals and gaining a higher internet and press presence.

To provide information, advice and resources about Kabuki Syndrome and establish a network of regional coordinators, who will provide support and help for newly diagnosed families in that region.

To fundraise, with both regional and national events, to support the charity's aims.

To organise and publicise family events and fundays, which will provide families affected with Kabuki Syndrome with a chance to meet other families and share experiences, as well as have an enjoyable break.

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Who are we?

Everyone involved with the running of this charity is a volunteer who has a child with Kabuki Syndrome and so we understand better than anyone the worries that come with such a diagnosis.

Tommy and Beth Allen - beth@kabukiuk.org.uk - Beth and Tommy Allen live in Shropshire with their five-year-old daughter Rosie and Charlie, aged 2, who was diagnosed with Kabuki Syndrome in September 2011. The struggles Beth and Tommy have faced in getting the right care and treatment for Charlie, as well as overcoming the lack of knowledge and awareness of Kabuki in the medical world, have made them determined to help current and future Kabuki families as much as they can.

Lisa Johnston - lisa@kabukiuk.org.uk - Lisa and Jason live in Bournemouth with their 5 children. Kitty was born in January 2009. She was diagnosed with kabuki syndrome at 13 months. It was a very isolating time for Lisa and Jason to begin with no support group to turn to. They have been passionate about fundraising and raising awareness of kabuki. They have held a kabuki family day at their home and have organised 3 successful fundraising events.

Sally Trewartha - sally@kabukiuk.org.uk - Sally and Paul Trewartha live in Essex with their son Harry who has Kabuki Syndrome and his little brother Toby who does not. Sally feels passionately that with the right help children with Kabuki Syndrome can go on achieve great things.

Emma Fisher - emma@kabukiuk.org.uk - Emma is the charity Treasurer

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What have we done?

Fundraising - walk for a walker

Kitty was diagnosed with Kabuki Syndrome at 13 months. Kitty's particular presentation of Kabuki Syndrome means that she is unable to weight bear and therefore cannot walk.

At almost three years old, Kitty was becoming increasingly frustrated at not being able to run around with her friends and siblings. Kitty was assessed for a walker but unfortunately there was no NHS funding available to pay for it.

Kitty's family and friends organised a sponsored walk to raise the money to buy a walker. This was so successful that, due to peoples wonderful generosity, they were able to buy not only the walker but a specially adapted bike as well.

Kabuki UK now hope to do similar, life changing, things for other children.

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What have we done?

Family days

Last Summer saw the first gathering of families with children with Kabuki Syndrome organised by the Kabuki UK trustees in Bournemouth. Five families enjoyed a day of fun on the beach.

These days are hugely important for the parents as most people do not know anyone else with a child with Kabuki Syndrome; this can make parents feel extremely isolated.

It is even more important for the children to grow up knowing other people who have Kabuki Syndrome and face similar challenges.

Kabuki UK plan to organise and fund family days on a regular basis in several locations across the country.

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How can you help us?

We are, like all charities, always looking for people to help us fundraise so that we can achieve our aims. There are a number of ways that you can help support Kabuki UK.

- Organise a sponsored event - whether it be taking part in a marathon or a walk!

- Donate money via Paypal (see www.kabukiuk.org.uk)

Any money which is given to Kabuki UK will be used directly to help people with Kabuki Syndrome. That could be a direct payment to a family to buy a walker, like Kitty, or to fund a family day where children can meet other people with Kabuki Syndrome for the first time. We are all volunteers and have minimal overheads.

Corporate sponsorship and sponsorship in kind

We are a small charity with big ambitions. To buy a walker like Kitty's costs £1,500, to put a brochure explaining Kabuki Syndrome in front of every GP in the country will cost a lot more.

We need help to achieve our aims. If your organisation could help us either by donating money, goods, expertise or time we would love to hear from you.

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Case study

Harry Trewartha - Kabuki UK case study

Essex toddler Harry Trewartha suffers from Kabuki Syndrome, a rare genetic disorder that led to open heart surgery and weeks in intensive care when he was just four days old.

Kabuki Syndrome is extremely rare and affects only one in 32,000 people. For Harry, who is four-years-old and about to start school, this means a horseshoe kidney, cleft palete, developmental delays, feeding difficulties, hearing problems and distinctive facial features, including protruding ears and large, bright blue eyes.

Harry's heart condition was diagnosed during pregnancy but the Kabuki Syndrome was not diagnosed until he was 18 months old by the genetics team at Great Ormond Street Hospital. Harry and his family have been part of groundbreaking genetic testing into the gene responsible for causing Kabuki Syndrome.

At the time of Harry's diagnosis there was no support group in the UK for families affected by Kabuki Syndrome. There was no one to turn to for advice or help and it is so rare that the majority of doctors have to google Kabuki before they see Harry.

Harry is about to start school where he will have specialist support in his learning, this will be a huge challenge for Harry as he is behind his peers in almost every way. However Harry is resilient and constantly surprises people around him with his ability to rise to a challenge.

Harry's Mum is one of six parents who have now formed Kabuki UK. They aim to support newly diagnosed families through the establishment of a network of contacts, to create awareness of the syndrome among the medical profession and to raise funds to pay for specialist equipment.

Yes, Harry has Kabuki but there is far more to him than that. He is the sweetest, most loving and friendly child, he is always smiling and he understands so much. His family hope that despite his challenges and with the right love and support Harry will grow up into a happy, caring, fun-loving man.

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Facts and figures

Kabuki Syndrome was first described in Japan in 1981, by Dr Niikawa and Dr Kuroki. It was later described in Europe and America around 1990.

On August 15, 2010 researchers at the University of Washington announced the discovery of the MLL2 gene mutations responsible for approximately 75% of incidences of Kabuki Syndrome.

Kabuki Syndrome is rare, affecting approximately 1 in 32,000 births.

50% of children with Kabuki Syndrome will lose their hearing before they are five.

92% of people with Kabuki Syndrome will have some learning difficulties.

Congential heart defects are often found in people with Kabuki Syndrome.

Other features which are often indicative of Kabuki Syndrome are;

Poor feeding and failure to thrive

Hypertonia

Genitourinary anomalies

Cleft lip and/or palate

Gastrointestinal anomalies

Ophthalmologic anomalies

Dental anomalies

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How to contact us

For fundraising information please contact;
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