Global Scientific JOURNALS

GSJ: Volume 8, Issue 2, February 2020, Online: ISSN 2320-9186
www.globalscientificjournal.com

SPEECH AND LANGUAGE CHARECTERSTICS IN DRAVET SYNDROME : A CASE STUDY

[1]MaithilyRai, [2]EaswariGopakumar , [1,2]Lecturer

Dr. M.V. Shetty College of Speech
Hearing, Mangalore-15

E-mail: manavalike.rai@gmail.com

Ph: +918762526124

Abstract

A 2 year 4 months old child with no family history of speech and language delay was referred to speech language pathologist with the complaint of limited speech and language output. He was diagnosed with Dravet syndrome, which is one of the most malignant form of epilepsy.(Oguni et al., 2001;Dravet et al., 2005), . The most common gene mutation linked to Dravet syndrome is in a gene called SCN1A (Rosander, 2015) but not all SCN1A gene lead to Dravet syndrome.

On evaluation it was observed that the child has severe delay in speech and language skills. Results revealed that Phonology, semantics, Play and Pragmatics of the child was within the age range of 9 to 12 months, Syntactic development of the child was nil. He started babbling by 1.5 years of age, bysyllabic utterances are not achieved. Feeding skills of the child are affected, he has difficulty in swallowing the food with solid consistency and uses smooth consistency food.Sudden unexpected death is high in Individual with Drawet syndrome. It is very important for a professional to have an idea about the seizure triggers for an effective rehabilitation.

Background

Dravet syndrome or Severe Myoclonic Epilepsy in infancy(SME) was described by Dravet in 1978.It is a rare genetic epileptic encephalopathy (dysfunction of brain) that begins in the post natal period or initial childhood.Incidences of probably less than 1 per 40,000 live births were reported in 1990 in the U.S.A (Hurst, 1990). Almost the same figure (1/20,000 or 30,000) was reported by Yakoub et.al (1992). In these studies males are more often affected than females in a ratio of 2:1. This syndrome is characterized by febrile and afebrile,

generalized and unilateral, tonic or clonic seizures. Seizures occur generally in the first year of life of a normal infant and later associated with myoclonus, atypical absence and partial seizure. Seizures lasting more than 10 minutes, seizures occurring on one side of the body, and seizures triggered by a warm water bath in children under 12 months old are considered significant risk factors for a Dravet syndrome. The most common gene mutation linked to Dravet syndrome is in a gene called SCN1A (Rosander, 2015) but not all SCN1A gene lead to Dravet syndrome.

The EEG, imaging, and development are usually normal at first, but abnormal EEGs and developmental delays often appear in the 2nd and 3rd year of life (Wirrell 2017). In coordination (ataxia) and low muscle tone (hypotonic) are often apparent in the early years and remain a characteristic of the syndrome throughout life (Villas 2017). Speech delay is frequently seen before age of 2 years. Physical occupational and speech therapy are recommended (Wirrell 2017). Other common characteristics and health problems include behavioral issues, growth and nutrition issues, unsteady walking and disruptions of the autonomic nervous system, which regulates things such as body temperature and sweating (Lagae).

Cognitive deficits and behavioral disturbances are a common trait. Developmental delay becomes progressively evident from the second year onwards. As a rule, children start walking at a normal age but an unsteady gait develops for an unusually long period. Patients fine motor abilities do not develop well and have poor eye hand coordination. Affected children are restless, do not listen to adults, and are not interested in playing with educational toys and participating in the usual activities of their age group. Language also starts at a normal age, but progresses very slowly, and many patients do not reach the stage of constructing elementary sentences. Not all these traits are present in all patients and the traits tend to be less severe in those with a recent diagnosis (Buoni et al., 2006; Ragonaet al.2010)

Case presentation

A 2 year 4 months old child with no family history of speech and language problems was referred to speech language pathologist with the complaint of limited speech and language output. Case history reports that, Child was conceived normally. Iron and folic acid supplements were taken by the mother regularly under the supervision of the physician.

Normal vaginal delivery was reported. He was a full term baby, weighing 3.5 kg with immediate birth cry.

He had presented with convulsion at 6 months with stiffness of all 4 limbs, up rolling of eyes and he was discharged with prescription Midaz nasal spray. At 8 months of age he presented fever with seizure and was discharged with Spy.Tegrital 2mg bd, At 14 months of age another episode of fever with convulsion had occurred. Patient is on regular follow up and medications for the same.

Child had attained head control by 9.5 to 10 months, rolled over in a year; started walking with support by the age of 2 but walking without support is still not accomplished. Gross motor skills like reaching the objects was accomplished by 6 months, however finer skills like, turning the page of book is still not attained. He started babbling by 1.5 years of age, bysyllabic utterances are not achieved.

Investigations

By the time of referral the child had undergone intensive investigations, Neurological evaluation was completed and suggested that the child had a normal sleep awake EEG and MRI was found to be typical, Reports also confirmed the concern of mutated SCN1A gene. The child was diagnosed as Global Development Delay secondary to Dravet syndrome.

Sensory Profiling

Childs sensory profiling was carried out by using Caregivers questionnaire Sensory profile by Winnie 1999. This involved questions related to sensory processing, Visual processing, Vestibular processing, Touch processing, Multisensory processing, Oral sensory processing, Sensory processing related to Endurance/ Tone, Modulation related to body position and movement, Modulation of movement affecting activity level, Modulation of sensory input affecting emotional responses, Modulation of visual input affecting emotional responses and activity level, emotional and social response, Behavioral outcomes of sensory processing and also items indicating threshold for response.

Reports of the profiling reveled that Child Frequently responds to unexpected or loud sound and also responds to name call by smiling and turning head towards the sound source suggesting a normal hearingfor the child. This was confirmed by the audiological evaluation

which confirmed that the child has normal hearing sensitivity in both ears.Visual tracking is present and there are no symptoms of difficulties in visual perception.he manages to sit for around 10 minutes without support. Response to pain by cry is also reported. Child was cooperative and gave good coordinated response for multisensory processing tasks.

Feeding skills of the child are affected, he has difficulty in swallowing the food with solid consistency and therefore he uses smooth consistency food, the child's motoric development is delayed. He exhibits most of the motoric problems like muscle weakness, moves stiffly and gets tired easily especially when standing or holding a particular body position, locks joint for stability, weak gasp and poor endurance. He can't lift heavy objects and appears to be lethargic. He has difficulty in walking and takes assistance to perform the same. Emotional response are reported to be intact as he cries when scared and when needs are not satisfied, smiles when seeing mother and familiar people.He is well affectionate with the parents and also has good eye to eye contact.

COMDEAL CHECKLIST

As a part of evaluation, COMDEAL CHECKLIST given by Prathibakaranth was administered to determine the age level for gross motor skills, fine motor skills, activity of daily living, receptive language, expressive language, cognitive skills, social skills and emotional skills. From the results, a 10 months delay was observed in performance of gross motor skills, fine motor skills and receptive language. Childs ability to perform the activities of daily living and his expressive language was severely delayed by 22 months.Parent child interaction was qualitatively and quantitatively good.

SPEECH AND LANGUAGE DEVELOPMENT CHART(SLDC)

Parameters of language was assessed using a standerdised test named SLDC given by Addy Gard et.al, Results revealed that Phonology,semantics,Play and Pragmatics of the child was within the age range of 9 to 12 months and theSyntactic development of the child was observed nil.

Oro peripheral Mechanism examination

Oral structures were structurally normal with poor lip seal. Child needs assistance for feeding and is able to swallow only smooth solid consistency of food. Vegetative skills such as biting, blowing and chewing are impaired. Thechild is bottle fed and no difficulty regarding swallowing was observed.

Non verbal mode of communication is used to express his needs. No significant behavioral issues were observed. The child indulges himself in solo play as well as peer play, but without verbal communication. Pre linguistic skills such as sitting tolerance, response to name call, eye to eye contact were fair. Pragmatic skills such as greeting, requesting, turn taking, narration, topic initiation, topic maintenance are not yet achieved.

SPEECH CHARACTORESTICS

His comprehension and expression of language was limited to one word utterances. This resulted in limited fluency although vocal characteristics were within normal limits.

The overall results of assessment revealed that the client has delay in speech and language.

Recommendation and follow up

It is recommended that, the client should attend speech language therapy. The client was attentive and cooperative throughout the assessment and parents support was found to be evident. Therefore the treatment prognosis for improvement of speech and language skills is judged to be good. Treatment targets may include speech therapy and home training.

Complications

Dravet syndrome is one of the most malignant forms of epilepsy. (Oguni et al., 2001;Dravet et al., 2005).Sudden unexpected death is high in this case. It is very important for a professional to have an idea about the seizure triggers such as stress, boredom, lack of sleep, irregular sleep and meal patterns, low blood sugar for an effective and risk-free management.

.

References

Hurst DL.(1990) Epidemiology of severe myoclonic epilepsy of infancy.*Epilepsia*

Yakoub M, Dulac O, Jambaque I, Plouin P. (1992) Early diagnosis of severe myoclonic epilepsy in infancy. *Brain Dev*

Rosander, C., &Hallböök, T. (2015).Dravet syndrome in Sweden: a population-based study. *Developmental Medicine & Child Neurology*,.

Wirrell, E. C., Laux, L., Donner, E., Jette, N., Knupp, K., Meskis, M. A., ...& Berg, A. T. (2017). Optimizing the diagnosis and management of Dravet syndrome: recommendations from a North American consensus panel.*Pediatric neurology*,

Villas, N., Meskis, M. A., &Goodliffe, S. (2017). Dravet syndrome: characteristics, comorbidities, and caregiver concerns. *Epilepsy &Behavior*,.

Ceulemans, B., Boel, M., Leyssens, K., Van Rossem, C., Neels, P., Jorens, P. G., &Lagae, L. (2012).Successful use of fenfluramine as an add-on treatment for Dravet syndrome.*Epilepsia*

Buoni S, Orrico A, Galli L, Zannolli R, Burroni L, Hayek J, Fois A, Sorrentino V. (2006) *SCN1A* (2528delG) novel truncating mutation with benign outcome of severe myoclonic epilepsy of infancy. *Neurology*