CARE

Given the multiple organ systems involved in Rett syndrome, a multidisciplinary approach ideally provided by a dedicated Rett Syndrome Clinic is vitally important to medical care. At home, an individual with Rett syndrome will need constant support from their family. It takes strength and energy to pursue the diagnosis, make an appropriate treatment plan and find people who will support it, in addition to providing all of the physical aspects of direct care giving. Even with all the challenges, there is great joy in raising a child with Rett syndrome. Their love is unconditional. They demonstrate amazing courage, patience and teach us many life lessons without speaking a word.

WHO IS O.R.S.A.?

The Ontario Rett Syndrome Association (O.R.S.A.) is a volunteer, not-for-profit charity for parents, caregivers, researchers, medical professionals and other interested support agencies and individuals. O.R.S.A. became incorporated in 1991. Its Board of Directors is comprised of parents and caring citizens. O.R.S.A. provides information through a website, a newsletter and conferences. Support is provided to families regionally. The association funds Canadian research projects, two Rett Syndrome Clinics, a resource centre and the Canadian Rett Syndrome Registry. O.R.S.A. advocates for the needs of individuals with Rett syndrome and their families provincially and nationally.

OUR MISSION

The Ontario Rett Syndrome Association (O.R.S.A.) exists to ensure that children and adults with Rett syndrome are enabled to achieve their full potential and enjoy the highest quality of life within their communities.

OUR SERVICES; HOW WE HELP

- Provide funding for research
- Funder and administrator of Canadian Rett Syndrome Registry
- Fund and support clinics in London, Ottawa and Toronto
- Advocate
- Host activities to provide caring support and networking opportunities
- Family Outreach Coordinator to assist families with access to clinics and networking
- Host conferences and medical symposiums
- Operate a Resource Centre
- Provide an information-based website and newsletter
- Provide all new parents/ guardians membership with a complimentary parent package

HOW YOU CAN HELP

- Join O.R.S.A.
- Make a donation or volunteer your time
- Attend or host a fundraising event
- Make others aware of O.R.S.A. and Rett syndrome
- Contact Us

CONTACT US: WWW.RETT.CA HELP IS AT YOUR FINGERTIPS

- Ontario Rett Syndrome Association
- P.O. Box 50030 London, ON N6A 6H8 T: (519) 474-6877
- Ontario Rett Syndrome Resource Centre T: (519) 850-RETT (7388)

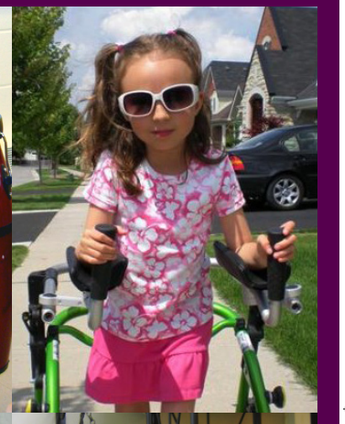
RETT SYNDROME

ORSA since 1991

Ontario Rett Syndrome Association
Building Healthy Tomorrows

Rett syndrome (RTT) is a unique developmental disorder that begins to show its affects in infancy or early childhood.

It is seen almost exclusively in females, and is found in a variety of racial and ethnic groups throughout the world.



Ontario Rett Syndrome Association
P.O. Box 50030 London, ON N6A 6H8
T: (519) 474-6877

Ontario Rett Syndrome
Resource Centre
T: (519) 850-RETT (7388)
www.rett.ca

RETT SYNDROME

www.rett.ca • (519) 474-6877

Charitable Donation #89017 8999RR0001

ENTERING A NEW WORLD

When a child is diagnosed with Rett syndrome, it can be very difficult for parents to accept. Families of newly-diagnosed children need time to grieve and sometimes a grief counsellor specializing in special needs is helpful. The Ontario Rett Syndrome Association (O.R.S.A.) is there to help you when your family is ready to move on. There are connections that can be made to other parents, to one of the Rett Syndrome Clinics in Ontario and to other supports. Information is available to you through O.R.S.A.'s resource centre, website and newsletter. The Association (O.R.S.A.) can provide you with a sense of belonging to an extended family that can help you manage through the difficult times. It is committed to support each family with up-to-date knowledge and new discoveries as they travel their journey with their loved one who has Rett syndrome.

WHAT IS RETT SYNDROME?

Rett syndrome (RTT) is a unique developmental disorder that begins to show its effects in infancy or early childhood. This disorder is seen in infancy and occurs almost exclusively in females. It is usually caused by a mutation of the MECP2 gene on the X chromosome. Rett syndrome is found in

all racial and ethnic groups throughout the world. It affects one in every ten thousand live female births. Early developmental milestones appear normal, but between 6-18 months of age, there is a delay or regression in development, particularly affecting speech, hand skills and gait. A hallmark of Rett syndrome is repetitive hand movements that may become almost constant while awake. Other more common medical issues encountered include epileptic seizures, muscle stiffness, osteoporosis and scoliosis. Despite its multiple handicaps, Rett syndrome is not a degenerative disease. Many individuals with Rett syndrome live long into adulthood. There is currently no cure.

DIAGNOSIS

Rett syndrome is diagnosed through a clinical examination that looks for specific signs and behaviours; it is confirmed through genetic testing. It can occur, however, without any of the currently known genetic mutations. As with any medical condition there is a large amount of variation in people with Rett syndrome. For example some girls learn to walk, although in an abnormal way, while others are never able to walk independently. There are two types of diagnoses to be made: typical or classic RTT and atypical or variant RTT.

Typical or classic RTT is characterized by a period of regression followed by recovery or stabilization. It requires the presence of 4 main criteria – partial or complete loss of acquired purposeful hand skills, partial or complete loss of acquired spoken language, gait abnormalities and stereotypical hand movements, and must meet 2 exclusion criteria – no brain injury secondary to trauma and no grossly abnormal psychomotor development in the first six months of life. Some other supportive criteria are often present in this type of RTT but they are not required for the diagnosis.

For Rett syndrome to be considered atypical or variant, a period of regression must be present followed by recovery or stabilization and 2 of the 4 main criteria above must be met as well as 5 of 11 supportive criteria. These supportive criteria are:

1. Breathing disturbances when awake
2. Teeth grinding when awake
3. Impaired sleep pattern
4. Abnormal muscle tone
5. Peripheral vasomotor disturbances
6. Curvature of the spine - scoliosis/kyphosis
7. Growth retardation
8. Small, cold hands and feet
9. Inappropriate laughing/screaming spells
10. Diminished response to pain
11. Intense eye communication - eye pointing

WHAT TO EXPECT

Even though there is not yet a cure for Rett syndrome, there are interventions that will make a significant difference. As Rett syndrome is not a degenerative disease, constant physical and mental stimulation will have a tangible impact, and it will prevent further physical deterioration due to non use. "Walk, walk, walk," Dr. Rett used to say. Each intervention will enhance the quality of life for those living with the condition.

Occupational, speech/ communication and physical therapies are essential supports. The therapists within these three services often work together to address issues and develop therapy plans. Aggressive therapy is always recommended and there may be a need for lifelong therapy support. A team approach including the family is the best practice when supporting a child or adult with RTT. At this point, led by therapists with expertise in augmentative communication, good results are being seen with the use of computers that respond to eye gaze and do not necessarily require hand skills.

Other effective therapy interventions often used are hydro, hippo, music and massage therapies. They provide support to occupational, physical and speech programs. Each has its benefits and should not be overlooked.

Children and adults with RTT can continue to learn and enjoy family and friends well into middle age and beyond. They experience a full range of emotions and show their engaging personalities as they take part in social, educational and recreational activities at home and in the community.

With Rett syndrome, lifelong learning is possible

RESEARCH

Canadian research into Rett syndrome is very well represented on the international stage. There are at least 20 different scientists or clinicians located in British Columbia, Alberta, Manitoba, Ontario and Quebec currently conducting projects relevant to Rett syndrome. In 2006, the Canadian Rett Syndrome Research Consortium was formed. As a result, there is a strong collaborative effort between Canadian researchers. In 2008, the first "team-based" research grant application was submitted to the Canadian government. Since its inception, O.R.S.A. has financially supported the Consortium. Over two decades, the Association has provided numerous individual research grants across Canada, as it is deeply committed to supporting and funding Canadian research into Rett syndrome.