Rett Syndrome - A Useful Summary

About the Syndrome

- Complex neurological disorder
- Affects mainly females, currently 6 boys in UK with a diagnosis.
- Genetic
- At least 1 in 12,000 females
- Statistically in the UK at least 2,500 have the disorder
- It's the most common genetic cause of profound & multiple learning disability in females

Diagnosis

- Through clinical observation and blood test
- Development stops from about 12 months, period of regression, skills in speech and hand movements reduce or are lost.

Common Features

- Breathing irregularities
- No verbal communication
- Epilepsy
- Muscles become rigid, joint deformities, muscle wasting
- Unsteady wide-based gait
- Crying / screaming spells
- Excessive drooling
- Aspiration pneumonia
- Development of scoliosis
- Impaired sleep pattern
- Cold, blue hands and feet
- Teeth grinding
- Difficulties with swallowing and chewing
- Gastric and oesophageal reflux
- Constipation

Genetics

- Mutation on the MECP2 gene on X chromosome
- This gene is responsible for turning off other genes when they are no longer needed in development.
- The mutation causes the turn-off mechanism to fail, allowing other genes to function abnormally.
- The connections (synapses) are then disrupted during brain development.
- Picture MECP2 gene as a book with 4 chapters.
- Mutations include having a missing page(s), extra page(s) or pages in wrong order.
- In some cases an entire chapter or two may be missing.
- Correct term for the chapters is exons.

Rett UK
Support today, hope for tomorrow

Langham House West, Mill Street, Luton LU1 2NA
Tel: 01582 798910 · Direct Support Line: 01582 798911 · Fax: 01582 724129
Email: info@rettuk.org · Website: www.rettuk.org