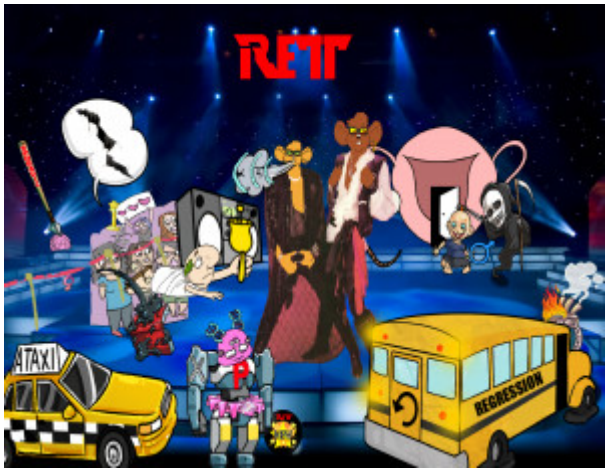
Rett Syndrome



PLAY PICMONIC

Pathogenesis

Sporadic Neurological Disorder
Sporadic-spear on Brain
Rett syndrome is a rare neurological disorder.The mutation present in this disease usually occurs sporadically, meaning that it's usually not inherited from a parent.

De Novo Mutation of MECP2 on X Chromosome
Mutant on a Mech-suit with Letter-P, a Two-tutu and an X-chrome
In Rett syndrome there's a mutation of the Methyl-CpG-binding protein 2 gene, or MECP2 gene, which codes for MECP2 protein. MECP2 protein is involved in forming neuronal connections and helps to silence or turn off other genes. When this protein is mutated, it can cause severe impairment.

RISK FACTORS & SYMPTOMS

Seen Almost Exclusively In Girls
Fan-girls
Females have 2 X chromosomes so one mutated MECP2 gene can be compensated for by a normal MECP2 gene on the other X chromosome.

Affected Males Die in Utero or Shortly After Birth
Baby with Male-symbol coming out of Uterus-door and Death-reaper
Males have only one X chromosome, so if there's a mutation in the MECP2 gene, then they cannot make functional MECP2 protein, and that might be why males with the mutation typically die in utero or shortly after birth.

Symptoms Usually Appear Between Ages 1-4
One-wand and Four-fork
Before 6 months of age, children with Rett syndrome develop normally, but starting around 6 months, children start to lose interest in play and no longer maintain eye contact. The second stage is the rapid deterioration stage and occurs between ages 1 to 4 years. In this stage there is a dramatic regression in speech, motor skills and intellect.

Developmental Regression
Developmental-bus backing up
Developmental regression is when a child loses an acquired function or fails to progress beyond a prolonged plateau after a period of relatively normal development. This begins to occur in patients with Rett syndrome during ages 1 to 4 years where they lose several skills.

Motor Impairment
Motor-on-fire
Rett syndrome patients present motor impairment due to motor skill regression. An example of this would be a child who used to grasp things with their hands may no longer be able to do that. Eventually muscles become weak or may become rigid or spastic with abnormal movement and positioning.

Intellectual Disability (Mental Retardation)

Tar Covered Autograph Book

Patients with Rett syndrome may also exhibit a decline in intellectual function. This deterioration can be rapid or gradual. Parents may notice a sudden change in their child’s behavior and health. Affected children may show diminished interest in people and objects.

Loss of Verbal Abilities

Broken-speech-bubble

Children with Rett syndrome typically begin to lose the ability to speak, to make eye contact and to communicate in other ways. Some children have rapid changes, such as a sudden loss of speech.

Seizure

Caesar

Most people who have Rett syndrome experience seizures at some time during their lives. Multiple seizure types may occur and are accompanied by an abnormal electroencephalogram (EEG).

Stereotyped Hand Wringing

Stereo and hand-ringing-bell

Children with Rett syndrome typically develop repetitive, purposeless hand movements, known as stereotypic movements. The most common stereotypic movement disorder seen in patients is hand wringing, which is the clasping together and squeezing of one's hands.

Decelerated Head Growth (Microcephaly)

Small-head

In patients with Rett Syndrome, their brain growth slows after birth. Smaller than normal head size (microcephaly) is usually the first sign that a child has Rett syndrome.

Breathing Problems

Abnormal-breath sound waves

Patients with Rett syndrome can display breathing problems such breath-holding, abnormally rapid breathing (hyperventilation), forceful exhalation of air or saliva, and swallowing air. These problems tend to occur during waking hours, but breathing disturbances such as shallow breathing or periodic breathing can occur during sleep.

Ataxia

A-taxi

Ataxia is a neurological sign consisting of lack of voluntary coordination of muscle movements. In patients with Rett syndrome, the first signs of ataxia often include reduced hand control and a decreasing ability to crawl or walk normally. At first, this loss of abilities occurs rapidly and then it continues more gradually. Eventually muscles become weak or may become rigid or spastic with abnormal movement and positioning.