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A Case Study on Management of Rett Syndrome by Wholistic approach

Case Report

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Abstract

Background: Rett syndrome-RS comes under Autism spectrum disorder-ASD which is a neurodevelopmental syndrome. It is diagnosed by the main differentiating features of lack of interpersonal and communication skills, poor eye contact, delayed speech with pervasive abnormal body movements. Aim and Objectives: This case report is aimed at dissemination of comprehensive role of Ayurveda in management of ASD, Rett syndrome. Material and Methods: RS is the severe form of ASD. This case study of 2.3 year's girl presented with RS and global delay, being treated with wholistic approach. It comprises Ayurveda chikitsa and other therapies like Yoga, hydrotherapy, occupational, music, physiotherapy and many more. Observation and Result: Patient has shown promising results in all developmental milestones such as gross motor, fine motor and personal social in 6 months duration except language. Different varieties of massage therapy, diet and Basti, Nasya (Panchkarma) procedures, Omkar mantra chanting, passive Yogasana were done. Conclusion: In this case report, mainly Ayurveda interventions were implemented with wholistic approach as an adjuvant, received good result in gross motor development which is very difficult in RS, hence it is a unique case. It also opened the door of wholistic approach with the hope to deliver the good result in similar disorders.

Key Words: Autism spectrum disorder-ASD, Developmental and Speech delay, Rett syndrome, Wholistic approach, Other therapies, Panchakarama, Yoga, Communication.

Introduction

Rett syndrome (RS) comes under the classification of Autism spectrum disorders. It is a X linked dominant, progressive neurodevelopmental disorder, prevalence of 1:10,000 ratio, resulting from genetic mutation of the MECP2 gene. (1) ASDs are increasing drastically, may be due to adoption of western lifestyle, stress, prenatal maternal infections/ complications, faulty diet and daily regimen.(2) The definite cause is not known but many environmental predisposing factors are responsible for the occurrence of this disease. It can be correlated with *Unmad* as per Avurveda to some extent as *Vata* is the controller of mind and in both the diseases, vitiation of mind is common. Unmad is an umbrella term for every defective psyche, described in Brihattrayee-Ayurved classics in detail. Charak has described five types-Vataj, Pittaj, Kaphaj, Sannipataj and Agaantuj Unmad. (3) RS is a severe form of ASDs. The clinical features include many complex symptoms such as behavioral problems, screaming, developmental delay, mood,

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anxiety disorder, obsessive-compulsive disorder, gastrointestinal upset, jadatva/mental retardness, phobia, sleeplessness etc. The disease starts with developmental delay and the symptom of not responding to family members or peers (social isolation). Patient may feel comfort in awkward posture for long time with stereotype hand writhing movements. (4) Autistic Spectrum disorder- ASDs can be classified as Asperger syndrome, Rett syndrome, Pervasive developmental disorder-PDD, Childhood disintegrative disorder-CDD. (5) Necessary investigations may help to rule out hidden associated features and differential diagnosis, such as Karyotyping, DNA analysis, Pet scan, STARCH test (Sexually Transmitted Disease, Toxoplasmosis, AIDS, Rubella, Cytomegalo and Herpes infection), thyroid profile, CBC, EEG, IQ test etc. DSM-V diagnostic criteria is helpful in making the diagnosis of ASD, mainly based on typical clinical features, examination and history. (6) Samprapti - the probable etio-pathogenesis is based on defective organogenesis; brain damage, metabolic changes in intra-uterine life may be due to genetic predisposition, radiation, dushivisha sevan and other known or unknown reasons. It is a Tridoshaj vyadhi with variation in their dominance such as Kaphaj, Vataj, Pittaj or Dwidoshaj or Sannipataj category (7). In contemporary science, the available management is based on supportive and symptomatic treatment modalities such as physiotherapy, occupational, speech therapy, behavioral and play therapy with need-based medicines. (8)

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Aim & Objective

Exposition of role of Ayurveda interventions with wholistic approach in the management of RS.

Case Report

Parents approached with a 2.3 years old girl child with the complaints of delayed milestones such as unable to recognize parents, drooling of saliva, speech delay since age of 6-7 months; unable to sit, stand & walk without support with repeated abnormal movements. She had lack of appetite, irregular bowel with hard stool, poor eye contact and bilateral squint. Child had poor social smile and was not sitting till 14 months of age also. After physiotherapy, child was able to sit with support at the age of 18-20 months momentarily but no improvement was seen in other milestones. Parents underwent treatment from many hospitals but no satisfactory result was seen. In birth history, mother was having history of stress at work place and positive TORCH infection (Cytomegalo, Rubella) before conception with non-consanguineous history. In Natal history, Full Term Vaginal Delivery, Birth weight 2.200 kg, cried soon after birth. In Post Natal history, she had given NICU care just after the delivery for observation due to peripheral cyanosis. She had received all vaccines as per routine and optional schedule. Family history was insignificant. Her sleep was disturbed with teeth grinding habit. The developmental history is delayed, reflected in table no. 1.

Table no 1: Showing developmental history at a

	glance in pre and post treatment (t/t)						
Sl. No	Param eters	Milestones	Achievement				
1	Gross Motor	Neck holding	Achieved at 6 months				
		Sitting with support	Achieved at 20 months with forward bending, post treatment sitting without support for long period also				
		Standing, walking with support	Post t/t achieved standing and walking for few steps with support only.				
2 Fine motor		Pincer grasp	Not achieved yet				
		Reaching & holding object	Achieved post t/t with palmer grasp				
3	Langu age	Speech	Delayed, post t/t -loud screaming, able to utter 2 monosyllables post t/t which is appreciable by parents.				
4	Person al- social	Social smile & recognizing relatives	Post t/t achieved it with much understanding & thoughtful but not expresses properly				
		Eye contact	Moody, achieved momentarily after t/t				

Figure No. 01: Showing abnormal writhing movements of extremities, Figure no-02 showing drooling of saliva and sitting with support posture. (Before treatment)





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Figure No-01

Figure No-02

On examination, pre t/t, she was prone to develop recurrent mild respiratory infections, but respiratory, cardiovascular system was not observed much derangements. During t/t, no respiratory infection occurred till 6 months and Post t/t also no episode of any illness was noted till 8 more months. There was sluggish peristalsis hence severe constipation, lack of appetite with tendency to keep oral medicines, semisolid food bolus in the mouth for long time. Sometimes, when in mood she used to consume her favorite mashed food at average speed without difficulty in deglutition. She remained non-active with her awkward posture of all extremities writhing movements for long period without trying to communicate with others. Her general and systemic examination is depicted in table no. 2

Table no 2: Showing the general & systemic examinations findings of Rett Syndrome on

admission			
General Examination			
Built: -Lean			
Appearance: -Lethargic			
Gait: -Scissoring			
Eyes: Pallor +			
Tongue: Coated			
Pulse rate:116/min			
RR: 24-26/min			
BP : 90/60mmHg			
Temperature: Normal,			
Afebrile			
Systemic Examination			
CNS-			
Conscious, but not oriented about time and place.			
♦ Reflexes bilateral-			
*			

- - 1. Planter- flexion
 - 2. Knee jerk- sluggish
 - 3. Ankle jerk-sluggish
 - 4. Biceps& Triceps- normal
- Muscle power- Grade 3/5 of B/L Upper and lower limbs
- Muscle tone- Hypotonic
- CVS- S1 S2 Audible, no adventitious sound
- **RS-** AE=BS, Chest clear
- **P/A-** Soft, No distention or organomegaly



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Central nervous and musculoskeletal systems were grossly affected. Severity was assessed with GMFCS-(Gross motor function classification system). Her head holding and sitting was in 2nd grade, standing-0 grade, fine motor grade was 0, personal-social grade was 1, Ashworth scale -0, Activities of daily living-

ADL score was 0 at the time of admission. Post treatment after 3rd sitting, GMFCS grade of sitting -5, standing, fine motor and Ashworth scale grade-1, Personal social and ADL score achieved 2. The anthropometry on pre-post admission is reflected in table no. 3

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Table no 3: Showing the Anthropometry on pre and post treatment

<u> </u>	<u> </u>
On admission: -	After 4 months & 3rd sitting of treatment: -
• Weight: 9.5 kg	• Weight: 10.7 kg
Height: 87 cm	Height: 90 cm
 Head Circumference: 44.5 cm 	 Head Circumference: 45.1 cm
 Mid Arm Circumference: 14 cm 	 Mid Arm Circumference: 15 cm
Chest Circumference: 47 cm	Chest Circumference: 48 cm
• BMI-12.55	• BMI-12.85
 PEM Grade-1 	• PEM Grade-1

Post treatment, constipation was relieved by *Basti* and there was no change in other factors. Table no. 4 is showing details of pre-post treatment status of *Ashtavidha pariksha*.

Table no 4: showing Ashtavidha pariksha on pre-post treatment

	1	Nadi	Kaphaj-Hansa gati	5	Shabda (Speech)	Cooing, screaming
	2	Mala(stool)	Once in 3-6 days interval	6	Sparsha (touch)	Sukhoshna/normal
			Post t/t-relieved constipation			
Ī	3	Mutra (urine)	8-10 times/day,1-2/night	7	Druka (vision)	Squint, no eye contact
	4	Jivha (tongue)	Coated	8	Akruti (built)	<i>Krish/</i> thin built

Post treatment, constipation was relieved with no coating on tongue but the intake of food and medicine was remained difficult, loud screaming started.

DSM-V and CARS score pre-post treatment was shown in Table no. 5

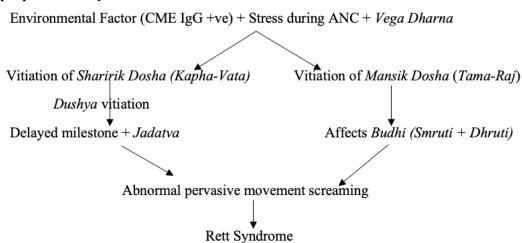
Table no. 5 depicting pre-post score of DSM-V and CARS in this RS case report

S.No	Assessment criteria of DSMV & CARS	Pre t/t score	Post t/t score
1	Social Relationship- Verbal	4	3 later 2 (post t/t followup)
2	Impairment in communication	4	3
3	Consistent attachment to some non-living objects & its use	4	3
4	Unstable & Indecisive mind:	4	2
	Visual response		
5	Fear and lack of courage or Nervousness	4	2
6	Total score	20	12

Samprapti Ghataka

In present case *Dosha involved were Kapha* +++, *Vata*++, *Pitta*+ with all *Dushya except Sweda*, urine, *Shukra* and *strotas Rasavaha* to *majjavah* were affected. It is with bad prognosis, *Asadhya*-lifelong condition with main *Dushti* of *Agni*, *ama* and *Vata-Kapha* affecting whole body and mind.

Probable Samprapti of Rett Syndrome



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Investigations

Her LFT, KFT, CRP, CBC was normal except mild leukocytosis. In Karyotyping 46XX, presence of MECP2 gene defect. DNA and EEG were normal, MRI was showing Fronto-temoporal atrophy, Cytomegalo- Ig G was positive (133 mg).

Diagnosis

The diagnosis of Rett Syndrome was made as per diagnostic criteria of presence of MECP2 (Methyl- CpG-binding protein 2) gene defect by molecular genetic testing which in turn leads to abnormal function of other genes too

Interventions

As Rett syndrome is a complex syndrome with *Yapya* or *Asadhya*/not curable category hence it requires multiple interventions to improve quality of life. Ideally, all types of management modalities of Ayurveda-*Yuktivyapashray*, *Daiv vyapashray* & *Satvavajay chikitsa* has been added in this case to improve the multiple symptoms in a short period of time due to practical difficulty of parents to stay in India and take many leaves. They also started with rehabilitative and behavioral therapy for short duration and discontinued as finding no outcome. Table no. 6 is showing the medicine details of 1st to 3rd sitting.

Table no. 6: showing medicines administered in 1st to 3rd sitting: -

	Table no. o. showing	incurcines auminister	cami to 5 sitting.		
Formulation			Dose with duration	Anupana	
1st	2nd	3rd		•	
Agnitundivati	Ksheerbala oil-101 Avartan		1/4th of 250mg twice a	Warm water	
	10 drops tid with milk	or water	day(After food) x10 days		
2 Medhya (memory boosting) polyherbal syrup		2.5ml BD (After food),	Water		
			twice x 1 month		
		Samvardhana	2.5 ml once a day	With Milk	
		Ghrita +	(Empty stomach) for 2		
		Suvarnaprashan	months		
		800 mg twice a day x 10	With honey		
Praval Panchamrit -125 mg +			days in a month		
Guduchi satva-100 mg					
Triphla Churn	a-250 mg				
Trikatu churna	<i>u</i> -250 mg				
Application of	Vacha + Yashtimadhu	+ Trikatu +Kantakari	1 Pinch application of	Honey	
Churna on ton	gue for speech stimulation	on	each on tongue		
	Agnitundivati Medhya (mem Ashwagandha (KKRyog/com Praval Pancha Guduchi satva Triphla Churn Trikatu churna Application of	Formulation 1st	Formulation 1st 2nd 3rd Agnitundivati Ksheerbala oil-101 Avartan 10 drops tid with milk or water Medhya (memory boosting) polyherbal syrup Ashwagandha Ghrit Samvardhan a Ghrit a + Suvarnaprashan (KKRyog/combination) Kumarkalyanakrasa-60 mg Praval Panchamrit -125 mg + Guduchi satva-100 mg Triphla Churna-250 mg Trikatu churna-250 mg	Agnitundivati Ksheerbala oil-101 Avartan 10 drops tid with milk or water Medhya (memory boosting) polyherbal syrup Ashwagandha Ghrit Samvardhan + Suvarnaprashan (KKRyog/combination) Kumarkalyanakrasa-60 mg Praval Panchamrit -125 mg + Guduchi satva-100 mg Triphla Churna-250 mg Application of Vacha + Yashtimadhu + Trikatu + Kantakari I /4th of 250mg twice a day(After food) x10 days 2.5ml BD (After food), twice x 1 month 2.5 ml once a day (Empty stomach) for 2 months 800 mg twice a day x 10 days in a month	

Agnitundi vati started initially as there was Agnimandya and Ama, Strotorodha present and later the condition was improved and hence withdrawn in next sittings. Kumar kalyan rasa/ KKR was continued for around 2 months out of six months treatment course intermittently. In 1st sitting, procedures were advised as in Table no. 7

Table no.7: showing procedures performed in all the 1st to 3rd sitting

Sr. No.	Panch	Medicine used	
	1st sitting of 10 days, 20 days gap for 2nd sitting	2 nd & 3 rd same sitting of 21 days with gap of one month	
1	Nasya with Bramhi tail	Bruhan-Rukshan; start & ends with Bruhan Brahmi Tail, 1 drop pratimarsh x3 days, Rasna churna- Pradhman nasya x 3 days alternate cycles till 21 days	Brahmi Tail & Rasna churna
2	Shirodhara	Shirodhara with Brahmi tail; Vatahar kwath dhara x3-3 days, alternate cycles till 21 days	Brahmi Tail
3	Utsadan-Udvartan (whole body) Dashmool Tail + Triphala Churna & Triphala churna alone for Udvartana on whole body	Udsadan followed by Taildhara & Udavartan followed by Kwath/kanji dhara x 3-3 days alternate Brihan & rukshan action alternate cycles till 21 days	Dhanwantar tail +Ksheerbala tail for Tailadhara Vatahar kwath/Kanji



- SARIT	International Journal of Ayurvedic Medicine, Vol 11 (2), 351-357					
4	Swedana	Shashtik shali pindasweda & patrapottali sweda, alternate 3 days each, 5 cycles till 21 days	With Vata shamak Kashaya (Nirgundi, Bilva, Bala, Eranda)			
5	Basti- started after 3 days of admission	Matrabasti -Dhanwantram Tail, 15 ml for 3 days alternate Sasneha niruh basti with Vatahar kwath70 ml with saindhav, honey 5 gm each & Dhanwantaram oil 10 ml x 3 days alternate cycles till 21 days	Dhanwantaram Tail & vatahar kwatha			

As Vata and Kapha Doshas were dominant hence Vata-kaphahar procedures were started. Application on tongue was started to stimulate speech. After 1st sitting, her screaming, fear and undue phobia were reduced with relief in constipation. She was able to sit few more minutes without much forward bending. In 2nd and 3rd sitting, alternate 3 days *Bruhan & 3* days Rukshan were given to pacify both Kapha and Vata for 21 days, in which 4 cycles of Brihan procedures (Nasya by oil, Udsadan, Tailadhara, Pindswed, Matra Basti) while 3 cycles of Rukshan procedures (Pradhaman Nasya, Udvartan, Kwath Dhara, Patrapottali and Sasneha Niruh) were included. (9) In between 2 sittings there was gap of 20-30 days. Figure No-03 showing sitting in position with no drooling of saliva and Figure no. 4 depicting standing posture, post Treatment





Figure No. 3: Showing sitting without support

Figure No. 4: Standing posture post treatment

Multimodal treatment for the whole system correction approach such as Play therapy, counselling of parents, Physiotherapy, Occupational therapy, music therapy, hydrotherapy, touch therapy, mantra chanting and Yoga with the help of therapist and parents were beneficial to improve quality of life. CBT- cognitive behavioral therapy, Speech therapy are not possible till eye contact get improves hence not started.

Discussion

Rett syndrome is a neurodevelopmental with genetic origin but child's parents did not reveal the positive family history in both the families except work stress and untreated positive cytomegalo and Rubella infection which was probably pregestational. Rett syndrome is degenerative and has tendency to deteriorate the motor function, called as Apraxia. It occurs with idiopathic cause due to mutation in gene

MECP2, exclusively occur in females, interfering with every body movement, eye gaze and speech too. Thus, require multiple interventions to correct many complex symptoms at a time. In this case of classic RS, many autistic features were present. There was moderate positive change in pre-post treatment as per DSM V and CARS criteria (10).

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Here, *Kapha-vatahar* medicines and procedures with other therapies were administered to check the efficacy of these Ayurveda interventions in Rett syndrome.

Treatment Guideline

As both Sharirik as well as Mansik dosha are involved in Samprapti hence multi-modal treatment approach is necessary for the whole system correction as individualized therapeutic approach (11). Starting with Yuktivyapashray Chikitsa-Agnideepan, Pachan and Anuloman was essential initial step in Rett syndrome achieved by Agnitundi ras.(12)] Sanvardhan or Bramhi Ghrit kalp- As the main centre of Vyadhi Adhishtan is brain hence any Ghrit kalp fortified with Medhya Dravyas or intake of PuranGhrit (old ghee) is very much important in treatment point of view. Kalyanak or Mahakalyanak ghrit, Panchgavya ghrit etc. also can be given. (13-14) Medhya Shamanaushadhi polyherbal syrup, Suvarnaprashan medicines to boost the action of neurotransmitters, coordination of synapse thus, augment brain function and acts as Medhya, Rasayna, Buddhivardhak (nootropic). (15,16) Regarding doses, it is already described by Acharya Charak and Vagbhat that as per Agni, Koshta, Vaya, Bala, Kala etc, Vaidya has to decide the dose which may vary time to time, so as per these variations dose had changed as per need.

Kumar Kalyan ras

It provides *Balya- Brihan & Rasayan* effect to enhance the function of all *Dhatu*, general immunity, boost growth and development of whole body and to reduce debility. It is *Sukshma, Stroto gami, Pachak, Anulomak, Vatahar* and *Rasayan*. (17)

Application on tongue

For the symptomatic treatment of saliva drooling or speech delay, Yastimadhu, Vacha, Kantakari and Trikatu mixed powder is beneficial as Vak shudddhikar yog with honey. (18) Panchkarma-Poorva karma- Rukshan, Deepan-Pachan, Snehan-swedan, & Pre-procedures like Talapodichhil/Shirolepa, Shirodhara, Udvartan, Udsadan, Annalepan, Patrapottali, Pindasweda, Kwathdhara, Takradhara, Kanjidhara or Tailadhara etc these found to be



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effective, improve micro-circulation, eliminate toxin from skin and provides nutrients too (19). These are hot so pacify *Kapha-vata* both, required to tackle global developmental delay in this case. Alternate *Rukshan-brihan* procedures act as controlled action of *Kapha-vatahar*. *Rukshan* leads to *Kaphhar*, *Vata* stimulant to activate motor function as there was quadriplegia while *Bruhana*- for the action of *Balya-Rasayna*, *Medhya* effect.(20,21) These help in mobilizing the deeply seated morbid *Doshas* from remote areas of the body to the nearest site to expel by pre-procedures and main *Panchkarma* procedures.

Pradhan karma

Nasya, Basti-Niruh, Matra Basti play an important role in RS. These Panchkarma specially Basti focuses on correction of metabolic disturbances, provides nutrition as well as excretes the waste products by its hot, unctuous, Vatahar properties, stimulates enteric plexus in turn central nervous system and regulates the normal functioning of not only Apan vayu but also other all types.(22) Venous drainage of rectum facilitates absorption of Basti dravya by portal

circulation and then systemic circulation, thus useful in all *Vatik* conditions hence called as '*Ardha chikitsa*'. Through *Nasya*, medicine directly goes to brain and stimulates its cognitive as well as sensory-motor function with correction of maladaptive behavior. (23)

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Paschat karma

Sansarjan krama stimulates appetite, thereafter Balya-brihan & Rasayan medicines effectively could act. The remaining latent Doshas were pacified by following alternative treatment modalities mentioned in table no.8. These modalities help to calm the triggering factors provoking regression of motor function and poor communication profoundly present in Rett syndrome. As in Rett Syndrome, there is reversal of motor function which patient achieves in toddler age group but after treatment we noticed a steady gain in different new milestones of gross motor, fine motor, personal-social as well as language and understanding, that is the most positive achievement of this case study. Also, gradation of DSM V features was shifted to less scoring pattern than more.

Table no.8: showing other Complementary Alternative Medicine interventions with mode of action and correlation with Avurveda *Chikitsa traya*

No	Name of therapy	Characteristic	Outcome	Ayurveda counterpart
1	Counselling (24)	Problem solving by communication with parents about tackling & training of care	Addresses the severity of grievances, helps to plan management with assurance & positivity	Satvavajay chikitsa (25)
2	<i>Yogasanas</i> -passive (26)	Typical body postures with performer's pressure	Improve blood circulation, flexibility & motor function	Satvavajay, swasthavritta
3	Omkar chanting with gentle touch marma therapy (27)	Gentle touch starts from skull to last part of spine,	Offers secure and love- caring feeling, facilitate parent-child bonding, relieve anxiety, fear	Daivavyapashray Chikitsa (28)
4	Music therapy (29)	Instrumental light music in slow volume	Provides relaxation, stops screaming, relaxes body & mind	Daivavyapashray Chikitsa
5	Occupational & Physio therapy (30-31)	Provide support by braces, shoes & helps muscle to move, strengthen	Improve range of motion	Yuktivyapashray Chikitsa-YC
6	Hydrotherapy (32)	Passive water exercises	To boost muscle strength	YC

These measures are very much beneficial in combating with complex syndromes like Rett. With multimodal and wholistic approach only there is a hope to achieve some positive results as RS is a life-long condition due to genetic origin.

Conclusion

The Autism Spectrum Disorders require the long-term interventions so as Rett syndrome also. The improvements seen after each course of management may amplify steadily. Ayurveda opens a door in the management of Autism and similar conditions and shows the spark of expectations and optimism to affected children with the help of all *Chikitsa traya (Satvavajay, Yuktivyapashraya* and *Daiv vyapashraya chikitsa)* modalities as per need.

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