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# A Celiac Mimicry: Kabuki Syndrome

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**Abstract** Kabuki Syndrome is a rare genetic condition with a large panel of manifestations, including digestive symptoms that may simulate a celiac disease. We present a case of a very rare presentation of Kabuki Syndrome type 2.

***Keywords:*** *celiac disease, kabuki syndrome, HLA*

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## 1. Introduction

As an autoimmune disorder and occurring on a particular genetic ground (i.e. HLA DQ2/DQ8), celiac disease (CD) can nevertheless mimic a very specific syndromic association. We present a case of a very rare presentation of Kabuki Syndrome type 2.

## 2. Case Presentation

We present the case of a 12-year-old boy, diagnosed since the age of 05 as CD with digestive signs and supported by digestive endoscopy with histology in favor of Marsh stage III villous atrophy. No initial celiac serology was performed when the diagnosis was made.

He started gluten-free diet since at that moment, and consulted for a follow-up of CD in the outpatient clinic and presented on clinical examination, in addition to typical facial dysmorphia, mental retardation and pronounced visual impairment.

The ophthalmology review reveals severe bilateral optic atrophy. Optical Coherence Tomography subsequently confirms these typical lesions of optical atrophy. Neurological investigation (neuropediatric consultation, EEG and brain MRI) did not find any other specific stigmata. Likewise, the cardiac evaluation (ECG, cardiac ultrasound) is without particularities. A complete class II HLA typing is ordered for this peculiar case and the result is unequivocal: absence of the risk genes HLA DQ2 / DQ8.

Finally, the diagnosis of Kabuki Syndrome is retained according to the 2019 consensus criteria [1]. This made it possible to release the child's diet with an appreciable weight gain (+ 2 kg) over 12 months.

## 3. Discussion

The association of these symptoms is almost pathognomonic of Kabuki Syndrome: a rare, genetic association characterized by growth delay, intellectual deficit, and typical facial dysmorphia (hence the name of Kabuki = make-up of Japanese theatre actors) and various skeletal, cardiac, renal and ocular malformations [1-7].

Dysmorphic features encompass long palpebral fissures, eversion of the lateral third at the lower eyelid; large and arched eyebrows; short columella, depressed nasal tip; malformed ears [2].

Neurological involvement is almost constant, particularly the intellectual delay, sometimes with specific scars on brain MRI [4].

Two distinct genetic types of Kabuki Syndrome are described [1,3,8]:

- Kabuki Syndrome Type 1, OMIM 147920: autosomal dominant, gene 12q13.12
- Kabuki Syndrome Type 2, OMIM 300867: X-linked, Xp11.3 gene

The specific genetic test could not be performed in our patient; nevertheless, the most likely type for this patient is type 2 due to the absence of similar manifestations in his parents.

Several presentations similar to our patient, mimicking or associating celiac disease, have already been described worldwide and reported sparingly in the literature [2,7]

There is a wide heterogeneous spectrum of clinical manifestations in KS, some authors recently suggested introducing additional diagnostic criteria for correct and early recognition of this syndrome [7].

The management of this syndrome is not specific and depends on the organ/system involvement [2,9].

## 4. Conclusion

Kabuki Syndrome is a rare genetic disorder that may induce malabsorption; diarrhea and growth failure and so simulate a celiac disease.

Cautious examination and a holistic patient' approach are mandatory.

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## References

[1] Adam MP, Banka S, Bjornsson HT, Bodamer O, Chudley AE, Harris J, Kawame H, Lanpher BC, Lindsley AW, Merla G, Miyake N, Okamoto N, Stumpel CT, Niikawa N; Kabuki Syndrome Medical Advisory Board. Kabuki syndrome: international consensus diagnostic criteria. J Med Genet. 2019 Feb; 56(2): 89-95.

[2] Adam MP, Hudgins L, Hannibal M. Kabuki Syndrome. 2011 Sep 1 [Updated 2021 Jul 15]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2021. Available from: https://www.ncbi.nlm.nih.gov/books/NBK62111/.

[3] Adam M, Hudgins L. Kabuki syndrome: a review: Kabuki syndrome. Clinical Genetics. 20 oct 2004; 67(3): 209-19.

[4] Boisgontier J, Tacchella JM, Lemaître H, Lehman N, Saitovitch A, Gatinois V, et al. Anatomical and functional abnormalities on MRI in kabuki syndrome. NeuroImage: Clinical. 2019; 21: 101610.

[5] Geneviève D, Amiel J, Viot G, Le Merrer M, Sanlaville D, Urtizberea A, Gérard M, Munnich A, Cormier-Daire V, Lyonnet S. Atypical findings in Kabuki syndrome: report of 8 patients in a series of 20 and review of the literature. Am J Med Genet A. 2004 Aug 15; 129A(1): 64-8.

[6] Digilio MC, Marino B, Toscano A, Giannotti A, Dallapiccola B. Congenital heart defects in Kabuki syndrome. American Journal of Medical Genetics. 2001; 100(4): 269-74.

[7] Di Candia F, Fontana P, Paglia P, Falco M, Rosano C, Piscopo C, Cappuccio G, Siano MA, De Brasi D, Mandato C, De Maggio I, Squeo GM, Monica MD, Scarano G, Lonardo F, Strisciuglio P, Merla G, Melis D. Clinical heterogeneity of Kabuki syndrome in a cohort of Italian patients and review of the literature. Eur J Pediatr. 2021 Jul 7.

[8] Ng SB, Bigham AW, Buckingham KJ, Hannibal MC, McMillin MJ, Gildersleeve HI, et al. Exome sequencing identifies MLL2 mutations as a cause of Kabuki syndrome. Nat Genet. sept 2010;42(9):790-3.

[9] Philip N, Devriendt K, Clayton-Smith J. Management of Kabuki Syndrome: a clinical guideline [Internet]. Version.