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# Rett Syndrome -A case study of Malayalam Speaking Child with an Autism Spectrum Disorder

Supritha Aithal, Manjunath Y.N., and Shyamala K.C., Ph.D.

There is no enigma like the mind. The normal mind functions at the speed of light, in a multitude of directions, and yet, there are certain shadows that are never dissolved by light, and remain a deep mystery, as when presented by a disordered human mind.

In this paper, an attempt is made to present the case study of a child with Rett syndrome/disorder, a sub group of the pervasive developmental disorder.

#### **Definition of Pervasive Developmental Disorders (DSM-IV-RT, 2000)**

Pervasive developmental disorders (PDDs), also called autism spectrum disorders (ASD) are characterized by dysfunction in three core areas of early childhood development namely communication and language skills, social interaction, behavior, specifically by the presence of stereotyped, repetitive behaviors and restricted activities and interests.

Diagnostic and Statistical Manual for Mental Disorders (DSM-IV-RT, 2000) Classification System of Autism Spectrum Disorders

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The Autism spectrum Disorders is an umbrella term under which there are group of disorders such as

- Autism
- Asperger's syndrome
- Childhood disintegrative disorder (CDD)
- Rett syndrome
- Pervasive developmental disorder not otherwise specified (PDD-NOS)

Out of all these disorders the specific symptoms, assessment and management of the rare condition "Rett disorder" is the highlight of this paper.

# **Objectives**

- 1. To highlight the specific symptoms of the rare condition "Rett disorder" among the Autism Spectrum Disorders.
- 2. Get to know in detail the crucial manifestations, neuropathology, differential diagnosis, assessment and management aspects of Rett disorder.

# **Definition of Rett Syndrome**

Rett syndrome is a neurodevelopmental disorder caused by X-linked genetic mutations that occur almost exclusively in females (Amir et al., 1999). It was originally described by Austrian pediatrician named Andreas Rett in 1966.

#### **Incidence**

The incidence of the disorder is about 1:10,000-20,000 (Percy, 2001). Indian literature quotes that the incidence in India is about 1 in 10,000 to 22,000 live births (Sitholey, Agarwal & Srivastava, 2005)

#### **Causes**

Rett syndrome is caused by mutations in the gene MECP2 located on the X chromosome. And these mutations can be of two types:

- 1) Sporadic Mutations
- 2) Germline Mutations

# **Sporadic Mutations**

Sporadic mutations are usually caused by a de novo mutation which are not inherited from either of the parent. They occur in 95% of the rett syndrome children. Parents are generally genotypically normal, without a MECP2 (Methyl-CpG-binding protein-2) mutation. Mutated

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MECP2 is usually derived from the male copy of the X chromosome. It is not yet known what causes the sperm to mutate. Such mutations are rare.

#### **Germline Mutations**

Rett disorder can also be inherited from phenotypically normal mothers who have a germline mutation in the gene encoding methyl-CpG-binding protein-2. MECP2 is found near the end of the long arm of the X chromosome at Xq28.

# **Atypical Rett Syndrome**

An atypical form of Rett syndrome, characterized by infantile spasms or early onset epilepsy, can also be caused by a mutation to the gene encoding cyclin-dependent kinase-like 5 (CDLK5). These individuals have generally tested negative for a MECP2 mutation. Not everyone with a CDKL5 mutation appears as atypical RTT. Other CDKL5 mutation disorders include Infantile Spasms, West Syndrome, Early Onset Seizures, and Autism.

# **Mortality**

Males with pathogenic MECP2 mutations usually die within the first 2 years from severe encephalopathy, unless they have an extra X chromosome (Klinefelter syndrome), or have somatic mosaicism. Females with Rett syndrome can live up to 40 years or more.

#### **Case Details**

Case number: 230795
Age/Gender: 4yrs/female
Language: Malayalam
Date of birth: 28.04.06
Date of report: 23.10.2009

#### Brief history of the child (23.10.2009)

The child was brought to AIISH by her parents with the complaint of poor speech and language skills. They reported that child appeared to be developing normally till 8 months of age. She showed regression soon after, in motor, speech and language skills.

#### Duration

According to DSM IV criteria there should be apparently normal psychomotor development at least through the first five months after birth in children with Rett syndrome. In the client regression was noticed after the age of 8 months.

#### **Nature and Onset:**

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Literature on the onset and the nature of the Rett disorder is progressive and gradual. In the child the condition is worsening and gradual.

## **Developmental History**

Speech milestones of the child were

• Babbling : 6months

First word:1 year /amma/First sentence :Not achieved

Motor Milestones of the child were

• Neck control: 3-4 months

• Turn over: 6 months but lost the turn over skill at 8 months

• Sitting: > 2 yrs after the physiotherapy and she lost the skill after discontinuing the therapy.

# **Medical History**

According to DSM IV TR (2000) main criteria for Rett disorder should be apparent normal prenatal and perinatal development.

In the child, the medical history as reported by the parents included the following information on:

Pre natal period – There was no significant history

Peri natal period - Full term normal delivery.

Birth weight: 3.5 kg Birth cry: Normal

Post natal period- EEG abnormalities were found but without apparent seizures. She was under medication to avoid seizures.

#### Family history

Literature suggests that more than 99% of cases occur among people with no family history. Rest 1% of cases have X linked dominant pattern of inheritance. The child is from joint family, and she has a normal elder sister and consanguinity was reported to be negative and no other family history was reported by the parents.

#### **Earlier investigations**

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The previous investigations were done at department of neurological sciences in Baby memorial hospital at Calicut on 26.12.2007 when the child was one and half years old. Digital Electroencephalography (DEEG) was done. The diagnosis made was "? Seizure."

Magnetic resonance Imaging (MRI) was done at Dr. Shaji's MRI Center on 31.12.2007 and the impression made was Bilateral Hippocampal Sclerosis.

## Recommendations made after the general history taking included the following:

- Detailed Speech and Language Evaluation
- Autism Spectrum Disorders unit (ASD) Evaluation
- Clinical Psychology Evaluation
- Physiotherapy /Occupational therapy Evaluation
- Neurological Evaluation
- Pediatric Evaluation
- Counseling
- Follow up

# **Speech and Language evaluation**

#### First evaluation was done on 23.10.2009

Tests administered were

a) Receptive Expressive Emergent Scales (REELS) – by Bzoch and League (1989) adapted by Madhu, Deepa, Suhas, Harshan Kumar & Shyamala (2009).

Receptive Language age (RLA) was 8-9 months

Expressive language age (ELA) was 4-5 months

Delay in both receptive and expressive language was seen in the child.

b) Modified Checklist for Autistic toddlers (M-CHAT)- by Robins, Fein, & Barton (1999)

It is a screening tool which can be administered to assess the children at risk for Autism Spectrum Disorders (ASD). It includes 23-items which are in a question format. There are six critical items which are behaviors that are majorly found in at risk children for ASD. Children who fail in two or more critical items has to be referred for detailed evaluation. M-CHAT was administered in the present child. She failed in all the critical items (2, 7, 9, 13, 14, and 15) which includes Q2 (Does your child take an interest in other children?), Q7 (Does your child ever use his/her index finger to point, to indicate interest in something?), Q9 (Does your child ever bring objects over to you (parent) to show you something?) ,Q 13 (Does your child imitate you?) ,Q14 (Does your child respond to his/her name when you call?),Q 15 (If you point at a toy across the room, does your child look at it?).

Recent evaluation was done on 10.08.2010

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Only REELS was administered and the scores were Receptive Language age (RLA) was 11-12 months Expressive language age (ELA) was 8-9 months

Provisional diagnosis made was Delayed Speech and language with Rett syndrome

Table 1: Criteria for Differential Diagnosis of Autism spectrum Disorders (Wetherby & Prizant, 2001)

	Autism	Asperger's	Rett	CDD	PDD-NOS
Onset/ Course	Prior to 3 yrs	Onset may be before or after 3 yrs *	Onset prior to 3 yrs. Normal development till 6 –18 months	Onset after 3 – 4 yrs of normal development **	May fail to meet autistic criteria
Delay	May or may not be associated with general delays	No general delay in cognition and language.  Motor clumsiness may be present **	Usually associated with mental retardation *	Usually associated with mental retardation*	May or may not be associated with developmental delays
Severity	Exceed standard threshold of a number of features	Most exceed threshold in social area *	Exceed standard threshold of a number of features	Exceed standard threshold of a number of features	May fall below threshold in 1 or more areas *
Domains affected	Social Communication Repetitive behaviors	Social Circumscribed interests *	Social Communication Repetitive behaviors (lesser than autism) *	2/ 3 domains of autism Social Communication	Social Communication Repetitive behaviors (lesser than autism) *

<sup>\*-</sup> May differ from Autism, \*\*- Always differs from Autism

Table 2: Differential Diagnosis of Rett syndrome from other similar syndromes

Characteristics	Rett Syndrome	Fragile X Syndrome	Angelman Syndrome
Genetic Mutations	MECP2 GENE,	FMR-1 gene	Deletion of long arm

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	CDKL 5 gene	Weak link of X chromosome	of chromosome 15(15q11-15q13)
Gender	Exclusively in females	In both males and females	In both males and females
Epidemiology	1-10,000 to 22,000	1-1,000(males) 1-2,000(females)	1 in 12,000 to 20,000
Dysmorphic features	Mild microcephaly only in few cases.	Long face, prognathism, macrocephaly	Triangular face, prognathism, macrostomia, widely spaced teeth, protruding tongue, and hypopigmentation.
Cognition	Severe cognitive impairment	Varies from mild to severe cognitive impairment	Mild cognitive impairment
Communication	Severe speech impairment, Receptive and nonverbal communication skills higher than verbal, Apraxia of speech.	Delayed speech milestones, Articulation difficulty, Alternations in rhythm and fluency, Repetitive speech (palilalia)	Severe speech impairment, Receptive and nonverbal communication skills higher than verbal

# **Autism Spectrum Disorder (ASD) Evaluation**

Tests administered were

# a) Differential Diagnostic Checklist for Autism Spectrum Disorders - DDC-ASD by Shyamala, Vijayashree, Sujatha&Rajkumar (2007).

This test was mainly used to differentially diagnose the autism spectrum disorders. In this test Part-B was administered which includes diagnostic criteria for rett syndrome. This section consists of

- Gender-female
- Normal development till 5 months followed by regression

This checklist assessed majorly three domains:-

1) Motor - Deterioration was seen in the child

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- 2) Behavior Loss of acquired previous hand skills and stereotyped hand movements e.g. hand wringing and hand washing and mouthing
- 3) Social Withdrawal from the surroundings, absence of reciprocal interaction and poor adaptive skills

The other associated problems present in the child were

- Abnormalities in gait (unsteady, scissored gait, wide based, stiff and toe walking)
- Reduced muscle tone and hypotonia.
- Reduced body fat and muscle mass (power or strength)
- Decelerated head growth after a period of normal development.
- Exhibits severely delayed intellectual development or profound mental retardation noticed from 5 months with severe learning disabilities

# b) Childhood Autism Rating Scale (CARS) - by Schopler, Reichier, and Renner (1988)

The Childhood Autism Rating Scale (CARS) is a behavioral rating scale used to identify children with characteristics of Autism and differentiate them developmentally delayed children who don't have autism features. And CARS can be administered to any child who is above two years of age. It consists of 15 items and which are rated on a 7 point-rating scale. Here each item represents a particular ability or characteristic or the behavior of children with autism. CARS can distinguish the severity of autism from mild to severe degree depending on the total scores obtained.

The various domains which were assessed in child are

- 1) Relating to people
- 2) Imitation
- 3) Emotional responses
- 4) Body use
- 5) Object use
- 6) Adaptation to change
- 7) Visual response
- 8) Listening responses
- 9) Taste, smell and touch response and use
- 10) Fear or nervousness
- 11) Verbal communication.
- 12) Non verbal communication.
- 13) Activity level
- 14) Level and consistency of intellectual response.
- 15) General impression

Total score was: 43/60

Impression made was Severe Autism.

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#### Other Evaluations

# **Clinical Psychology Evaluation**

# First Evaluation was done on 23.10.2009

Test administered was developmental screening test

Developmental Age (DA) was 8-9 months

Developmental Quotient was 21

Diagnostic formulation made was severe developmental delay

#### Recent Evaluation was done on 10.08.2010

Test administered was Vineland Social maturity Scale

Social age (S.A) was 7 months

Diagnostic formulation made was

Expressive language Disorder, developmental delay type,

Profound grade M.R (I.Q < 20) and poor psychomotor coordination

#### PT/ OT Evaluation

Impression made was Developmental delay with sensory issues and recommended for Developmental therapy.

#### **Neurological Evaluation**

Impression: Rett syndrome

#### **Pediatric Evaluation**

Head circumference of the child was 46 cm which was less than that of normative.

Tone: Hypotonia in the lower limbs power is 3/5.

Impression was Rett syndrome.

After all the evaluations the provisional diagnosis made was Developmental delay with Rett syndrome.

Other characteristic features of the Rett syndrome present in the child was

- 1) Hypotonia( usually first to appear)
- 2) Deceleration of head growth
- 3) Breathing problems

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- 4) Severe speech and language impairment
- 5) Loss of purposeful hand movements, replaced by stereotyped hand movements
- 6) Loss of social engagement
- 7) Sleep disturbances.
- 8) Bruxism (grinding of teeth)
- 9) Poor circulation that can lead to cold and bluish arms and legs
- 10) Gastrointestinal problems like constipation and GERD
- 11) Frequent Seizures
- 12) Reduced eye contact and lack of interest in play

Other characteristic features of the Rett syndrome that are not present in the child but can occur at the latter stages of the disorder are

- 1) Shaky, unsteady, or stiff gait; or toe walking
- 2) Apraxia.
- 3) Ataxia
- 4) Scoliosis
- 5) Excessive saliva and drooling
- 6) Anxiety, panic, occasionally self –injury

# **Neuropathology in Rett syndrome:**

- 1) **Brain weight reduction**: MECP2 is responsible for the production of a type of Glial cells. One of the types of glial cells are asterocytes which contribute to the majority of the brain weight. When there is a mutation in the MECP2 gene then there is reduction in the glial cells which in turn reduces the brain weight.
- 2) Reduction in brain weight is not generalized because the cerebral hemispheres are affected more than the cerebellum in Rett disorder.
- 3) **Brain volume** is affected in the brain areas such as prefrontal area, posterior frontal area and anterior temporal regions.
- 4) Normal brain volume is preserved in posterior temporal area and occipital area.
- 5) Decreased pigmentation in subtantia nigra and pars compacta can be seen.
- 6) Abnormalities in neurotransmitters like acetylcholine, dopamine, serotonin, glutamate substance p production and new nerve growth factor can also be noticed.
- 7) Decreased dendritic territories in regions like cortex, thalamus, basal ganglia amygdala and hippocampus( decreased neuronal size and increased density)
- 8) Neuronal loss at anterior horn and gliosis in cortico spinal tracts of spinal cord.
- 9) Breathing irregularities, heart rate variability, cold feet, constipation and swallowing suggests autonomic impairment in Rett disorder.

#### Management

Currently, there are no readily available therapies that radically change the course and evolution of Rett Syndrome (Zwaigenbaum & Szatmari, 1999).

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#### **Aims of the Intervention:**

- 1) Preserving physical and psychosocial functioning in children with rett syndrome
- 2) Enhancing quality of life.
- 3) Providing education and support to families.

#### **Team Members**

The team members include various medical and nonmedical professionals who help in the intervention of children with Rett syndrome.

- Speech language pathologist
- Psychologist
- Family members
- Special-education teachers
- Orthopedic surgeons
- Physio / occupational therapist
- Nurse
- Neurologist
- Pediatrician
- Cardiologist
- Pulmonologist
- Gastro enterologist.

Following are a few management approaches general to all the autism spectrum disorders including Rett disorder.

# **Medical Management**

#### a) Drug therapy

The drug therapy helps to improve the various characteristic features of rett syndrome. They are as follows:

- 1) Poor sleep patterns
- 2) Aggression towards others
- 3) Chronic constipation
- 4) Anxiety disorders
- 5) Self injurious behaviors
- 6) Depression
- 7) Hyperactivity

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# b) Vitamin and minerals therapy (Pimland, 1989)

High dose of vitamin B6 with Magnesium is given to children with Autism Spectrum Disorder. The autism research institute proposed that many studies have shown that vitamin B6 and magnesium supplements have a good rate of success by improving the ability to relate and communicate.

# c) Gene therapy

It is the insertion, alteration, or removal of genes within an individual's cells and biological tissues to treat disease. Here insertion of functional genes into an unspecified genomic location in order to replace a mutated gene is carried out. But this technology is still in its infancy.

# **Non-Medical Management**

It deals with communication education and overall rehabilitation.

# a) Speech Language and Communication Therapy

It helps the children with autism spectrum disorders to communicate verbally or nonverbally. There are several approaches that are in practice. Some of them are mentioned here.

- i) Lovaas method/Applied Behavior Analysis (ABA), 1987: It is a form of applied behavioral analysis (ABA) in which the trainer models to the child a skill to be mastered and then encourages the child to replicate.
  - Here the principles of operant conditioning are used to teach language. Very popular with early childhood educators.
  - It uses careful behavioral observation and positive reinforcement or prompting to teach each step of a behavior.
  - A child's behavior is reinforced with a reward when he or she performs each of the steps correctly.

# ii) Treatment and education of autistic and related communication handicapped children (TEACCH) - Schopler (1970)

• This was founded by Dr. Eric Schopler in the early seventies. The main purpose of the curriculum is to teach the child with autism communication skills versus speech skills. The main emphasis is on remodeling the environment to accommodate difficulties faced by the children with ASD.

# iii) Play therapy - Hickman (1997)

Play therapy is an intervention method to promote skills designed to facilitate positive social interactions, either verbal/nonverbal.

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• Play designs allow the clinician to structure activities to accommodate the child's level of functioning and create unique opportunities for new skills (Hickman, 1997).

# iv) Option method/Son-rise program - Kauffman (1970).

- It's a home based, child centered approach. Encourages the parents to participate with their child to increase the child's motivation.
- The philosophy behind treatment is that we have to accept the child with autism
  rather than change the child to suit us, second the child is always given options or
  choices so that some amount of control is given to the child rather than being
  controlled.

#### v) Augmentative and Alternative communication - Sigafoos & Woodyatt, (1996)

- This method relies on the comprehension abilities of the child. It helps to improve communication and stimulate interaction. Commonly used methods are:
- Picture Exchange Communication System (PECS)- Bondy and Frost (1994)
- Visually mediated communication Hogdon (1999). Depending on the child's need, the techniques can be selected.

# b) Physical therapy and occupational therapy:

- This is required to develop, maintain and restore maximum movement and functional ability.
- Splints and braces can be given to the child to assist coordination and balance.
- This is also useful to increase purposeful use of hands in daily life activities.
- Sensory integration therapy helps them process what they see, hear, taste, smell and touch.
  - Following are all alternative and supportive therapies that can be used for children with ASD.

## c) Hydro therapy (Bumin, Uyanik, Yilmaz, Kayihan, & Topcu, 2003).

• Hydrotherapy can be used to improve gross motor abilities especially spontaneous movements and also for muscle relaxation and it decreases the body tension.

# d) Hippotherapy

• Use of movement of the horse as a treatment strategy in physical occupational therapy sessions and this improves gait and emotional well being of the client.

# e) Music therapy - Yasuhara & Sugiyama (2001)

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• Music therapy uses music to improve learning, build self-esteem and reduce anxiety or any of a number of treatment goals that are specific to the patient.

# f) Yoga and meditation

• This is used with children with ASD and have been found effective in calming and destressing.

# Speech and language therapy given to the child:

The goals taken up for the child based on the baseline (see table 3) and its progress are tabulated below.

Table 3: The goals taken up for the speech language therapy and the progress in the child

Goals	Progress
<ul> <li>To improve the functional communication</li> <li>To improve the concept of self and family members</li> </ul>	To indicate yes smile and to indicate no she will turn her face.     Says /ta/ inconsistently
<ul> <li>Response to name call.</li> <li>To improve the child's ability to comprehend polar questions</li> </ul>	Says /amma/ /appa/ /akka/ meaningfully.
<ul> <li>To improve the comprehension and expression of food items, body parts and common objects</li> </ul>	Says /bibi/ for biscuits, /mika/ and /vava/ for doll inconsistently

Table 4: The speech and language abilities of the child with Rett syndrome

Speech and Language abilities	Verbal		Non verbal
Comprehension	General	Malayalam words in IPA	Attends to sounds easily. Example: children's
	Responds to name call. /bubLu/ Shows inconsistent responses to short commands. Identifies few objects like		cry. Attracted to television advertisements.

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	fan, light, switch, glass,		
	phone, book, cycle,		
	fruits like banana — /palam/		
	flower /pu:vu:/		
	crow	/ka:ka/	
	cup	/kappu:/	
	doll →	/mikki//va	n:va/
	Comprehends some		
	action verbs like	verbs like	
	eating /tinnunu/		/
	sleeping —	/uraŋunnu	/
Expression	Differential cry and vocalizations		Smiles to indicate 'yes'
	present.		and turn her face to
	Says /ta/ inconsistently.		indicate 'no'.
	Says /amma/ for mother, /appa/ for		
	father, /akka/ for sister (actual		
	name /akhila/) meaningfully.		
	Says /bibi/ for biscuits, /mika/ and		
	/va:va/ for doll inconsistently.		

# **Summary**

- The manifestations are highly variable with this genetic syndrome
- This child shows majority of features of the Rett syndrome reported in the literature.
- Malayalam language skills in the present child indicate better comprehension than expression; though expression is also limited to a few words.
- Although poor prognosis is indicated in literature, rigorous and continuous developmental therapy is recommended in all domains of motor, speech/language/communication as well as other behaviors as she has shown slow but steady improvement.

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Supritha Aithal

III rd B.Sc (Speech and Hearing) Student All India Institute of Speech and Hearing Manasagangotri Mysore-570006 Karnataka, India supritha.aithal@gmail.com

Manjunath Y.N III <sup>rd</sup> B.Sc (Speech and Hearing) Student All India Institute of Speech and Hearing Manasagangotri Mysore 570006 Karnataka, India nkpamanju@gmail.com

Shyamala.K.C., Ph.D.

Professor in Language Pathology & OC-DHLS Department of Speech Language Pathology

Language in India www.languageinindia.com

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Supritha Aithal, Manjunath Y.N., and Shyamala K.C., Ph.D.

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All India Institute of Speech and Hearing Manasagangotri Mysore570006 Karnataka, India Shyamalakc@yahoo.com