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SPEECH AND LANGUAGE PROFILING AND EFFECT OF VIDEO FEEDBACK INTERVENTION ON 4 YEAR OLD CHILD WITH SEGAWA SYNDROME

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ABSTRACT

An irregular gait (clumsy or uncoordinated walking) and dystonia (uncontrollable shaking) are two of the characteristics of Segawa syndrome, which is a rare genetic condition. Generally, it is possible that the symptoms of Segawa syndrome are remarkable with diurnal fluctuations, particularly in adolescents and adults. Well-known clinical illustrations of dopaminergic deficits seen in Segawa syndrome affecting speech and language control in patients range from a fundamental inability to control motor aspects of speech production will result in moderate to profound cognitive language impairments, which in turn formulate difficulties with phonological processing, syntactic complexity and language comprehension. VFI is a technique aimed at fostering positive parenting that has been shown to be beneficial for child development and parent-child interaction in a variety of settings, including at-risk and clinical populations, in varied ways. The concept of promoting more cost effective assistance for people with special needs entrusts the major responsibility for therapeutic administration to the parents, in the home environment, in contrast to the typical service delivery paradigm. The aim of the present study is to profile about the speech and language characteristics of a 4-year-old child with Segawa Syndrome and the effect of video-feedback intervention given, in improving parent-child interaction, after six months of treatment.

Key-words-Syndrome, Intervention, Segawa, Video-Feedback

INTRODUCTION

Segawa et al. initially described dopa-responsive dystonia (DRD) in 1976. It is often referred to as 'hereditary progressive dystonia with diurnal changes' or 'Segawa's syndrome'. It is not uncommon for this uncommon disorder to be misdiagnosed as hysteria, hereditary spastic paraparesis, or cerebral palsy due to a lack of awareness. Segawa syndrome is a rare genetic illness marked by a clumsy or uncoordinated method of walking (abnormal gait), as well as dystonia (uncontrollable shaking). Generally speaking, dystonia is a word that refers to a collection of muscle disorders that are characterized by involuntary muscle contractions that cause the body to move and position itself in unnatural, sometimes painful, ways (postures). Dystonia in Segawa syndrome mainly affects the legs, however in some cases, dystonia in the arms may be the initial sign of the disorder. It is possible that the symptoms of Segawa syndrome, particularly in adolescents and adults, will become noticeably worse or more visible in the afternoon and evening hours as opposed to the morning hours on occasion (marked diurnal fluctuation).

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When levodopa is administered to children with Segawa syndrome, they often experience a significant and long-lasting improvement. It is an amino acid that can be turned into dopamine, a brain molecule that acts as a neurotransmitter, through the process of oxidation. Children with Segawa syndrome have low levels of dopamine in their bodies. Mutations in the GCH-1 gene are the root cause of the illness. The GCH-1 gene mutation is passed down through families in an autosomal dominant manner.

In the speech system, recent studies have shown that dopaminergic transmission might influence synchronic linguistics speech process (Tettamanti et al., 2005), verbal long-term memory and word generation (Cervenka et al., 2008). On the opposite hand, the role of dopaminergic neuromodulation in traditional speech control remains unknown.

Well-known clinical illustrations of dopaminergic deficits seen in Segawa syndrome affecting speech and language control in patients range from a fundamental inability to control motor aspects of speech production (e.g., monotone, hypotonic speech with reduced loudness and pitch, decreased accuracy of articulation; moderate to profound cognitive language impairments (e.g., difficulties with phonological processing, syntactic complexity and language comprehension.(Heinz, 1998; Kataoka and Ueno, 2010; Ludlow, 2008; Thompson., 2009).

VFI is a technique aimed at fostering positive parenting that has been shown to be beneficial for child development and parent-child interaction in a variety of settings, including at-risk and clinical populations, in varied ways. VFI is increasingly being used with parents of children with neurodevelopmental impairments (ND; for example, cerebral palsy, sensory and/or psychomotor delay, and genetic abnormalities)Provenzi et al (2020).

The present study focuses on describing the speech and language characteristics of a four year old child with Segawa Syndrome and the effect of video-feedback intervention given, in improving parent-child interaction.

REVIEW OF LITERATURE

While significant progress has been made in the understanding of mother-child interactions (Bornstein, 1989; Cyr, Dubois-Comtois, & Moss, 2008; Gartstein, Crawford, & Robertson, 2008; Landry, Smith, Swank, & Miller-Loncar, 2000),In the context of families parenting children with particular developmental disorders, relatively little of this information has been developed. However, in recent years, parenting behaviour in the context of a developmental disability has gained attention (e.g., Ruble, McDuffie, King, & Lorenz, 2008; Rutgers, van IJzendoorn, Bakermans-Kranenburg, et al., 2007; Warren & Brady, 2007).As a crucial variable in the study of unexceptionally developing newborns, maternal responsiveness or sensitivity has recently inspired attention in the research of children with syndromes and disorders of developmental delay (Slonims, Cox & McConachie, 2006; Venuti, Giusti, & Bornstein, 2008; Warren & Brady, 2007)..At the very least, maternal responsiveness has the potential to influence language development, which is critical for all children but especially those with developmental disabilities or syndromes, such as autism, that can delay or prevent language development (Baker, Messinger, Lyons & Grantz, 2010.)

The inheritance of rare single gene illnesses is typically characterized by Mendelian patterns of inheritance. Autosomal dominant diseases, autosomal recessive conditions, and X-linked disorders are among the many types of genetic abnormalities. Hannah Blencowe et al., (2018)developed a method for estimating the burden of these disorders in children up to the

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age of five in areas with limited empirical data as non-communicable diseases. This approach uses population-level demographic data, combined with assumptions based on empirical data from settings with data available, to provide population-level estimates which programme policy-makers or the professionals, when planning services can use.

AIM - The aim of the present study is to profile about the speech and language characteristics of a 4 year old child with Segawa Syndrome and the effect of video-feedback intervention given, in improving parent-child interaction

METHODOLOGY-

The initial evaluation in the department of speech and language diagnostics was profiling the speech and language procurement of the child using proforma for childhood language disorders and standardized test materials such as REELS-3,3- Dimensional – Language acquisition test, Reynell's attention scale and Westby play scale.

The child was enrolled for video-feedback intervention, with specific interest in meeting parent child interaction atreceptive and expressive language, speech sound production and social communication skills. Schedule of regular sessions and time bound goals were set to score the prognosis post 6 months. Questionnaire modified from the study - Efficacy of Parent Child Interaction and Parental Self Esteem with a Video Feedback Intervention in Children with Autism Spectrum Disorder (Mohan, Kumar and Mohan, 2020) was administered to check improvement and parental satisfaction and standardized diagnostic materials were used to score pre and post therapy administration.

Parents were asked to record the administration of the techniques within the natural environment of the child. The therapist acted as the guider and the parents were asked to analyze and reflect on video-clips of their own interactions, with the child.

Questionnaire was administered post six months. Parents were asked to rate each question between 1 to 5 on a visual analog scale, where 1 was strongly disagree and 5 was strongly agree.

RESULTS AND DISCUSSION

The child was referred from XXX hospital for speech and language intervention followed by the reported delay in language acquisition at the age of 1.9 years. Her last evaluation in this hospital was at the age of 10 months for genetic study revealing the presence of Segawa and the child was under regular medication of levodopa and carbidopa with increase in dosage on regular visit for the medical conditions and sleeping disturbances. Her parents reported that their main concern that day is regarding her communication skills.

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She is the first child of consanguineous second-degree parentage with significant natal history such as oligohydramnious, preterm caesarian delivery with significantly low birth weight of 1.9kg and delayed birth cry. She was also treated for neonatal hypoxia under specialized ICU for about 2 days soon after birth. Detailed profiling of the child eliminated all other causes for possible congenital abnormalities or impairments except the genetic disturbance occurred. She presented with a significant delay in expressive language and cognition - delay of 1 year, and barely noticeable delay of 3-5 months in receptive language. Considering diagnostic criteria of overall noted delay in motor and speech milestones with significant medical and natal history, dystonia of the foot, with normal play scales and attention with emerging pragmatic skills she was along with the presence of professionally diagnosed as an individual with global developmental delay secondary to Segawa syndrome. Further the client was recommended for a multi-disciplinary management of Speech and language therapy, Physiotherapy, along with ongoing medications focusing in improving overall quality of life. The figure 1 shows the pre and post therapeutic scores obtained on standardized language tests and Figure 2 shows graphical representation of prognosis.

The child was enrolled for video feedback intervention from home aiming improvement in parent child interaction. The preliminary goal for therapy was set to improve the functional communication of the child, with the therapist explaining about the speech and language stimulation techniques and its demonstration.

Parents reported increased rating for question 1, which indicated that there was an increased vocabulary level, after video feed-back, Question 2 which reported that there was significant improvement in social interaction, question 3 which indicated reduction in parental anxiety regarding communication skill, Question No:4, indicating parents' ability to encourage language development through video feedback and parent child interaction, Question No: 6, which indicated the improvement in parental self- esteem through video feedback and parent child

Pretherapy at 3.5 years of age	Post therapy at 4 years of age
3D LAT <i>Language comprehension</i> -Group VI, 24-26 months (Understands whose and what happened, where questions with spatial relationships and simple how questions evaluating quality, but do not comprehend how questing evaluating cause) <i>Language Expression</i> -Group II, 12 -14 months (Begun to express needs and naming 4-5 lexical items, but not started saying words to indicate actions/event as well as repeating after the other) <i>Cognition</i> : Group IV, 18-20 months (child has started to use actual words, memorizes past events and replies to questions)	Extended REELS <i>Receptive language</i> Age- 36-42 months (3-3.5 years) (Comprehends numerical markers, and first-order embedded sentences) <i>Expressive language</i> age- 27-30 months Can express self by using pre spontaneous speech, express prima correctly, and can count up to ten) <i>Cognition (3D LAT)</i> : Group IX, months 3 (presents with concept of though work in the family, and correction while talking, however not with future plans)
Reynell's Attention Levels and Strategies Stage 2; 1-2 years (maintains single channeled attention and ignore external stimulus)	Stage 4; 3-4 years (Maintains single c attention and able to shift between ta
Developmental Play scale <i>Play</i> - Symbolic level I; 17 to 19 months (Has some concept on tool use, hidden objects and life like props at everyday familiar situations with coherence at short isolated schemas even with self-representation) <i>Communication</i> - Phase 1; pre-symbolic level II; 13-17 months (Uses context related single words and exhibit requesting, protesting, labeling, greeting and commanding)	<i>Play</i> -Symbolic level V; 3 years (does play with modification, involving s role play and associative play) <i>Language</i> -Symbolic Level III; 2 ye gives comments on self-activity and simple sentences containing phra emerging morphological markers)

Figure 1

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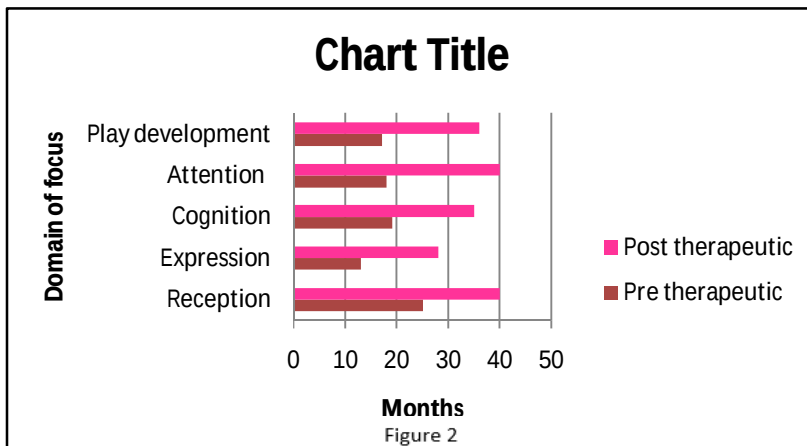
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interaction. Question 5 specifically addresses the effect of the feed-back interaction from father's perspective.

Father reported that the intervention increased their knowledge of their child's ideas and feelings, as well as their communication and relationship with their infant. The reply from fathers was overwhelmingly positive. The ability to conduct sessions at home (or at dads' places of work) and the ability to schedule sessions according to father's schedules were highlighted as critical to successful delivery. The finding is in agreement with the study done by Lawrence, Davies and Ramchandani (2012) which suggested that father's involvement in interventions enhances its effectiveness.

CONCLUSION



Parent child interaction is the soul of communication that nourish the innate ability of verbal mode of message transmission for a normal hearing child with special needs. Governments are promoting a more cost-effective type of speech and language assistance that is delivered in the

home. This concept entrusts the major responsibility for therapeutic administration to the parents, in the home environment, in contrast to the more typical service delivery paradigm in which the speech-language pathologist is the primary agent of change. That is, the parent-centered with professional support (which is also considered the best practice) is now slowly replacing the therapist-centered model.

FUTURE WORK & SUGGESTIONS-

The present study is a single-case study. The effect of Video-feedback intervention on improving communication skills and improving parent-child interaction can be assessed, on rarely occurring syndromes.

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APPENDIX

On a scale from 1 -5 please rate the following statement about the efficacy of parent child interaction video feedback and self esteem.

1= Strongly disagree

2 = Disagree

3 = Neutral

4 = Agree

5 = Strongly agree

1.வீடியோகருத்துதலையீட்டில்கலந்துகொண்டபிறகு

என்னுடையகுழந்தையின்சொல்வளம்அதிகரித்துள்ளது.

2.வீடியோகருத்துதலையீட்டில்பங்கேற்றதற்குபிறகு

என்னுடையகுழந்தையின்சமூகத்திறன்கள்அதிகரித்துள்ளது.

3.வீடியோகருத்துதலையீட்டில்பங்கேற்றதற்குபிறகு

எனதுகுழந்தையின்மொழிவளர்ச்சிகுறித்தகவலைஎனக்குகுறைந்துள்ளது.

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- 4.வீ டியோகருத்துதலையீட்டில்பங்கேற்றதற்குபிறகு
என்னுடையகுழந்தையின்மொழிவளர்ச்சியைஊக்குவிக்கும்திறன்
எனக்குஅதிகரித்துள்ளது.
- 5.வீ டியோகருத்துதலையீட்டின்பங்கேற்றத்திற்குப்பிறகுமனம்அழுத்
தம்குறைவதோடும்ட்டுமல்லாமல்தந்தைக்கும்குழந்தைக்கும்இடை
யிலானதொடர்பும்அதிகரித்துஉள்ளதா
- 6.நான்,வீ டியோகருத்துதலையீட்டில்பங்கேற்றதால்என்னுடையசுய
திறனைமேம்படுத்தமுடிந்தது.