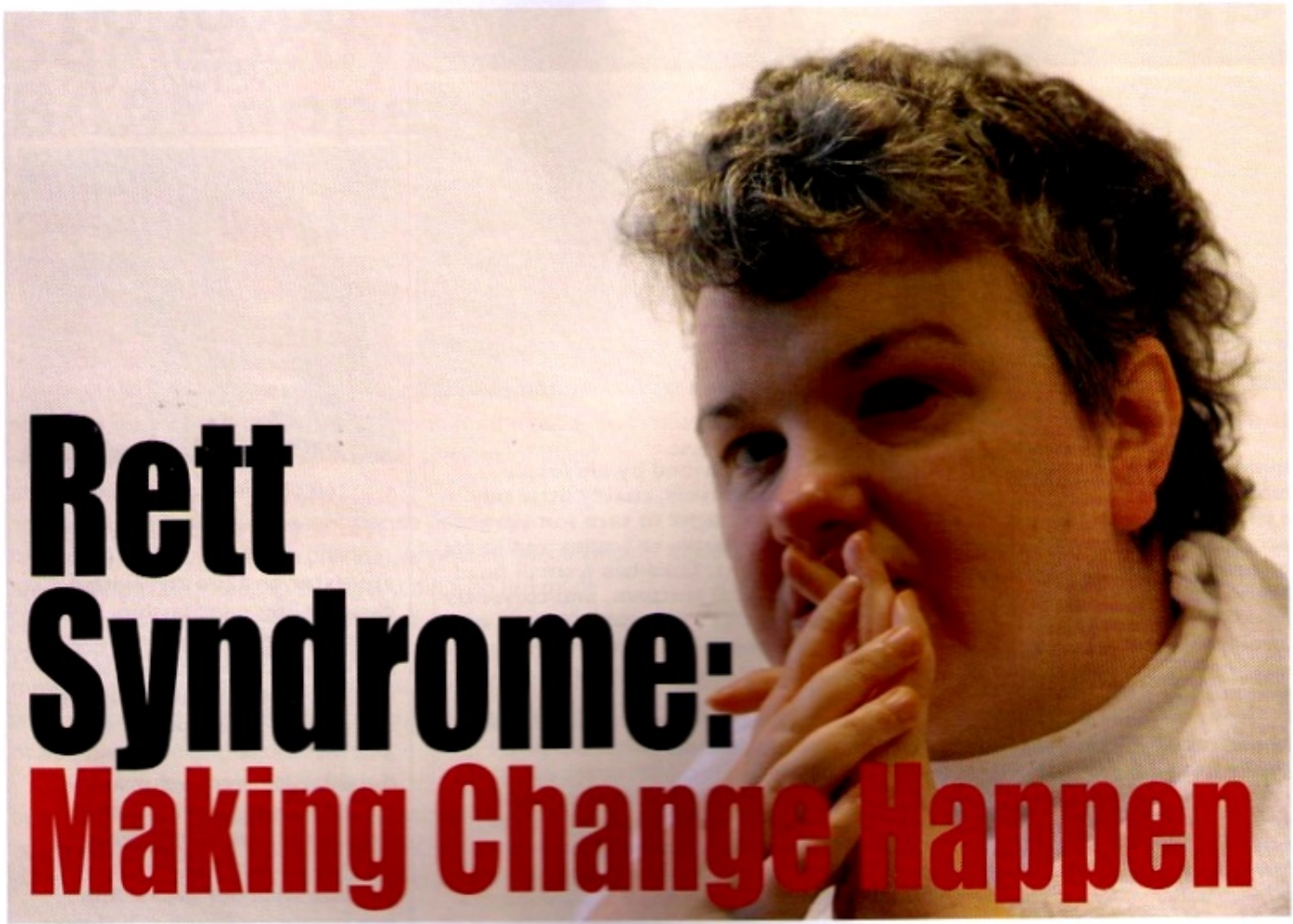

# Rett Syndrome: Making Change Happen

Last October, the 6th World Rett Syndrome Conference was held in Paris at the Maison de la Mutualité. It was the first such conference to bring together Rett syndrome suffers and related spectrum disorders. Despite this, very little about the syndrome has infiltrated our media thereby seeping into public consciousness. Many people do not even know of its existence.

Yet Rett Syndrome can be a debilitating condition, caused by a genetic mutation in the gene MECP2[1]. As the gene only acts on the X chromosome, girls are more likely to develop Rett Syndrome than boys. According to Melanie Ekless, Family Support Services Manager of the Rett Syndrome Association, there are just five boys in the UK who have the condition, and of the entire population in Britain, one in ten thousand people have been diagnosed.

Many specialists see Rett Syndrome at the extreme end of the Autistic Spectrum due to some shared characteristics (little or no speech, constantly wringing hands etc.) yet Autistic Spectrum Disorder affects mainly boys, up to seventy per cent.

"[Rett Syndrome] is a profound and multiple learning disability," Melanie explains. "Those with Rett Syndrome will need twenty-four hour support and supervision."

Diagnosis is made by clinical diagnosis (the practitioner observes the child and assesses their speech, language and motor skills) and by a recently developed blood test which tests for the MECP2 variation. Due to the nature of the condition, diagnosis is not made until the child is around nine to eighteen months old, when the development of the infant either slows down or stops altogether. For parents, the apparent cessation of their child's development can be a frightening time, especially if they do not understand why it has happened. But thanks to the discovery of the genetic cause, diagnosis and treatment can be relatively straightforward, providing treatment is set up early on.

Speech therapists can aid with

*"While every child diagnosed will not experience the same symptoms, some symptoms are commonly experienced in varying degrees, including Scoliosis."*

A physiotherapist supports a woman with Rett's syndrome in the pool

communication, and can teach children how to communicate their needs if verbal speech isn't possible. It is estimated that up to fifty per cent of those with Rett syndrome are unable to speak.

For those children who develop Scoliosis (the curving of the spine) as part of the condition, early treatment is essential: "Scoliosis tends to worsen over time, so physiotherapy is really important," says Melanie Ekless. "Joints contract too, particularly in the feet, so involving a physiotherapist specialising in learning disabilities is a must."

While every child diagnosed will not experience the same symptoms, some symptoms are commonly experienced in varying degrees, including Scoliosis. Many will also have Epilepsy and experience breathing difficulties: "autonomic difficulties are quite common," Melanie explains. "therefore it will be difficult to swallow because of chewing and moving food to the back of the throat. Many children may grind their teeth too." These children will need their food puréed, or in very rare cases, gastronomy tubes will need to be used, if malnutrition is evident.

Many will find it difficult to walk, and those that can will do so with a wide gait. Others will develop involuntary hand movements and twitches. As Melanie explains: "Rett Syndrome is a spectrum disorder of sorts, so it is often confused with ASD. Some people may have a higher level ability in one area and a lower level in another, so it varies." Yet Melanie does not believe there are obvious similarities between the two spectrum disorders. "Autism has a lot of hand movements," she says. "When you know them well and can recognise them, these movements are actually quite distinct. Autism has become quite well known over the past ten to fifteen years, but Rett is still fairly unknown, so people don't always know how to differentiate between them."

Making music: A young boy with Rett's Syndrome plays the tamborine

Organisations like the Rett Syndrome Association work hard to promote understanding, awareness and research, thereby improving the quality of life for people with the condition. The Rett Syndrome Association brings families together, and offers them support and advice. Specifically, it organises annual family weekends which gives families a chance to meet and share experiences and attend workshops and seminars run by professionals. A buddy scheme is offered to families attending for the first time. Melanie, who personally organises the programme and the speakers says: "It is a time for families to come and be together and learn a bit more about Rett syndrome and support one another." She adds, "We organise speakers who have either direct relevance to the syndrome or indirect, such as the orthopaedic consultant last November who spoke about Scoliosis." The weekend, which took place last year at the Hilton Hotel, Northampton, included a dinner and dance, holistic massage, a quiz session and music workshops as well as the informative seminar sessions with medical professionals.

The Rett Syndrome Association needs to raise £500, 000 to continue to run its valuable services in 2009. Every donation counts. If you would like to donate, please visit the website at **www.rettsyndrome.org.uk** or to find more information about current campaigns and support services for families affected by Rett Syndrome. S

[1]This gene mutation was first discovered to be the 'cause' of the condition as recently as 1999.

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