

Case Report

Oropharyngeal dysphagia and language delay in partial trisomy 9p: case report

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ABSTRACT. The phenotype of partial trisomy 9p includes global developmental delay, microcephaly, bulbous nose, downturned oral commissures, malformed ears, hypotonia, and severe cognitive and language disorders. We present a case report and a comparative review of clinical findings on this condition, focusing on speechlanguage development, cognitive abilities and swallowing evaluation. We suggest that oropharyngeal dysphagia should be further investigated, considering that pulmonary and nutritional disorders affect the survival and quality of life of the patient. As far

as we know, this is the first study of a patient with partial trisomy 9p described with oropharyngeal dysphagia.

Key words: Deglutition evaluation; Language evaluation; Deglutition disorders; Language delay; Partial trisomy 9p