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An Overview of Rett Syndrome with its Homoeopathic Management

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ABSTRACT

Rett syndrome (RS) is characterised by a severe neurodevelopmental disorder that contributes significantly to severe intellectual disability in females worldwide. The Majority of the cases were caused by mutations in the X-linked gene methyl-CpG-binding protein 2 (MECP2). The Mutation of MECP2 was seen in 70% -80% cases and remaining 20%-30% of the cases were attributes to unknown mutation. Neurodevelopment has been referred to as neurological regression that severely affects cognitive, motor, communication skills and by autonomic dysfunction and often seizure disorder, a disturbed breathing pattern with hyperventilation, periodic apnoea, scoliosis, growth retardation and gait apraxia. The most common oral findings were bruxism. No etiological treatment is available in modern medicine. We discuss the clinical aspects and review in clinical and homoeopathic management.

Key words: Rett Syndrome, Neuro-developmental disorder, Neurological regression, Individualisation, Homoeopathy

INTRODUCTION

Rett syndrome (RS) is characterised by severe neurodevelopmental disorder that causes severe intellectual disability in females worldwide[1]. It is considered to be the second most common cause, after Down's syndrome, of mental retardation in females[1].It was first identified by an Austrian paediatric neurologist called Dr. Andres Rett in 1966. Dr. Rett noticed in his clinic normal early development, followed by a period of regression and loss of purposeful hand movements. He also called it "cerebroatrophic hyperammonaemia" in a 1996 German publication.[2] However, the first published report was noted in 1983 by Dr. BengtHagberg et al., who reported the condition of 35 patients with strikingly similar clinical features of progressive autism, loss of purposeful hand movements, ataxia and acquired microcephaly which led to a significant increment in interest and awareness about this rare condition[3].

Rett syndrome has been consider that the periods of apnea may contribute to brain damage because oxygen saturation levels as low as 50% have been noted. Infants with RTT show normal growth and development until the age of 6–18 months, followed by regression of skills and development; they often exhibit autistic behaviours in the early stages[4]. Neurodevelopment has been referred to as neurological regression that severely affects cognitive, motor, communication skills and by autonomic dysfunction and often seizure disorder, a disturbed breathing pattern with

hyperventilation, periodic apnoea, scoliosis, growth retardation and gait apraxia[5].

EPIDEMIOLOGY

Rett syndrome (RTT) is a childhood neurodevelopmental disorder, with an approximate prevalence of 1 in 10,000–15,000 female live births[2].

STAGES OF RETT SYNDROME [2,11]

The development of RS is usually described in four stages

Stage I: Early Onset Stage - Between 3 months and 3 years

- Loss of acquired skill like hand function, vocalization.
- Developmental stagnation (e.g. sitting, crawling, vocalization)
- Slowing head growth
- Postural delays
- Hypotonia
- Babbling and new word appear but poor.
- Insomnia may also develop.
- Child may not make eye contact with family members.
- Child may also develop a lack of awareness for her environment especially plays activities.

Stage II: Rapid Destructive (regression) Stage- Between 1 to 4 years

- Rapid regression of language skills.
- Loss of acquired motor skills like clapping, washing.
- Extreme screaming and crying episode by 18- 24 months.
- Bruxism.
- Exploratory character of her play and grasping for object is lost.
- Hand sterotypic movements
- Autistic like behaviour and panic like attacks.
- Breathing irregularities like episodic apnea, hyperpnea.
- Microcephaly worsens
- Convulsion may occur.

Stage III: Plateau Stage/ pseudo-stationary stage- Between 2 to 10 years old

- Severe mental retardation.
- Increased alertness of her surroundings.
- Prominent hand apraxia.
- Spasticity.
- Seizures are common

Stage IV: Late Motor Deterioration Stage- usually occurs around age ten on and lasts for years.

- Growth retardation
- Wasting, dystonia and bradykinesia
- Scoliosis, muscular atrophy, muscle rigidity.
- Wheelchair dependency in some cases
- Eye contact improved.
- Seizure activity typically decreased

Each person affected with Rett syndrome age of onset, duration, severity of symptoms may vary.

Atypical Rett syndrome variants: [12]

Early onset RS/ Congenital RS

This is more severe variant. Child showing congenital hypotonia and infantile spasm. Early normal development is completely absent making its diagnosis very difficult.

Preserved speech variant

In this form of RS, partial language is preserved; they retain some speech and the ability to walk. Most of these individuals also show a milder expression of other signs of RS.

Forme Fruste (FF)

This is a known variant of RS with milder clinical features. In this form of RS, the clinical signs appear at around age 2–3 years after the child has already expressive her language. Motor skills are well preserved.

Male with RS

Males with a 46XY karyotype, can also be affected by mutations in MECP2.

MANAGEMENT[4]

There are recently no known specific treatments for RTT in modern medicine. Clinical Care and homoeopathic treatment and genetic counselling (with DNA tests to rule out familial transmission), support and advice for the families, and physiotherapeutic measures reduce scoliosis development to the extent possible. During regression, certain features of RTT are similar to autism; misdiagnosis of RTT as autism is, therefore, likely.

Standard treatment Guidelines according to Hahnemann[6]

§153- the more striking singular, uncommon and peculiar (characteristic) signs and symptoms of the case of disease….. in order to constitute it the most suitable for effecting the cure.

§164- The small number of homoeopathic symptoms present in the best selected medicine is no obstacles to the cure in cases where these few medicinal symptoms are chiefly of an uncommon kind .....under such circumstances without any particular disturbances.

§284 foot note of 6th edition – The power of medicines acting upon the infant through the milk of the mother or wet nurse is wonderfully helpful……..But the case of mothers in their (first) pregnancy by means of a mild antipsoric treatment , especially with sulphur dynamizations prepared according to the directions in this edition, is indispensable in order to destroy the psora that producer of most chronic diseases which is given them hereditarily; destroy it both within themselves and in the foetus, thereby protecting posterity in advance. This is true of pregnant women thus treated; they have given birth to children usually more healthy and stronger, to the astonishment of everyday.

Homoeopathic concept of causation and its relationship with miasms[10]

Dr. Hahnemann used the term miasm with two perspectives: one is the 'pollution' that 'causes the disease' and another is the 'noxious influence or atmosphere' that influences the disease process. Dr. Hahnemann grouped miasm into psora, sycosis and syphilis. Tubercular is a diathesis, a combination of Psora with sycosis or syphilis also called pseudo-psora. Loading from heredity along with the predisposition determines the miasmatic evolution in disease. The chronic miasms in the mother were responsible for hereditary disorders in the child when he/ she was in the intra-uterine state, and these could be treated by the homoeopathic remedy.

Opinion of different stalwarts on cause

Stuart closes view:[13] Dr. Stuart close's writes in his The genius of homoeopathy, Lecturers and Essays Homoeopathic Philosophy " The real cause is the whole of the antecedents , and we have no right , philosophically speaking, to give the name of the cause to one of them , exclusively of the others". The most pernicious medical error is to assume that a disease had a single cause , and to direct all efforts and agencies aganist that. This error is responsible for failure in treatment.

Roberts view:[14] Dr. H. A Robert's writes in his book " The principle and art of cure by Homoeopathy" under chapter disease classification that it was Hahnemann's teaching that the removal of the cause was the first step in the proper method to cure. So in Homoeopathic therapeutics, prescription on the basis of causative factor is a unique feature. It meets with the management of the psychic, psycho-neurotic, psycho-somatic illness and other maladies in which hereditary predisposition plays an important role.

REPERTORIAL APPROACH

Phatak Repertory[8]

- Children – Talk late- Agar, Bar-c, Calc-p, Nat.mur, sanic etc
- Children – Walk late- Agar, Calc, Calc.p, caus, sil, sulp.
- Speech-Development, retarded- Phos

- Slow comprehension, thinking etc- Bar.Carb, Bry, Hell,Phos, Plb, Puls, sulp. etc
- Growth affected, disorder of- Bar-C, Calc, Calc-p, phos, phos-ac, sil., Thyr.

Homoeopathic medical repertory – Robin Murphy[9]

Children

- Learning, disabilities, - AGAR. BAR-C., CALC., CARC. LYC.
  - Understanding difficult- BAR-C., CARC.
- Retarded mentally- BAR-C., CARC.
- Walk general late learning to- BAR-C., CALC., CALC-F., CALC-P., CAUST. NAT-M.
  - Tardy, development of bones- CALC-P.
  - Weak, legs, with- CALC.
- Development delayed or arrested- BAR-C., CAL., CAL-P., CARC.
  - Bones of- calc., calc-p.
  - Muscles of – calc., nat-m.

Kent Repertory[7]

- Mind- Dullness, sluggishness, difficulty of thinking and comprehending: Arg-n., Bapt., Bar-c., Bar-m., Bell., Bry.,Calc., Cal-p., Cal-s., Carb-v., Graph., Guaj., Hell., Kali-br., Kali-c., Lach., Laur., Lyc., Nat-a., Nat-c., Nat-m., Nux-m., Op., Ph-ac., Phos., Pic-ac., Plb., Puls., Seneg., Sep., Sil., Staph., Sulph., Tub., Zinc.
- Mind- Speech, incoherent: Bry.,Cann-i., Hyos., Lach., Phos., Rhus-t., Stram.
- Head- Convulsions of the right side of head: Mygal
- Teeth- Grinding: Apis., Bell., Hyos.
- Respiration- Difficult: Anac., Ant-t., Apis, Ars., Bry., Cact., Carb-v., Caust., Chel., Chin.,Chlor., Cina., Crot-t., Cupr., Cupr-ar., ferr., Hep., Ip., Kali-ar., Kali-c., Kali-i., Lach., Lob., Meph., Merc-c., Naja, Nat-s., Nux-m., Op, Phos., Puls., Sel., Sil., Spong., Squil., Stann., Stry., Sulph, Tarent., Verat.
- Extremities- Motion, difficult: *camph., con., cup., pic-ac.*
- Extremities- Motion, loss of power of: *Apis.,stram., tarent.*
- Extremities- Walking, difficult: Aur., chin., olnd., ter. Cite this article: Kyle SM, Vashi N, Justice

CONCLUSION

From this above discussion, I conclude that for any success in the clinical practice knowledge are most essential factors. If anyone tries to take maximum care while taking case and analyses according to Hahnemann, the prescription would be given us symptomatic relief.

Declaration by Authors

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REFERENCES

1. Weaving LS, C Jellaway, J Gecz, Rett syndrome : Clinical review and genetic update . 2004, oct15 : 1-7 doi: 10.1136/027730
2. Stephanie kyle, Nett V. Rett syndrome: a neurological disorder with metabolic components.2018, Feb 20.http://dx.doi.org/10.1098/rsob.170216
3. Mahdi, S.S.; Jafri, H.A.Allana, R.; Amenta, F.; Khawaja, M.;Qasim, S.S.B. Oral Manifestations of Rett Syndrome - A Systematic Review. Int. J. Environ. Res. Public Health 2021, 18, 1162. https://doi.org/10.3390/ijerph18031162
4. Jalal Gharesouran a, Azizeh Farshbaf Khalili b, Noushin Sorkhkoh Azari c, Leila Vahedi d. First case report of Rett syndrome in the Azeri Turkish population and brief review of the literature 2015, feb23 http://dx.doi.org/10.1016/j.ebcr.2014.11.001 2213-3232/© 2014 Published by Elsevier Inc.
5. E.E.J. Smeets K. Pelc B. Dan. Rett syndrome.2012, april 16 DOI: 10.1159/000337637
6. Hahnemann S. Dudgeon RE, Organon Of Medicine. 28th impression. New delhi : B jain publishers Pvt. Ltd, 2012

7. kent JT, Repertory of Homoeopathic materia medica, Second rearranged ed.37th Impression. New Delhi : B. Jain Publishers Pvt. Ltd;2009. P 624-636
8. Phatak SR. A concise repertory of homoeopathic medicines, 4th ed. New Delhi: B. Jain publishers Pvt. Ltd; 2005.
9. Murphy R. Homeopathic Medical Repertory. 3rd ed. New Delhi; B jain publishers Pvt. Ltd; 2005
10. Nikumbh SB. Studying the evolution of miasm in autism spectrum disorder: A case series. J intgr stand Homoeopathy 2020;3(2):43-50
11. http//www.orpha.net/data/patho/GB/uk-Rett.pdf
12. Meir Lotan and Bruria Ben. Rett syndrome. A Review with Emphasis on clinical characteristics and intervention. The scientific WORLD JOURNAL(2006)6,1517-1541, ISSN 1537-744X; DOI 10.1100/tsw.2006.249
13. Close, S: *The Genius of Homoeopathy , Lecturer and Essays Homoeopathic Philosophy* ; Reprint Edition-2001 ; Published by jain publishers (P) Ltd; New Delhi.
14. Roberts , H.A: *The Principles and Art of Cure by Homoeopathy , Modern Text Book* ; Reprint Edition -2001 ; B. Jain Publishers (P) Ltd., New Delhi.

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