



CHARGE SYNDROME: CASE REPORT

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OBJECTIVE: To describe a case with clinical features of Charge Syndrome that underwent speech therapy. CLINICAL REPORT: This study describes the characteristics of a female patient treated at HRAC for bilateral cleft lip and palate who presented with Charge syndrome. Patient's phenotypic characteristics included: prominent eyes, short neck, prominent frontal, small ears, heart disease, short stature, a delay in the development of motor skills, small nails, and hearing loss. In the initial clinical assessment the patient had mixed hearing loss with deep loss on the right ear and severe on the left ear. Intelligibility of speech was impaired by severe hypernasality, presence of compensatory articulation, as well as receptive and expressive language below expectations for her age. The treatment emphasized the hearing rehabilitation with bilateral hearing aid fitting and the oral communication using pharyngeal bulb for management of velopharyngeal insufficiency. Speech therapy addressed both, phonetic-phonological aspects and velopharyngeal function for speech. Sensory cues were used as strategies to facilitate speech production. After 155 sessions of therapy the patient showed improvement in speech intelligibility, particularly within therapy but maintained use of as systematic compensatory articulation during spontaneous speech. **CONCLUSION:** The patient had significant improvement of oral communication after the speech therapy intervention with significant improvement in quality of life.

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