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Research Paper

A case study on Cri-Du-Chat syndrome: early intervention and rehabilitation

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ABSTRACT

Cri-du-chat syndrome also known as crying cat syndrome is a rare genetic condition that is caused by the deletion of genetic material on the small arm of chromosome 5. It is a rare syndrome with reported incidence of 1 in 50000 live births.2 This is a case of 2 and half year old girl with be hypotonic, microcephaly, cat like crying, distinct facial dysmorphism, speech & cognition was affected, flat footed resulted in unstable gait and frequent falls. Child showed violent behavior with poor attention and concentration. An early diagnosis resulted in early intervention in motor, speech, psychological and pre academics skills at child development centre. This case is successfully managed with conservative rehabilitation plan preparing him for normal school.

Keywords: *Cat like cry, Microcephaly, Hypotonic, Rehabilitation.*

The Cri du Chat syndrome (CdCS) is a genetic disease resulting from a deletion of short arm of chromosome 5 (5p-).[1] Its clinical and cytogenetic aspects were first described by Lejeune *et al*. in 1963 .It is also known as 5p Syndrome [2].The incidence ranges from 1:15,000 to1:50,000 live-born infants.[3] The most important clinical features are a high-pitched cat-like cry (hence the name of the syndrome), distinct facial dysmorphism, microcephaly, development delay, severe psychomotor and mental retardation.[3]

Distinctive facial features include small head size –microcephaly, widely-spaced eyes, round/plump-moon face, a broad nasal bridge, crossed eyes abnormally small jaw and improper alignment of upper and lower teeth. The most common heart defect is patent ductus arteriosus, less common is inguinal hernia, gastro esophageal reflux and syndactyly.[4]

Most of the children have difficulty with language, but can express with few basic words, gestures, sign language[5]. Other characteristics include feeding difficulties, delay in walking, hyperactivity, scoliosis and retardation. These children are usually friendly, happy and enjoy social interaction. [5]Affected children exhibit delays in the acquisition of skills requiring

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coordination of musculature and mental activities (psychomotor disability) and moderate to severe intellectual disability[6].It is diagnosed by FISH genetic analysis (fluorescence in situ hybridization.

Case Presentation

This is a case of 2 and half years old child who was admitted to child development centre in July 2017, with chief complaints of child is unable to stand and walk independently, cannot speak age appropriately, cries extensively and throws objects on others. Child was evaluated on following heading- Birth history: child is 2nd issue to non consanguineous parents. Full Term baby with avg birth weight (2.4 kg) with history of neonatal jaundice and microcephaly was reported. Personal history: the child is irritable and hyperactive.

On physical examination child was found to be hypotonic ,hyper mobile joints, mid brain reflexes not yet integrated, flat footed resulted in unstable gait and frequent falls. The child had abnormal motor development pattern (a w sitting and frog jumping), bilateral flat foot and poor hand manipulation and fine motor skills were reported.

Speech examination child had high arched palate and widened nasal bridge which resulted in high pitched nasal sounds. She communicated for needs with gestures and crying. Child had limited vocabulary of 5-7 words. She understands lexical items and responds to family members.

Behavior assessment observed that child was continuously licking her nose with tongue, restless, does not follow commands, hits other children and throws objects .child was totally dependent for activities of daily living like dressing, bathing and toilet skills. Child has shape, color concept.

Diagnostic Assessments: The child was assessed on following scale:

1. Portage guide to early intervention checklist: socialization skills 2 years, language skills 12 months, self help skills 12 months, motor 12 months, fine motor skills 6 months, and cognition skills 12 months.
2. Gross motor fine motor scale (GMFM-88) consist of 5 subsets that evaluate lying (100%), sitting (100%), crawling and kneeling (78%),standing (7.6%) and walking (16.6%) of the child total score was 60.44%.
3. Receptive Expressive emergent language scale –Extended (REELS-EXTENDED) is used to identify receptive and expressive language problems and calculate receptive and expressive age of the child .the child's RLA was: 12-14 months and ELA was 8-9 mths. Bilateral hearing sensitivity was within normal limits.
4. On Denver Developmental Screening Test (DDST): The Developmental Age of the child was 12 months and Developmental Quotient was 57 indicating mild delay in developmental functioning.
5. On Vineland Social Maturity Scale (VSMS): The Child's Social Age was 15 months and Social Quotient was 67 indicating mild deficits in socio adaptive functioning.
6. On BASIC MR: The score of the child bear significant in domain of Violent and Distractive Behaviour-2, Self Injurious Behavior-3, Repetition Behavior -4.

Interventions

After a detailed evaluation of the child a short and long term goal were targeted and was given regular physiotherapy, speech therapy, cognitive training, behavior modification and pre academics training.

Physiotherapy: It focused on gross motor development: correction of abnormal pattern of motor development for w sitting and frog jumping using verbal feedback. Use of medial arch supports as insoles in shoes while standing and walking was introduced. Child was trained for standing balance exercises with changing base of support .Parallel bars was used for gait training of the child. Swiss ball exercises helped in improving protective/ equilibrium reflexes and thus here fear of fall also reduced. For coordination: equilibrium exercises and frenkel exercises were used. Fine motor development: constraint induced movement therapy was used to control his fine motor movements. Emphasis was given on proprioceptive neuromuscular facilitation techniques for training bimanual tasks.

Cognitive training: It focused on cognitive functioning: She was given problem solving training through tower of color rings, shape puzzle task and reasoning exercises.

Attention enhancement training: She was started with beading activities, and sorting images activity.

Memory training: With the help of brain toys like (wooden tree shape puzzle, colorful abacus beads) to improve her cognitive abilities with the size, color and number organization concept.

Behavior modification: She started screaming whenever she used to get excited. During sessions, she was instructed to make proper eye contact with verbal cues and good and bad behavior was explained to the child. Orientation of basic emotions and way to expressing the emotions was also taught. She was told about few bad habits like licking nose, teeth grinding are not good which she does normally.

Psychosocial training: She was made to socialize with other play mates cooperatively. She was given instructions about to introduce herself first, when in group and show gratitude by saying "thank you' after the game with her play mates.

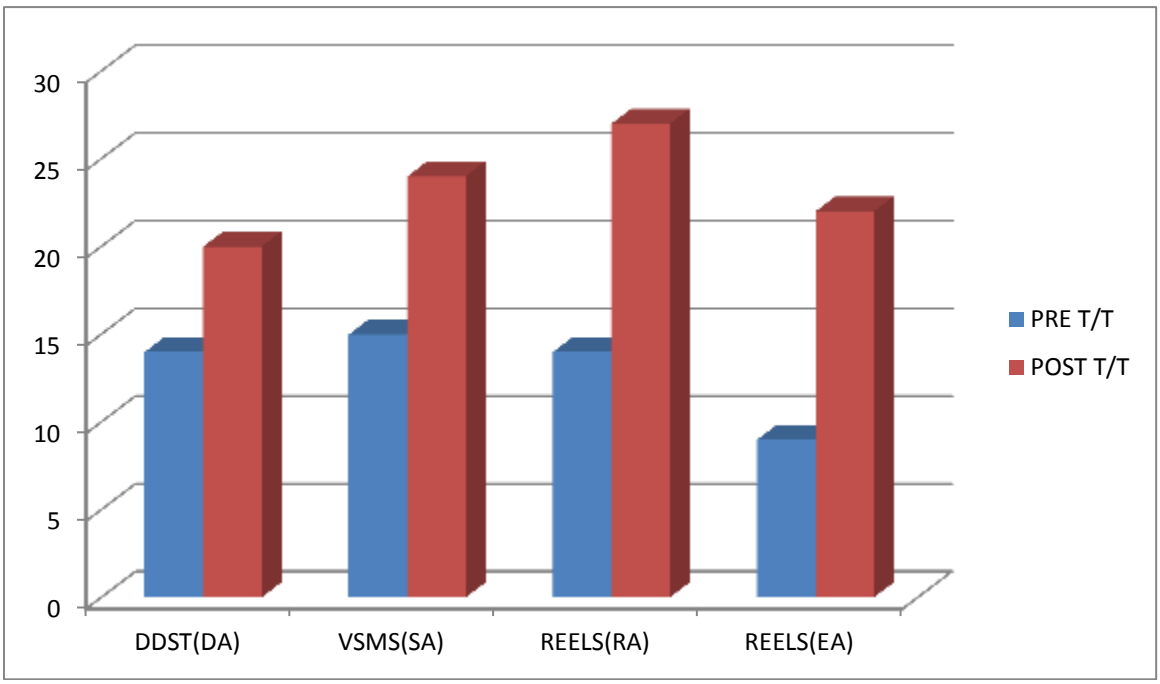
Speech therapy: Imitation technique, repetition technique, recitation technique was used. Respiratory exercises were also taught to the child. Blowing activities, exercise of tongue, lips and oral cavity and other techniques like counting aloud and biofeedback was also used.
Pre academics sessions: It included pre writing skills, fine motor development skills, reading skills and recitation of rhymes.

DISCUSSION

Every quarter the child performance was evaluated and new treatment goals were set. After 15 months the therapy gain of the child is as follows:

Physiotherapy: the child has reduced w –sitting and frog jumping and now follows reciprocal crawling; child can stand and walk independently. Child has started running a few steps too. Fine motor control has improved example: improved timing of Pecs's board activity. Bimanual activities requiring coordination example: stringing beads has also improved. On GMFM scale, child motor skills improved and she scored 80.12%.

Behavior modification
DDST - After assessments, the child **DA** was 20 months and **DQ** was 62.
VSMS - The child **SA** was 24 months and **SQ** was 72.
Basic MR Total score 4 suggesting no significant behavior problems



Speech: The child was reassessed on **REELS** scale. His **RLA** has improved to 24-27 month and ELA **to** 20-22 months. For clarity of speech, repetition of words ra, la, ta, na and simple sentence recitation is done.

On Portage scale, socializations skills improved to 3 years, language skills to 2 years, self help skills to 3 years, motor skills to 2 years, fine motor skills to 2 years and cognitive skills to 3 years.

Pre academics: child had poor pencil grip and no reading /writing skills. Now the child can scribble standing line, slanting line and sleeping line. Child can write letter A & B. Child recognizes 10 colors. Child can read 3 letter words, read picture book of fruits/animals/birds /opposites/vehicles/part of body.

CONCLUSION

It is challenge for the rehabilitation team to treat such a child but a team approach is a must for such a case. Mostly such cases remain undiagnosed so emphasis on proper diagnosis is a must so that a treatment could be started as early as possible.

An early diagnosis, proper counseling to the parents and early rehabilitation can help child to overcome his disability. This case has been successfully managed with the conservative approach, which itself is a rare occurrence.

REFERENCES

1. Nandhagopal, R., & Udayakumar, A. M. (2014). Cri-du-chat syndrome. *Indian Journal of Medical Research, 140*, 570–571.
2. Rodriguez-Caballero, A., Torres-Lagares, D., Rodriguez-Perez, A., et al. (2010). Cri du chat syndrome: A critical review. *Medicina Oral, Patología Oral y Cirugía Bucal, 15*(3), e473–e478
3. Higurashi, M., Oda, M., Iijima, K., et al. (1990). Livebirths prevalence and follow-up of malformation syndromes in 27,472 newborns. *Brain & Development, 12*, 770–773.

4. Chen H. (2016) Cri-Du-Chat Syndrome. In: Atlas of Genetic Diagnosis and Counseling. Springer, New York, NY
5. Cornish, K. M., & Munir, F. (1998). Receptive and expressive language skills in children with Cri-Du-Chat Syndrome. *Journal of Communication Disorders, 31*, 73–80, quiz 80–81.
6. Cornish, K. M., & Pigram, J. (1996). Developmental and behavioural characteristics of cri du chat syndrome. *Archives of Disease in Childhood, 75*, 448–450.
7. Dykens, E. M., & Clark, D. J. (1997). Correlates of maladaptive behavior in individuals with 5p- (cri du chat) syndrome. *Developmental Medicine and Child Neurology, 39*, 752–756.

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Conflict of Interest
The author declared no conflict of interests.

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