

Communication Profile of de Lange Syndrome

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ABSTRACT

Introduction

de Lange Syndrome is an autosomal dominant disorder which was initially described by Vrolik (1849) in a child with severe oligodactyly.

Case Report

A case of 2y2m/Female child came for speech and hearing evaluation. Audiological examination (BOA, DPOAE, ABR/BAER) and Speech-Language Evaluation, Psychological Examination were done by expertise clinicians. Previously, the child was diagnosed with de Lange Syndrome. Audiological evaluation diagnosed the child to have moderately severe hearing loss. After speech and language evaluation the child was detected with speech language disorder. After psychological evaluation the child was diagnosed with severe developmental delay and having low IQ range.

Discussion

de Lange syndrome is an autosomal dominant developmental disorder characterized by the sample number of characteristics which requires more rehabilitation options have to be planned. Unfortunately, there is dearth of literature that addresses the characteristics, assessment and intervention of individuals with de Lange syndrome.

Keywords

De Lange Syndrome; Communication Disorders

In the 1930s Cornelia de Lange, a Dutch pediatrician reported two unrelated girls with similar features and named the condition “degeneration typus amstelodamnesis”.^{1,2} In honor of her formal characterization the term de Lange syndrome is widely used.

de Lange syndrome is a syndrome of congenital anomalies mainly characterized by a prenatal and postnatal growth deficiency; Intellectual disability that usually ranges from moderate to severe; Hearing loss; distinctive facial appearance including arched eyebrows that is often meet in the midline (synophrys), low set ears, small and widely spaced teeth and a small upturned nose; a psychomotor delay; Hirsutism; feeding difficulties; behavioral problems and upper limb defects were also evident. Jackson et.al (1989)³ clinically review 310 patients and reported that 33% of these children were delivered prematurely and intrauterine growth retardation was also evident in many children.

de Lange syndrome is a very rare disorder that is

apparent at birth (congenital). Males and females appear to be affected in equal numbers. It has been estimated that de Lange syndrome occurs in approximately one in every 30,000 to one in every 50,000 live births.⁴

Case Report

A retrospective case study design is adopted for the present study. A 2 year 2 months old male child reported with a complaint of speech & language problem and reduced hearing sensitivity from both the ears. A detailed case history was taken which reveals that baby was born as premature and suffered with jaundice at birth and all the developmental milestones were also delayed.

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Table I: Report of Behavioural Observation Audiometry

SOUND STIMULATION	INTENSITY	RESPONSE
Clapping	80dBHL	Searching
Name Calling	75dBHL	Searching
Bell	85dBHL	Startle
Drum	75dBHL	Eyeblink
Daphli	80dBHL	No Response
500 Hz	75dBHL	Startle
1KHz	80dBHL	Ceasation of activity
2KHz	85dBHL	No response
4KHz	85dBHL	No response

A detailed Audiological and Speech and language Evaluation has been done.

Audiological evaluation:

As the age of the child was 2years 2months, so as per the protocol for the assessment initially Behavioral Observation Audiometry (BOA)⁵ was performed. (Table 1)

This report (Table I) indicates that child responds to all verbal stimulus (such as speech cues, name calling) and nonverbal sound stimulus (such as Drum, Bell, Daphli, frequency specific warble tones) between the intensity of 75- 85 dBHL and the responses were eye widening, searching, cessation of activity and startle

responses. BOA responses were indicative of bilateral moderate to moderately severe hearing Loss.

Further the child was recommended for DP-OAE. DP-OAE screening indicated "REFER" for both the ears indicative of abnormal outer hair cell functioning in the cochlea in both ears.

After doing DP-OAE screening, Auditory Brainstem Response (ABR) test was recommended for the child.

ABR test was done by using an instrument with ANSI S3.1-1999 specifications for the background noise. This test was done by using the protocol.⁶ (Table II)

This ABR finding (Fig.1) reveals, identifiable peak V was obtained till 65 dBnHL in right ear which was indicative of moderate hearing loss and identifiable

Table II: ABR Test Protocol

PARAMETER	SELECTION
TRANSDUCER	·ER-3A inserts
TYPE	·Click
POLARITY	Rarefaction
RAMPING (Window)	Blackman
RATE	·Click:21.1/sec



Fig. 1. ABR Report Findings of the Case of de Lange syndrome

peak V was obtained till 75 dBnHL in left ear which was indicative of moderately severe hearing loss.

Speech language evaluation:

The primary mode of communication was predominantly nonverbal through crying no true word or vocalizations were present.

On OPME examination, all articulators are normal in appearance and functions except the tongue elevations and lateral movements are affected. All the vegetative skills such as blowing, chewing are also affected except sucking.

For assessing receptive expressive language skills REELS was administered on the patient which reveals receptive and expressive language age of the patient is 0-3 months. Level of expression was also which reveals intentional communication behavior for both vocal and motor gestural.

Cognitive prerequisites for language development showed poor skills for all aspects of language development.

COMM DEALL shows delay in all aspects of language development(i.e. Gross motor, Fine motor,

Activity of Daily Living, Receptive, Expressive, Social, Cognitive and Emotional skills all are delayed).

Psychological evaluation:

Mild Developmental Delay was reported secondary to de Lange syndrome (DQ=52).

EEG reports revealed normal sleep record.

MRI of brain reports revealed no detectable significant



Fig.2. The child with de Lange Syndrome

Table III: Comparison of ranges between unaided and aided responses

SOUND	INTENSITY (UNAIDED)	RESPONSE (UNAIDED)	INTENSITY (AIDED)	RESPONSE (AIDED)
Clapping	80dBHL	Searching	50dBHL	Searching
Name Calling	75dBHL	Searching	50 dBHL	Eye Blink
Bell	85dBHL	Startle	45 dBHL	Eye widening
Drum	75dBHL	Eyeblink	45dBHL	Startle
Jhankara	80dBHL	No Response	50dBHL	Searching
500 Hz	75dBHL	Startle	45 dBHL	Ceasation of activity
1KHz	80dBHL	Ceasation of activity	50dBHL	Startle
2KHz	85dBHL	No response	50 dBHL	Eye Blink
4KHz	85dBHL	No response	50 dBHL	Head movement

abnormality with both the hemispheres were normal in appearance and show abnormal signal intensity, centrum semi ovale and white matter tracts are normal, corpus callosum is normal in size and shape with normal signal pattern, sulci and gyri are normal, cisterns ventricle are normal with septum in the midline, no evidence of hippocampal sclerosis is seen.

Gene karyotyping was done, reports revealed translocation between chromosomes 1 and 4. Breakage and reunion have occurred at the bands 1q21 and 4q31.3. The segments distal to them have been exchanged i.e. 46, XY, t (1; 4) (q21; q31.3).

Clinical features: clinical examination of the patient reveals following features: Developmental delay; Synophrys: thin eyebrows often meet in the midline; Long eyelashes; Thin downturned lips; Oligodactyly; Low set ears; Hearing loss; Communication delay; Hirsutism: excessive hairs on head and back; small widely spaced teeth, small upturned nose. (Fig.2)

ABR responses of the child revealed moderate hearing loss in the Right ear and moderately severe hearing loss in the left ear. For management, regular use of the hearing aid is recommended to the child which is followed by the Hearing Aid Trial. Patient was recommended for Hearing Aid Trial. (Table III)

BTE hearing aid was tried which was given under

the ADIP Scheme or Assistance to Disabled Persons for purchasing/ fitting of aids/ appliances. Where, the patient (Aided) responds to the entire verbal and non- verbal sound stimulus between the intensity of 40- 50dBHL. Responses were eye widening, searching, cessation of activity and startle. (Table III)

For management, firstly hearing aids were given to the child with proper programming. Unaided and aided responses were recorded (Table III) to check functional gain of hearing aid and ling six sounds test were to check the child's hearing skills. (Table IV)

Discussion

The aim of the study is to know the Audiological, Speech Language characteristics and clinical features for the individuals with de Lange syndrome. Due to this rarity speech and language features of de Lange syndrome lack literature. This study may help to investigate the specific clinical features, assessment, and management protocol best suited for patients with de Lange syndrome.

de Lange syndrome is a dominantly inherited developmental disorder characterized by the sample number of characteristics which requires more rehabilitation options have to be planned. Unfortunately, there is dearth of literature that addresses

Table IV: Comparison of ranges between unaided and aided responses in six ling sound test

SOUNDS	WITHOUT LIP READING				WITH LIP READING: AIDED			
	UNAIDED RESPONSE		AIDED RESPONSE		UNAIDED RESPONSE		AIDED RESPONSE	
	3ft	5ft	3ft	5ft	3ft	5ft	3ft	5ft
/a/	×	×	√	√	√	×	√	√
/e/	×	×	√	√	√	×	√	√
/u/	×	×	√	√	√	√	√	√
/s/	×	×	×	×	×	×	×	×
/sh/	×	×	×	×	×	×	×	×
/m/	×	×	×	×	×	×	√	√

the characteristics, assessment and intervention of individuals with de Lange syndrome. Therefore more clinical empirical studies are needed to confirm and understand speech & language characteristics in individuals with de Lange syndrome.

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