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PROFILING SPEECH, LANGUAGE AND SWALLOWING CHARACTERISTICS IN INFANTILE POMPE DISEASE – A SINGLE CASE STUDY

Clinical Research

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ABSTRACT

Pompe disease is a rare progressive neuromuscular genetic disorder caused by the deficiency of enzyme Acid Alpha-Glucosidase (AAG), that accumulates lysosomal glycogen in multiple tissue and cell types which affects skeletal, cardiac and smooth muscle cells. This case report discusses about a child who is diagnosed with Pompe disease who presented with various speech, language and swallowing difficulties. Assessment and management of speech, language and swallowing functions of the child were documented.

KEYWORDS

Pompe disease, Speech, Language, Swallowing

INTRODUCTION

Pompe disease (PD) is a rare progressive neuromuscular genetic disorder. The incidence of the disease is approximately 1 in 140,000 for infantile GSD II and 1 in 60,000 for adult GSD II (Nivargi, Kulkarni & Makhale, 2016). It is a deficiency of the enzyme Acid Alpha – Glucosidase (AAG) that hydrolyzes lysosomal glycogen (Kishnani, 2004). It results in problem to heart and skeletal muscles due to the deficiency of AAG which leads to generalized glycogen accumulation (Fecarotta et. al, 2013). The phenotype of the disease ranges from early-onset forms to late-onset forms. It is classified as the classic infantile form, which is the common form of PD presents within the first month of life with progressive hypotonia, hypertrophic cardiomyopathy, macroglossia, feeding difficulties, recurrent respiratory infections, and failure to thrive. If it is left untreated, it leads to death. Non-classic infantile-onset forms leads to limited to ECG abnormalities/heart problems and less mortality rate. In the late-onset forms symptoms may present at any age from childhood to late adulthood, cardiac involvement is generally absent and the clinical manifestations are mostly related to skeletal muscle involvement, with progressive motor impairment, respiratory failure, and premature death (Fecarotta et. al, 2013).

Individuals with Pompe disease are commonly reported to have swallowing as well as respiratory difficulties. Major developmental milestones, both motor and speech, are either delayed or not achieved. The mental development is usually normal. Hearing can be affected in most of them (Reuser, 2017).

Dysphagia is the major concern in individuals with PD. This complication impact on the quality of life of these individuals and this also lead to malnutrition, dehydration, weight loss and failure to thrive. In addition, aspiration may also occur which increase the risk of respiratory infections, ineffective cough, due to the involvement of respiratory muscles and contributes to the risk of aspiration which leads to pneumonia (Fecarotta et. al, 2013; Swift et. al, 2016). Enzyme Replacement Therapy (ERT) is the only effective treatment available for PD. The long term effects of ERT on dysphagia were being explored in the recent past. No significant improvements in oral feeding were reported in this population with ERT (Swift et. al, 2016).

CASE REPORT

A 7 year old female child was accompanied by her parents to the department with the complaint of aspiration during liquid consumption. The child is diagnosed with Pompe disease by a molecular genesist.

Medical history:

The child has been undergoing ERT. She has a History of Otitis media with effusion, bilateral myringotomy and grommet insertion was done on 9/11/16. Parents reported history of reduced hearing sensitivity in both ears. The child has scoliosis and abnormal gait. She requires assistance for her routine.

Oral Peripheral Mechanism Examination

	Structure	Function
Lips	Normal	Pursing, puckering, rounding, spreading were observed to be inadequate
Tongue	Normal	Protrusion, elevation, retraction and lateralization: inadequate Lip seal: Inadequate (drooling present)
Teeth	Underbite	Biting and chewing: inadequate
Jaw	Normal	Restricted range of motion
Hard palate	High arched	
Soft palate	Normal	

Impression: Malocclusion (underbite) of teeth, high arched palate was observed. Functions of articulators were observed to be inadequate.

Vegetative skills

Blowing, sucking, biting, chewing and swallowing were observed to be inadequate.

Speech Profile:

Speech subsystems

Respiration: The child's breath support for speech and non speech activities was observed to be affected. She was observed to have thoracic pattern for breathing.

Phonation: On perceptual analysis, loudness of voice was inadequate, pitch and quality could not be assessed.

Articulation: The child's articulation could not be assessed but as observed she had weak pressure consonants with reduced loudness in her speech.

Prosody: Could not be assessed since the child was not able to produce phrases or sentences with appropriate pitch and loudness

Language Profile

Informal assessment of language

Reception: The child follows simple one step commands. She can understand lexicals such as nouns (vegetables, fruits, animals, vehicles, common objects), which the child predominantly exposed to on her daily living basis. She also comprehends simple verbs, pronouns, prepositions, adjectives, 'wh' questions – what, which, who, where, time concept, colours, numbers and shapes.

Expression: The child predominantly communicates verbally using one word utterances. Attempts to use 2 – 3 word utterances on demand. She also uses gestures such as head nodding appropriately for simple questions.

Swallowing Profile:

Informal assessment of swallowing was carried out using food consistencies such as liquid and semi solids.

Symptoms	Food consistency	
	Thin liquids	Semi-solids
Drooling	Present (occassionally)	Present (occassionally)
Tongue moves forward to start swallow	Absent	Absent
Delay in oral transit time	Present	Present
Reduced chewing, delayed or absent rotary chewing	Not applicable	Present
Reduced tongue co-ordination	Present	Present
Repeated swallows per bolus	Present	Present
Gag reflex	Absent	Absent

Swallowing with solid food consistency could not be assessed as the child had difficulty in chewing due to muscle weakness

Impression: (?) Oropharyngeal Dysphagia

Treatment

The child attended demonstration therapy for a week with each session lasting for a duration of 45 minutes/day. Following which a home training plan was given to the parents for speech and swallowing difficulties.

Goals worked on

Swallowing and communication therapy aimed at improving the child oro-motor strength, range of motion of articulators, improving loudness, breath support, compensatory strategies and postural & diet modifications for swallowing, and alternative means for communication.

Goals for swallowing:

1. Worked on improving the child's breath support for speech and non-speech activities
2. Worked on improving the muscle strength (isotonic) and range of motion (isometric) of oral structures using oro-motor exercises
3. Assisted in improving the child's swallowing functions using compensatory (multiple swallow) and postural strategies (head extension)

Goals for communication:

4. Aided in improving the communicative abilities of the child for basic needs, wants and to convey emotions using Alternative and Augmentative Communication (high tech device)

Treatment progress could not be evaluated since the client attended minimal sessions. There is still a dearth in literature regarding the prognosis of individuals with Pompe Disease post treatment who are undergoing ERT and other rehabilitative measures.

CONCLUSION:

Children with Pompe disease are characterized by various swallowing and communication difficulties due to their muscular degeneration. Oro-pharyngeal dysphagia is a commonly reported symptom in children with Pompe disease (Jones et al, 2009; Swift et al, 2017). The present case study also profiled similar findings related to oro-pharyngeal dysphagia. Communication is also severely affected in these individuals. Appropriate training such as AAC helps these individuals to communicate despite their communicative difficulties, since they have good receptive skills. Future directions can include further investigations regarding the prognosis and treatment effects of swallowing and communication therapy in individuals with Pompe Disease.

Limitation of the study

1. Formal evaluation for speech, language and swallowing was attempted but could not be completed due to the child's fatiguing condition.
2. Since the child attended limited number of therapy sessions prognosis could not be determined.

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