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Phoniatric assessment in Kabuki Syndrome and its contributions to an individualized approach to school learning challenges: Case report

Avaliação foniátrica na Síndrome de Kabuki e suas contribuições para abordagem individualizada dos desafios de aprendizagem escolar: Relato de caso

Evaluación foniatrica en el Síndrome de Kabuki y sus contribuciones a un abordaje individualizado de los retos del aprendizaje escolar: Reporte de caso

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Abstract

Introduction: Kabuki syndrome is a rare disease with variable clinical manifestations, often associated with intellectual disability and comorbidities that impact academic performance. Phoniatric assessment can provide an individualized perspective, assisting in the management of learning difficulties in these patients. **Objective:** To report a case of Kabuki syndrome evaluated in a phoniatric outpatient clinic, highlighting the role of phoniatric assessment in directing individualized interventions that consider important skills for school learning. **Method:** A phoniatric assessment was performed on a child diagnosed with Kabuki syndrome, referred due to school difficulties. **Results:** The assessment identified specific auditory perceptual deficits that affected academic performance, in addition to visual skills that could facilitate learning. These findings guided the development of personalized management strategies. **Conclusion:** Kabuki syndrome presents great clinical variability. In the reported case, the phoniatric assessment contributed to the identification of altered auditory and visual perceptual skills that facilitate school learning, directing therapeutic interventions and school adaptations.

Keywords: Learning Disorders; Human Genetics; Academic Failure; Learning; Child Development.

Resumo

Introdução: A síndrome de Kabuki é uma doença rara com manifestações clínicas variáveis, frequentemente associada a deficiência intelectual e comorbidades que impactam o desempenho acadêmico. A avaliação foniátrica pode oferecer uma perspectiva individualizada, auxiliando no manejo das dificuldades de aprendizagem nesses pacientes. **Objetivo:** Relatar um caso de síndrome de Kabuki avaliado em ambulatório de foniatria, destacando o papel da avaliação foniátrica no direcionamento de intervenções individualizadas que considerem habilidades importantes ao aprendizado escolar. **Método:** Realizou-se avaliação foniátrica em uma criança com diagnóstico de síndrome de Kabuki, encaminhada devido a dificuldades escolares. **Resultados:** A avaliação identificou déficits perceptuais auditivos específicos que afetavam o desempenho acadêmico, além de habilidades visuais que poderiam facilitar a aprendizagem. Esses achados orientaram a elaboração de estratégias de manejo personalizadas. **Conclusão:** A síndrome de Kabuki apresenta grande variabilidade clínica. No caso relatado, a avaliação foniátrica contribuiu para a identificação de habilidades perceptuais auditivas alteradas e visuais facilitadoras do aprendizado escolar, direcionando intervenções terapêuticas e adaptações escolares.

Palavras-chave: Transtorno de Aprendizado; Genética humana; Insucesso escolar; Aprendizagem; Desenvolvimento infantil.

Resumen

Introducción: El síndrome de Kabuki es una enfermedad rara con manifestaciones clínicas variables, a menudo asociada con discapacidad intelectual y comorbilidades que afectan el rendimiento académico. La evaluación foniatría puede proporcionar una perspectiva individualizada, ayudando en el manejo de las dificultades de aprendizaje en estos pacientes. **Objetivo:** Reportar un caso de síndrome de Kabuki evaluado en una clínica ambulatoria de foniatría, destacando el papel de la evaluación foniatría en la orientación de intervenciones individualizadas que consideran habilidades importantes para el aprendizaje escolar. **Método:** Se realizó una evaluación foniatría a un niño diagnosticado con síndrome de Kabuki, derivado por dificultades escolares. **Resultados:** La evaluación identificó déficits perceptivos auditivos específicos que afectaron el rendimiento académico, además de habilidades visuales que podrían facilitar el aprendizaje. Estos hallazgos orientaron el desarrollo de estrategias de manejo personalizadas. **Conclusión:** El síndrome de Kabuki presenta una gran variabilidad clínica. En el caso reportado, la evaluación foniatría contribuyó a la identificación de habilidades perceptivas auditivas y visuales alteradas que facilitan el aprendizaje escolar, orientando intervenciones terapéuticas y adaptaciones escolares.

Palabras clave: Trastorno del Aprendizaje; Genética humana; Fracaso escolar; Aprendizaje; Desarrollo infantil.

Introduction

Kabuki syndrome (or Niikawa-Kuroki syndrome), first described in Japan in 1981[1], has a prevalence of 1:32,000 and is considered a rare congenital disease. Since its discovery, the number of diagnoses has been progressively increasing, allowing for a better characterization of the clinical presentation[2,3].

The two genetic types (type 1 due to variants in *KMT2D* and type 2 in *KDM6A*) share five main features: (1) dysmorphic facial features (100% of cases); (2) skeletal anomalies including fifth-finger brachydactyly and/or vertebral deformities (92%); (3) dermatoglyphic abnormalities with prominent digital pads (93%); (4) mild to severe intellectual disability (92%); and (5) postnatal growth retardation (83%). Additionally, mixed or conductive hearing loss is frequently observed (up to 65% of cases)[4].

Affected children face complex challenges in language and academic development, arising not only from cognitive and hearing impairments but also from the psychosocial impact of dysmorphic facial features[5]. Recent studies highlight the heterogeneity of neurocognitive profiles, reinforcing the need for individualized assessments[2,3,6,7,8,9].

In this context, phoniatric assessment can help identify both deficits and strengths, guiding multidisciplinary interventions and personalized school adaptations. This study reports a case in which phoniatric evaluation contributed to the planning of individualized strategies.

Case presentation

J.P.M., an 11-year-old boy, was referred to the phoniatrics service by his father due to persistent school learning difficulties. The patient has a confirmed genetic diagnosis of Kabuki syndrome type 1, with a pathogenic variant in the *KMT2D* gene (OMIM 602113), and bilateral moderate mixed hearing loss, using individual sound amplification devices (hearing aids) since the age of 8 with good adaptation.

His history includes preterm birth at 35 weeks due to oligohydramnios, with neonatal ICU stay due to breastfeeding difficulties. During early childhood, he had frequent choking episodes until around age 2. He underwent two surgeries for correction of post-foramen cleft palate: the first at 12 months, and the second at 5 years. Brain MRI performed in 2021 revealed structural changes such as pointed frontal bones (suggestive of trigonocephaly due to early closure of the metopic suture), slight enlargement of the atria of the lateral ventricles, and mild hypoplasia of the bilateral periventricular white matter. He received speech therapy for two years, which was discontinued by the family due to perceived improvement in communication needs.

During the phoniatric evaluation — which included a detailed anamnesis, physical examination, and specific task application — the left tympanic membrane appeared retracted and a central perforation was noted in the right membrane, with adequate use of hearing aids. The patient demonstrated excellent cooperation, sustained attention, and willingness to complete the proposed tasks. In reading tasks, he showed significant difficulties in decoding, which affected fluency, although he retained global comprehension of the text and was able to retell the story accurately.

Spontaneous written production revealed significant challenges: poorly developed cursive writing, immature pencil grip, spelling errors, inappropriate word separation, and inconsistent use of punctuation. However, the richness and coherence of the ideas expressed were noteworthy, suggesting preservation of higher cognitive functions. Complementary tasks indicated stronger performance in visuospatial working memory compared to auditory memory, along with praxic difficulties (manual and oral), global motor incoordination, and limitations in the mental manipulation of numerical quantities.

Discussion

Kabuki syndrome presents heterogeneous clinical features, with intellectual disability being a frequent finding, albeit with varying degrees of severity. In this case, although the patient faces significant challenges in reading and writing, his ability to comprehend text and formulate ideas suggests relative cognitive preservation. This is a fundamental consideration in educational planning, indicating the potential for academic progress when appropriate strategies are applied.

Language development in Kabuki syndrome is particularly complex, involving sensory factors (such as the hearing loss present in this case), as well as motor and cognitive aspects. The literature

highlights the absence of a uniform linguistic profile, with substantial variation among individuals in phonological, morphosyntactic, and pragmatic abilities[9,10]. The decoding and writing difficulties observed in this patient may be related to auditory components as well as specific linguistic processing deficits described in the syndrome.

The findings underscore the importance of a comprehensive clinical assessment that considers both limitations and strengths. Furthermore, the phoniatric assessment emphasizes the value of a multidisciplinary approach, supporting therapists and educators by clarifying functional aspects related to medical findings and suggesting school adaptations. In this case, relatively preserved visuospatial abilities can be used as compensatory strategies for phonological processing difficulties[11]. Similarly, graphomotor limitations justify the implementation of school accommodations, such as the use of technological tools or adjustments to written production requirements.

Phoniatric assessment proved essential in characterizing the patient's specific profile, identifying both deficits and preserved skills. This detailed analysis enabled the development of an individualized intervention plan, encompassing educational adaptations and the reintroduction of targeted therapies. The approach should address not only academic issues but also the development of socio-emotional and functional skills, aiming to promote greater autonomy in adult life[10,11].

This case reinforces the importance of phoniatric assessment in patients with complex genetic syndromes, demonstrating how a thorough understanding of the individual functional profile can guide more effective interventions and improve overall developmental outcomes.

Final comments

The prognosis for children with Kabuki syndrome has become increasingly favorable, especially when they receive early and individualized interventions. Clinical experience shows that with appropriate support, many patients reach significant developmental milestones and achieve satisfactory levels of autonomy in adult life.

In this context, phoniatric evaluation plays a crucial role, offering a comprehensive analysis of the organic and functional aspects involved in language and cognitive development. Its multidimensional nature allows for the identification of both limitations and potential, guiding the development of personalized therapeutic strategies and effective educational accommodations.

Systematic monitoring of academic performance is essential not only for intellectual development but also as a key factor in fostering functional independence and psychosocial well-being. At the same time, initiatives promoting social inclusion, accessibility, and awareness of the syndrome are vital to ensure the full development and active participation of these individuals in society[13].

This case supports the value of medical phoniatric assessment within interdisciplinary care and management of Kabuki syndrome, helping clarify medical aspects and suggesting targeted interventions that transform challenges into opportunities for growth and development.

References

1. Souza JC, Ribeiro TCC, Ribeiro RC. A síndrome da máscara do Cabúqui / Kabuki make-up syndrome. J Pediatr (Rio J). 1996; 72(5): 341-4.

2. Ho HH, Beirais LC. Kabuki make-up (Niikawa-Kuroki) syndrome: cognitive abilities and autistic features. Dev Med Child Neurol. 1997 Jul; 39(7): 487-90.

3. Sanz JH, Lipkin P, Rosenbaum K, Mahone EM. Developmental profile and trajectory of neuropsychological skills in a child with Kabuki syndrome: implications for assessment of syndromes associated with intellectual disability. Health Sci Res Commons. 2010.

4. Barozzi S, Di Berardino F, Atzeri F, Filipponi E, Cerutti M, Selicorni A, Cesarani A. Audiological and vestibular findings in the Kabuki syndrome. Int J Pediatr Otorhinolaryngol. 2009.

5. Schoen-Ferreira TH, Ramos JMP, Ávila MEB, Dabbur RR, Lima TA, Marteleto MRF. Síndrome de Kabuki: estudo de caso a respeito das características comportamentais, cognitivas, sociais e fonoaudiológicas. Rev Soc Bras Fonoaudiol. 2010.

6. Matsumoto N, Niikawa N. Kabuki make-up syndrome: a review. Am J Med Genet C Semin Med Genet. 2002 Dec 17.

7. Wessels MW, Brooks AS, Hoogeboom J, Niermeijer MF, Willems PJ. Kabuki syndrome: a review study of three hundred patients. Clin Dysmorphol. 2002 Apr; 11(2): 95-102.

8. Van Lierde KM, Van Borsel J, Van Cauwenberge P. Speech patterns in Kabuki make-up syndrome: a case report. J Commun Disord. 2000 Nov-Dec; 33(6): 447-61; quiz 461-2.

9. Defloor T, Van Borsel J, Schrander-Stumpel CT, Curfs LM. Expressive language in children with Kabuki syndrome. Am J Med Genet A. 2005; 132A (3): 256-9.

10. Brito MC, Misquiatti ARN. Intervenção fonoaudiológica na síndrome de Kabuki: relato de caso. Rev Soc Bras Fonoaudiol. 2010.

11. Dębska A, Wójcik M, Chyl K, Dzięgiel-Fivet G, Jednoróg K. Além da área de forma visual da palavra - uma caracterização cognitiva do córtex occipitotemporal ventral esquerdo. Neurosci do zumbido frontal. 28 de julho de 2023; 17: 1199366. DOI: 10.3389/fnhum.2023.1199366. PMID: 37576470; PMCID: PMC10416454.

12. Silke M Göbel, Rebecca Terry, Elise Klein, Mark Hymers, Liane Kaufmann. Impaired Arithmetic Fact Retrieval in an Adult with Developmental Dyscalculia: Evidence from Behavioral and Functional Brain Imaging Data. Brain Sciences, 2022, 12 (6), pp.735. 10.3390/brainsci12060735. halshs-03865926

13. Rotta NT, Ohlweiler L, Riesgo RS. Transtornos da Aprendizagem: Abordagem Neurobiológica e Multidisciplinar. Porto Alegre: Artmed; 2006.