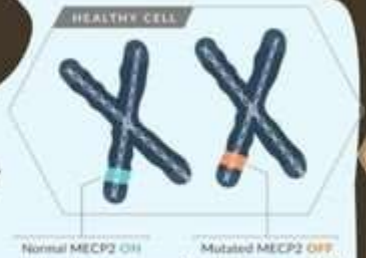
RETT SYNDROME

PRESENTED BY,

GAYATHRI.N.S

- Neurodevelopment disorder
- appearing in girls during infancy and early childhood
- mutations in the methyl-CpG-binding protein 2 (MECP2) gene
- lose skills such as the ability to crawl, walk, communicate or use their hands

Genetic Basis



- It is an x-linked dominant genetic disorder
- It is largely sporadic
- More than 95% of females meeting RS consensus criteria will have mutations
- In MECP2. MECP2, located at xq28
- The mutation appears to result in problems with the protein production critical for brain development.

Symptoms

- Microcephaly
- Loss of normal movement and coordination
- Loss of communication abilities
- Abnormal hand movements
- Unusual eye movements
- Breathing problems
- Cognitive disabilities
- Seizures
- Abnormal curvature of the spine
- Irregular heartbeat
- Pain

Stage 1: early onset.

Stage 2: rapid destruction

Stages of Rett syndrome

Stage 3: plateau

Stage 4: late motor deterioration

Stage 1

- early onset.
- starts between 6 and 18 months of age
- can last for a few months or a year.
- Babies show less eye contact and start to lose interest in toys.
- They may also have delays in sitting or crawling.

Stage 2

- rapid destruction
- Starting between 1 and 4 years of age, children lose the ability to perform skills they had earlier.
- Symptoms occur, such as slowed head growth, abnormal hand movements, hyperventilating, screaming or crying, problems with movement and coordination, and a loss of social interaction and communication.

Stage 3

- Between the ages of 2 and 10 years and can last for many years.
- Problems with movement continue, behavior may have limited improvement, with less crying and irritability, and some improvement in hand use and communication.
- Seizures may begin

Stage 4

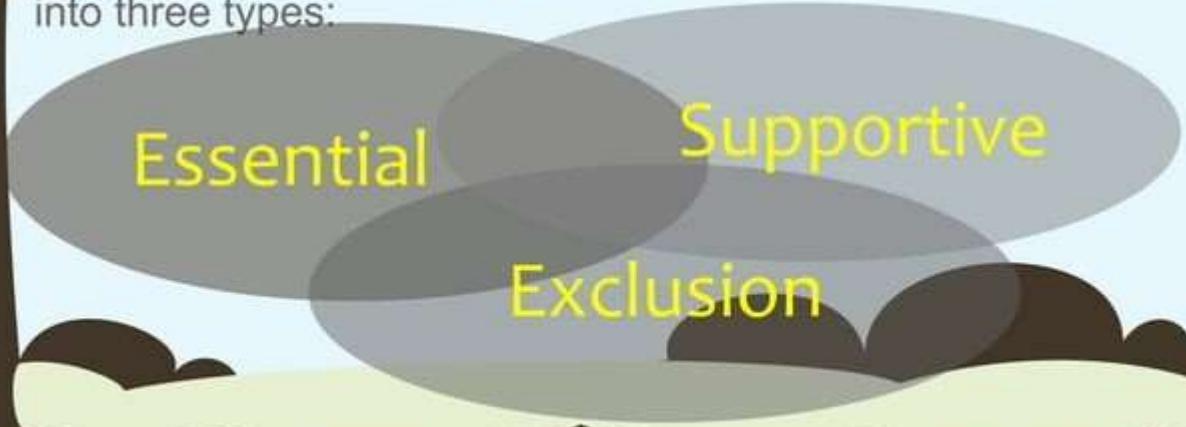
- begins after the age of 10 and can last for years or decades.
- It's marked by reduced mobility, muscle weakness, joint contractures and scoliosis.
- Understanding, communication and hand skills generally remain stable or improve slightly, and seizures may occur less often.

Diagnosis

- Physical exam and detailed information about the child's development and medical history.
- Genetic DNA blood test to support the diagnosis
- CT scan, MRI scan
- Electroencephalograms

Criteria for diagnosis

The guidelines for diagnosing Rett syndrome are divided into three types:



Essential criteria

- A period of normal development until between 6 to 18 months followed by a loss of skills, then recovery or stabilization of skills
- Partial or complete loss of purposeful hand skills
- Partial or complete loss of spoken language
- Dyspraxic gait
- Repetitive hand

Supportive criteria

- breathing irregularities while awake, such as apnea, hyperventilation and air swallowing
- teeth-grinding
- abnormal sleep patterns
- abnormal muscle tone (hypotonia, rigidity or spasticity)

- scoliosis or kyphosis (curvature of the spine)
- delayed growth
- small hands and feet
- inappropriate laughing or screaming spells
- reduced response to pain
- intense eye communication or “eye pointing”
- poor circulation in the hands and feet, with cold and bluish to red hands and feet

Exclusion criteria

- a neurometabolic disease or other inherited degenerative disorder
- a neurological disorder resulting from severe infection or head trauma
- evidence of brain damage acquired after birth
- grossly abnormal development in the first six months of life

Treatment

There is no currently targeted treatment or gene therapy

- Physical therapy
- Occupational therapy
- Speech-language therapy
- Nutritional support
- Behavioral intervention
- Support services

Supportive care


OCTOBER IS RETT SYNDROME AWARENESS MONTH
These supergirls have RETT SYNDROME, a severe movement disorder that has taken away their ability to speak, use their hands and in most cases, walk independently.
They also have patience, bravery, beauty, love, strength and a whole lot of awesomeness!
Rett Syndrome Awareness
Cure Rett Syndrome
cure rett