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Communication profile of a child with Ring chromosome 14 syndrome: A case study

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Abstract
Introduction: Ring 14 syndrome is a rare genetic condition due to the realignment of fourteenth chromosome into ring-like structure. Intellectual disability and language disorders were observed in children with ring syndrome.
Aim: The aim of the study was to document the communication profile of a child with a diagnosis of Ring chromosome 14.
Method: A detailed speech, language, and communication assessment was done for the child using informal and formal methods of evaluation.
Results and Discussion: The Child was diagnosed to have Receptive and expressive language disorder secondary to Ring chromosome 14. Also, deficits in oro-motor skills were observed.
Conclusion: The present study would help students and professionals understand the communication profile of a child with Ring 14 syndrome and additionally help in the rehabilitation process.

Keywords: Ring 14 chromosome syndrome, communication, speech, language

Introduction

Ring 14 syndrome is a rare genetic condition due to the realignment of fourteenth chromosome into ring-like structure. In an affected individual, the typical karyotype is 46, XY or XX, r(14). The ring formation requires breakpoints of two chromosome, first on the short arm and second on the long arm. The former has obtained little scrutiny since it happens within the heterochromatin of the short arm, devoid of protein-coding genes. The latter is more relevant, which cause losing of the gene-rich terminal band of the long arm. Comparative genomic hybridization assay is usually used to detect the deletion, differing in size between 0.3 and 5 Mb. Although, in few cases the deletion is too small to be detected by comparative genomic hybridization and the ring seems to be "complete" (Zollino *et al.*, 2012) [1]. In 1971, Gilgenkrantz *et al.* [2], for the first time described about this syndrome.

The clinical characteristic feature of Ring 14 syndrome includes shortness of stature, microcephaly, ocular abnormalities especially modified retinal pigmentation, atypical strabismus and macula, intellectual disability associated with aggressive and hyperactive behavior in few cases and pharmacoresistant epilepsy (Zollino *et al.*, 2012) [1]. Dysmorphic features consist of high and prominent forehead, widely spaced eyes with epicanthus and blepharophimosis, an elongated face along with puffy cheeks, a flat nasal bridge associated with a prominent nasal tip and low-set ears (van Karnebeek *et al.*, 2002; Zollino *et al.*, 2009) [3, 4].

During the first year of life, Epilepsy onset usually occur with a high variety of seizure types. Seizures can be generalized tonic– clonic, minor motor and complex partial (Battaglia & Guerrini, 2005) [5]. The interictal EEG pattern may be normal or described by multifocal spikes as well as spike-and-wave complexes. Seizures are usually drug-resistant and the outcomes are severe (Bahi-Buisson *et al.*, 2005) [6]. Rearrangement of chromosome 14 in the form of Ring shaped is frequently linked with a deletion of the terminal 14q region (Van Karnebeek *et al.*, 2002; Zollino *et al.*, 2009) [7, 8]. Few studies reported that children with ring 14 chromosome syndrome (both with and without deleted material) generally appear more impaired compared to children with linear 14q deletions (Zollino *et al.*, 2009) [8]. This could be because of higher presence of associated disorders, such as a increased occurrence of epilepsy (Van Karnebeek *et al.*, 2002) [7] and autistic traits (Zampini *et al.*, 2014; D'Odorico *et al.*, 2011) [9, 10].

Zollino *et al.* (2012) [1] reported that in their study, Intellectual disability especially of severe degree, was found in all patients with ring 14 chromosome syndrome except in one patient,

who had a non-deleted ring and seizure disorder (mild) as well. Behavior disorders, usually hyperactivity with bursts of aggressiveness occasionally, stereotypic motor movements, such as hand flapping and echolalia, were also observed occasionally.

Zampini *et al.* (2017) [11] described the developmental track of communicative skill of five children with chromosome 14 aberrations. Among the five children with chromosomal 14 aberrations, one child had Ring 14 syndrome. They reported that one child with Ring 14 syndrome had myoclonic seizure, profound intellectual disability, lower cognitive skills, autistic traits, stereotyped behaviors, limited gesture and word productions.

Zampini *et al*. (2012) [12] reported that intellectual disability and language disorders have been found in almost all children with Ring 14 syndrome. There was significant individual variation in their communication abilities (Rinaldi *et al*., 2017) [13]. A survey study was conducted in which it was reported that out of 12 children and young adults with ring 14 syndrome, only four used words to communicate (Zampini *et al.*, 2014) [9].

The existing studies highlight the clinical picture of ring 14 chromosome but with marginal focus on communication skills. Hence, this present case report documents the detailed communication skill of a 7-year-old male child with a diagnosis of Ring chromosome 14.

Case Report

A 7-year-old male child brought by the parents with the complaint of not speaking age appropriately. Detailed case history showed no significant prenatal, and perinatal history, which was mentioned by the parents. The child was born to non consanguineous parents. No history of communication disorders in family was reported. The child was exposed to Tamil and English languages. Most of the time, child was exposed to Tamil which was the mother tongue. The child was attending integrated pre-school where he was exposed to English language. Developmental histories including motor and speech-language milestones were reported to be delayed. He was diagnosed with Ring chromosome 14. He had symptomatic epilepsy. He was under medication for the same. He was right handed. His muscle tone was normal. On Psychological evaluation, his intellectual level was found to be moderate intellectual functioning. On VSMS, his social age was found to be 3 years 4 months. Hearing evaluation indicated normal hearing sensitivity in both ears.

Communication profile

Informal communication assessment based on the mother child interaction was done. The mother was instructed to interact with the child as she do at home using the toys given such as ball, dolls, fruits and vegetable models, kitchen set, story books and this interaction was video recorded for ten minutes. The communicative function predominantly exhibited by the child during parent child interaction was behavior regulation (requesting objects and actions). The communicative gestures that were observed during parent child interaction include pointing, reaching, giving, head shaking to express denial and object recognitory gestures. Few instances of combination of gesture and vocalization (pointing plus vocalization) and combination of gesture and verbalization (giving gesture plus single word utterance) were observed. Child expressed few kinship words and functional words (single word utterance) occasionally during the interaction with his mother.

Speech and language assessment

Detailed speech and language evaluation was done. On oral peripheral mechanism examination, restricted movements of articulators with respect to extended speed and movements was observed. Restricted elevation, lateralization and sweeping movements of tongue were observed. Inadequate pursing of lips and reduced intra oral breath pressure were observed. On pre-linguistic skills evaluation, eye contact present fairly, joint attention was not sustained adequately, inadequate non-verbal and verbal imitation skills were observed.

On language assessment, Comprehension skills include the child can differentiate and identify family members. He can comprehend lexical items such as fruits, vegetables, vehicles. He can comprehend prepositions and colors. He can follow two step verbal commands. He can understand the use of common objects. Expression skills include the child can express kinship words meaningfully. He can express his needs through few words with gestures. He can say few functional words.

Formal language evaluation was carried out using standard measures such as *Assessment of Language development* (ALD; Lakkanna, Venkatesh, & Bhat, 2008) [14], his receptive language skills correspond to 18 – 24 months and expressive language skills correspond to 12 – 18 months.

Diagnosis and intervention

Based on the comprehensive evaluation of communication, speech and language skills using informal and formal tests, the child was diagnosed to have Receptive and expressive language disorder secondary to Ring chromosome 14. Parents were counseled about the importance of intensive speech and language stimulation at home and also, recommended to attend continuous speech and language therapy in order to improve the child's overall communication skills.

Discussion

The child had deficits in intellectual and language skills, which is supported by evidence in literature (Zampini *et al.*, 2017; Zampini *et al*., 2012) [11, 12] who reported that children with Ring 14 syndrome have been found to have intellectual disability and language disorder. Also, this child had difficulties in oro-motor skills especially restricted articulatory movements in relation to extended speed and movements. This child used combination of gesture (pointing) and single word utterance occasionally during interaction. With respect to communication skills of children with Ring 14 syndrome, high individual variability was reported (Rinaldi *et al*., 2017) [13].

Conclusion

We can conclude that Ring 14 syndrome is a rare genetic condition that affects speech, language, and communication skills. To our knowledge, this is the first case study from South India that highlights the speech and language characteristics of a child with Ring chromosome 14. The present study would help students and professionals better understand the communication profile of a child with Ring 14 syndrome and additionally aid in the rehabilitation process.

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