Rett Syndrome Awareness - A Parents Experience

Karen and Micheal Borg

What is Rett Syndrome?

Rett Syndrome is a complex neurological disorder. It affects mainly girls. Although signs of Rett Syndrome may not be initially obvious, it is present at birth and becomes more evident during the second year. People with Rett syndrome are profoundly and multiply disabled and totally dependent on others for their needs throughout their lives. The pattern of characteristics associated with Rett syndrome was first described in 1966 by the Austrian doctor, Professor Andreas Rett.

Why does Rett Syndrome occur?

Rett syndrome is genetic in origin. It is probably the most common genetic cause of profound intellectual and physical disability in girls, occurring more commonly than 1 in 10,000 female births. Recently it is been detected that a large proportion of people with Rett syndrome have a mutation, or fault, on the MECP2 gene on the X chromosome.

How is Rett syndrome diagnosed?

Rett syndrome is now confirmed in most cases by carrying out a genetic test and also through clinical diagnosis – looking for signs and behaviours. Here are the typical signs. After appearing to make normal progress, a period of stagnation in development from about the end of the first year, which lasts until regression occurs;

- A regression period when skills in speech and hand movement are reduced, which occurs between about 9 and 30 months;
- Development of repetitive hand movements (wringing, patting, clapping, tapping, mouthing);
- Appearance of stiff or clumsy posture or gait;
- A normal head circumference at birth but a slowing of head growth between 2 months and 4 years approximately;
- Absence of other disease, syndrome or injury to account for the above features;

Other features which are often seen include:

- Breathing irregularities including hyperventilation, breath-holding and air swallowing;
- EEG abnormalities;

- Epilepsy – over 50% of people with Rett syndrome have some form of epileptic seizure at some time;
- With age, muscles become increasingly rigid, joint deformities and muscle wasting may occur;
- Unsteady wide-based gait (about half of those with Rett syndrome achieve independent mobility);
- Development of scoliosis (curvature of the spine);
- Growth retardation.

Theresa' Story

Theresa, who has just turned 5 in September, is a child with special needs. She does not speak and cannot communicate her needs. She also has poor motor co-ordination and cannot use her hands properly. She is totally dependent for all her needs. She does not play with toys or with her sister. But Theresa loves music, especially nursery rhymes and action songs. These songs tend to calm her down when she is agitated and are a wonderful tool just to hear her laugh! Theresa was one year old when we, her parents, began to question her development. This is her story.

Theresa was born in September of 1997, and was a good and quiet baby. It seemed to us that she was leading a rather normal life, until we became concerned that she did not speak by the time she was 1 year and six months. Our first appointment was with a speech pathologist who suggested that we carry out an audio test to check if Theresa could hear properly. When a detailed hearing test, the ABR, resulted in Theresa having a normal hearing, our paediatrician suggested we carry out routine tests, to find out if Theresa was just slow or mentally disabled.

We passed through a whirlwind of blood tests, urine tests, a CT Scan, MRI, EEG. Two separate blood tests were sent for genetic testing for Fragile X and for Angleman's Syndrome, but both were negative. Between October and December 1999, Theresa was assessed by the team at the CDAU (Child Development Assessment Unit) at St. Luke's Hospital. Her initial diagnosis stated Communication Disorder (mainly because Theresa could not talk, point, gesture or communicate). Since then, other professionals in the field, both privately and through hospital have come up with a number of 'diagnosis' from a form of mental retardation, autism, and finally a global developmental delay. It was only in November of 2001 when Theresa was seen at St. Luke's Hospital by a visiting consultant from the UK, that he suggested that we should carry out genetic testing for Rett Syndrome. The blood sample was taken in December 2001, and we were told that it would take over 6 months for the results of this test.

As six months is a long time to wait, especially when you own child's health is concerned, we decided to go to England during Easter, to visit a neurologist at a private clinic. The neurologist and other members of his team including a geneticist, confirmed their clinical diagnosis as that of Rett Syndrome. And sure enough, when we received the results of the blood test in Malta in July, it also confirmed that Theresa had Rett Syndrome.

Theresa's Education

During the trying time until Theresa's diagnosis was confirmed, we did not just wait around for the results. We invested in Theresa's educational programme immediately.

In fact, Theresa has been attending the Eden Foundation since she was 2 ½ years old. She attends the STEP programme (Structured Training & Education Programme), mainly geared for children with autism. As Theresa was thought to have had autism when we first contacted Eden, she was put on the STEP programme, but her education programme has since been adapted to suit her specific needs. Theresa attends her programme on two days. Theresa's school facilitator also visits Eden once a week and joins in the activities with Theresa and her Eden tutor. In this way there is certain continuity for Theresa between Eden and school.

Theresa is very happy at Eden and we too are pleased that we have taken the decision to send her there. As parents we are very involved in the work that is done with our daughter. We are kept informed of anything new that she does and together with all the professionals involved with Theresa, we regularly attend a review of her progress and plan the next step together. We very much feel that we are part of the Eden 'family'. We also feel that we have made lots of friends here, not just with Theresa's tutors and therapists, but also with lots of parents whose children attend the Eden Foundation.

On the other three days in the week, Theresa goes to school where she has a full time facilitator. Theresa also loves her school, especially her classmates. They include her in everything they do, from painting to singing, even if she cannot do anything herself. She loves to watch the children and just stay next to them. They all love sitting in a circle and singing songs best, because during song time Theresa is the happiest. She will laugh and smile at them and the other children are encouraged to make her laugh and smile again. They all take turns banging on the tambourine and Theresa joins in here and when it is her turn the other children encourage her to hit the tambourine. Theresa is not a 'disabled child' to her classmates, but just one of them.

Theresa's family

Theresa also has a 7 year old sister, Gillian, whom she loves very much. Although Theresa cannot join is her sister's play, she likes to watch her playing and misses her terribly when Gillian is not at home. Gillian has learnt all the songs that Theresa loves best by heart, and will sing to her often, just to hear her giggle. It is not easy being the sibling of a child who requires so much attention and care and Gillian has had to act older than her age on more than one occasion.

As a family we try to lead as 'normal' a life as possible. We take Theresa with us wherever we go. We love to do things all together. We have learnt to accept Theresa for who she is, not for what we would have liked her to be. We also believe that by living the present moment we are encouraged to keep going. The past is past, the future does not belong to us. Today is what we have to work with.

We would just like to add that we would like your support in simple things. Help children to be sensitive to the needs of other children, especially those who cannot ask for help. Families of children with a disability have to pass through a whirlwind of trials to get along in life. It would be much easier for all concerned if our children are accepted for who they are. We have to stop looking at the Disability, but at what Ability these children have, and when you really get to know them you will find out how happy they are and that they only want to be loved. On their part they give UNCONDITIONAL love – no matter what.

Looking for support

When we were in England after we received Theresa's diagnosis, we took the opportunity of contacting the Rett Syndrome Association UK, based in North London. This is a national organization giving help, advice and support to parents, carers, siblings and professionals, in fact, anybody involved with a child or adult who has Rett Syndrome. We were given books, leaflets and even videos covering the subject of Rett Syndrome, which we have found very helpful. We have also subscribed to their quarterly newsletter RETT NEWS. They also offer telephone support on 7 days a week. In our case we use the e-mail and have been in constant contact with each other since our return from the UK. For further details contact: Rett Syndrome Association UK on info@rettsyndrome.org.uk www.rettsyndrome.org.uk. The Association also organize a Family Weekend every year in October where families from all over the UK meet together to share their experiences and where professionals in the field update parents on the latest research in Rett Syndrome. Besides information and support, they also offer respite care for the child with Rett Syndrome, and organize outings and fun programmes for their siblings while parents attend the talks.

Contacting parents

In Malta hardly anyone has heard of RETT SYNDROME. So far we only know of two other families whose daughter has been given a diagnosis of Rett Syndrome. We would like other families who have a daughter with this syndrome to contact us, for the sole reason of being of support to each other. If you would like to know more information about Rett Syndrome please contact Karen and Michael Borg on telephone no. 21421075 or via e-mail at michael.r.borg@gov.mt

OCCUPATIONAL THERAPY VIEWS

There are several areas that the Occupational Therapist focuses on with children with Rett's Syndrome. These include Sensory processing, motor planning, minimising stereotypical hand movements, maximising hand use for functional activities, toilet training and advice regarding use of specialised equipment. The use of splints in the treatment and management of Rett Syndrome in girls is documented in literature. (Crawford M.,1999) In fact the use of elbow blocking splints was used in Theresa's case where the hand-to-mouth movements were decreased significantly.

A brushing protocol was also used the aim of this programme is to increase the child's understanding of the information that comes into her body through the senses. The purpose of the programme is to help the child take sensory information into her nervous system in a more normalized manner without a defensive or negative response. The programme involves both brushing and compression to the joints. Together, the brushing and joint compression help to activate the areas of the brain that will regulate responses to sensory information.

In Theresa's case decreased discrimination of vestibular and proprioceptive input is also present. This is exhibited by poor posture, frequent falling, clumsiness and poor balance. Treatment focuses on providing intensive vestibular and proprioceptive information and improving postural responses. (Nackley VL, 2001) A weighted jacket was also used to increase proprioceptive input and to decrease tiptoe walking. Simple motor planning activities are also presented regularly.

Liaison with other professionals i.e. her facilitator and Eden tutors is carried out regularly. Moreover her sessions are held jointly by the occupational therapist and the physiotherapist.

REFERENCES

- Sensory Integration Special Interest Section Quarterly Volume 24 No 1, March 2001. Sensory Diet Application and Environmental Modifications, A Winning Combination.
- Crawford M.J.(!999) A Creative Approach to Treating Rett Syndrome. O.T. Practice