Psychiatry Research Case Reports 2 (2023) 100150

Contents lists available at ScienceDirect

Psychiatry Research Case Reports

journal homepage: www.elsevier.com/locate/psycr

Gómez-López-Hernández syndrome: An adult case diagnosed after psychiatric symptoms

Bilge Targitay Ozturk [a,*], Tutkucan Guvendik [a], Ummuhan Nilay Agac [b,c], Burak Celik [a], Eldem Guvercin [a], Cemre Ozenbas [d,e], Nuri Karabay [d], Can Cimilli [a], Tolga Binbay [a]

[a] Department of Psychiatry, Dokuz Eylul University Medical School, Mithatpaşa Street, 1606, Balcova, Izmir, Izmir, Turkey
[b] Department of Child and Adolescent Psychiatry, Dokuz Eylul University Medical School, Izmir, Turkey
[c] Department of Child and Adolescent Psychiatry, Uludag University Medical School, Bursa, Turkey
[d] Department of Radiology, Dokuz Eylul University Medical School, Izmir, Turkey
[e] Department of Radiology, Kars Harakani State Hospital, Kars, Turkey

ARTICLE INFO

Keywords:
Gómez-López-Hernández syndrome
Rhombencephalosynapsis
Schizoaffective disorder

ABSTRACT

Gómez-López-Hernández Syndrome or cerebellotrigeminal dysplasia is a rare condition characterized by the triad of rhombencephalosynapsis, alopecia, and trigeminal anesthesia. The genetic background of this syndrome which has many phenotypic features such as brachycephaly (a lower-than-normal ratio of the skull's length to its width), midface hypoplasia, low-set and posteriorly angulated ear structure, hypertelorism (increased distance between orbits), thin lips, and motor mental development retardation, is not known clearly yet. In reports most of the cases are in childhood, it has been observed that psychiatric disorders can often accompany this syndrome, moreover, may have an early onset. Magnetic resonance imaging was performed on a 58-year-old male patient with a diagnosis of schizoaffective disorder, treatment-resistant psychiatric symptoms and epilepsy. As a result, rhombencephalosynapsis was observed on magnetic resonance imaging. Rhombencephalosynapsis can be isolated or accompany many congenital anomalies or syndromes. The most common congenital syndrome associated with rhombencephalosynapsis is Gómez-López-Hernández Syndrome. Detailed physical examination which included mild brachycephaly, regional alopecia, absence of bilateral corneal reflex, sensory loss in the trigeminal nerve area, hearing loss, downslanted palpebral fissures, head shaking, ataxia and a significant history of psychiatric disorders suggested a diagnosis of Gómez-López-Hernández Syndrome. In this article, role of rhombencephalosynapsis and related syndrome is discussed in terms of severe psychopathology.

Introduction

Gómez-López-Hernández Syndrome was first described by Gómez in a female child with trigeminal anesthesia, regional alopecia, brachycephaly, hypertelorism, strabismus, and cerebellar ataxia in 1979 (Gomez, 1979) and confirmed by López-Hernández (1982). This rare neurocutaneous syndrome consists of trigeminal anesthesia, alopecia and rhombencephalosynapsis triad. Phenotypic findings such as craniosynostoses, facial anomalies, deafness, motor mental retardation, ataxia can also be seen in this syndrome, whose etiology is not clearly realized yet (Muñoz et al., 1997). It has been suggested that the findings other than alopecia and rhombencephalosynapsis show less consistency in the assessments. In a case series of 13 patients, alopecia was observed in 100%, rhombencephalosynapsis in 83% and trigeminal anesthesia in 45% of patients (Perrone et al., 2021). Therefore, alopecia and rhombencephalosynapsis have been proposed as consistent and sufficient criteria (Sukhudyan et al., 2010). However, recently, it has been stated that the absence/hypolasia of the trigeminal nerve and the detailed evaluation of its related symptoms are an important predictor of the diagnosis of the syndrome (Perrone et al., 2021). Since it was observed that psychiatric conditions such as bipolar disorder and attention deficiency could accompany the syndrome, apart from mental developmental delay, the phenotypic features have been expanded to include psychiatric conditions by Brocks et al. (2000).

Case

A 58-year-old male patient with a diagnosis of schizoaffective

* Corresponding author.
E-mail address: bilge.targitay@deu.edu.tr (B. Targitay Ozturk).

https://doi.org/10.1016/j.psycr.2023.100150
Received 27 September 2022; Received in revised form 23 July 2023; Accepted 24 July 2023
Available online 25 July 2023
2773-0212/© 2023 The Authors. Published by Elsevier B.V. This is an open access article under the CC BY-NC-ND license (http://creativecommons.org/licenses/by-nc-nd/4.0/).

disorder and treatment-resistant psychiatric symptoms was hospitalized in the psychiatry service for planning his treatment and determining the conditions that may cause treatment resistance. The first psychiatric complaints of the patient started when he was 15 years old as mood episodes accompanied by psychotic symptoms, and the patient was diagnosed with bipolar disorder type 1 in this period. The patient, whose parents did not have consanguineous marriage, was born as the second of 5 siblings and all of his siblings were diagnosed with bipolar disorder type 1 at an early age. Since this family history was obtained from the patient, the patient's siblings were invited to the hospital for a detailed psychiatric interview by the patient's doctor. However, these evaluations could not be carried out due to the fact that his siblings live outside the city.

The patient, who was hospitalized in psychiatry services many times, has been showing persistence of psychotic symptoms for the last 8 years, even outside of the mood episodes. Therefore, the diagnosis of the patient was updated as schizoaffective disorder 7 years ago. Despite using many antipsychotic drugs including clozapine and mood stabilizer drugs at effective doses and durations, psychotic symptoms and mood episodes persist. In addition, the patient was diagnosed with moderate epilepsy 15 years ago and his seizures continue despite dual antiepileptic therapy (valproic acid and lamotrigine). Side effects such as tremor, dystonia, increase in epileptic seizures, neutropenia, etc. due to the drug treatments that he used, were observed, so the functionality of the patient further decreased. Manic episode symptoms such as decreased sleep duration, increased libido, increased desire to spend money, and irritability were observed along with jealous-paranoid delusions and auditory-tactile hallucinations in the mental state examination performed during hospitalization. The Young Mania Rating Scale score was 29 and the Positive and Negative Symptoms Scale total score was 113 at the time of hospitalization. Treatment-resistant symptoms of the patient were basically related to the content of psychotic thought and behavioral changes related to it. On the YMRS score, he got the highest scores for the sections that included delusions, hallucinations and irritability. Therefore, the patient's psychotic symptoms fit the definition of marked illness in terms of PANSS score and moderate mania in terms of YMRS score.

Magnetic resonance imaging (MRI) and electroencephalography (EEG) were performed to the patient. MRI revealed hypoplasia in the cerebellar vermis, partial continuity in the cerebellar hemispheres (rhombencephalosynapsis), mild atrophy in the corpus callosum and hippocampus, mild dilatation in the lateral ventricles (Fig. 1a), and bilateral trigeminal nerve hypoplasia (Fig. 1b). EEG showed no epileptic discharges.

In the neurocognitive evaluation; the patient's personal information, location orientation and abstract conceptualization were preserved. Moderate/severe impairment has been detected in all other neurocognitive processes in attention, executive functions and memory fields.

After evaluating the patient's family history and current findings, a detailed physical examination was performed. Mild brachycephaly (Fig. 2a), regional alopecia at the bilateral parietooccipitotemporal junction (Fig. 2b, c), absence of bilateral corneal reflex, sensory loss in the trigeminal nerve area on the entire face, hearing loss at a level that required the use of a device decided as a result of the hearing test, downslanted palpebral fissures (Fig. 2d), head shaking, and ataxia were detected in the patient.

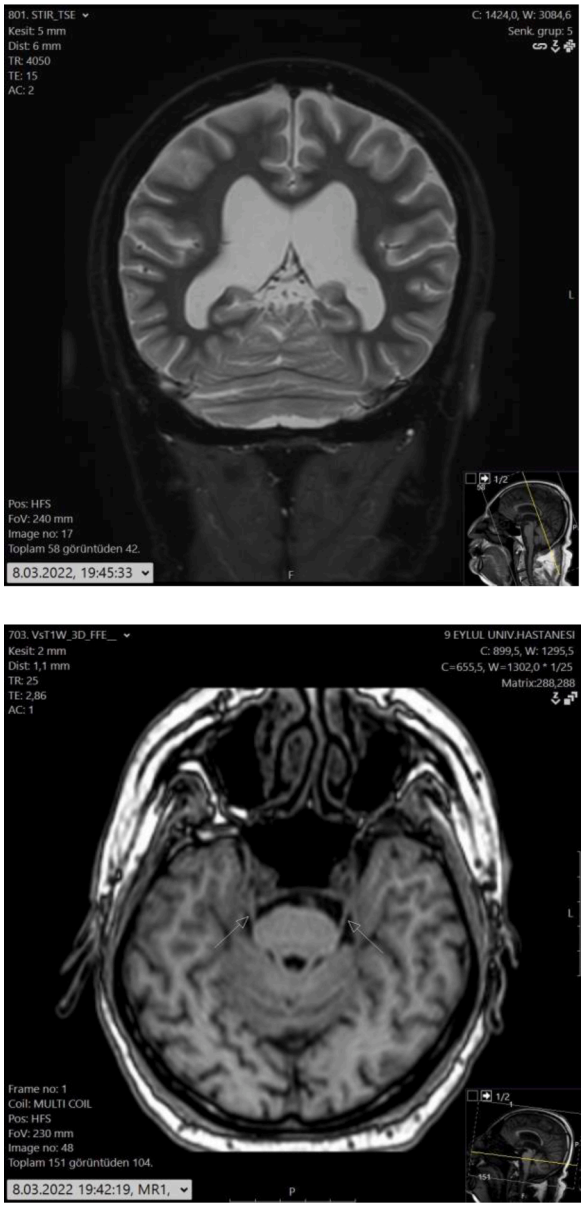


Fig. 1. a: Hypoplasia in the cerebellar vermis, partial continuity in the cerebellar hemispheres (rhombencephalosynapsis), mild dilatation in the lateral ventricles in coronal short tau inversion recovery (STIR) sequence, Fig. 1b: Bilateral trigeminal nerve hypoplasia in axial 3D T1 sequence.

Discussion

The thought that the patient had Gómez-López-Hernández Syndrome with his current findings was realized after the appearance of rhombencephalosynapsis on MRI. Rhombencephalosynapsis is a rare congenital anomaly that is characterized by partial or total agenesis of the cerebellar vermis (mainly dorsal portion) with midline fusion of cerebellar hemispheres, dentate nuclei, and superior cerebellar peduncles. Rhombencephalosynapsis was first described in 1916 in the postmortem examination of a 28-year-old man who died by suicide (Fouda et al., 2022). Rhombencephalosynapsis can be isolated or accompany many congenital anomalies or syndromes. The most common congenital syndrome associated with rhombencephalosynapsis is Gómez-López-Hernández Syndrome (Lindsay et al., 2020). At least 90 cases of rhombencephalosynapsis were described in the literature with approximately 25% of those associated with Gómez-López-Hernández Syndrome (Ishak et al., 2012). In the etiology of Gómez-López-Hernández Syndrome, it is generally suggested that problems in the development of the ectoderm from which the cerebellum, trigeminal nerve and epidermal cells originate, are the main factors. The common embriyologic origin in the neural ectoderm provides a clue to the obscure pathogenesis of Gómez-López-Hernández Syndrome (Fernández-Jaén et al., 2009).

Although the syndrome was initially defined by the triad of rhombencephalosynapsis, partial alopecia and trigeminal anesthesia, it was observed that the trigeminal anesthesia finding was not consistent in all



Fig. 2. a: Mild brachycephaly, Fig. 2b, c: Regional alopecia at the bilateral parietooccipitotemporal junction, Fig. 2d: Downslanted palpebral fissures.

cases as a result of the case reports. For this reason, it was stated by Rush et al. (2013) that rhombencephalosynapsis and alopecia should be core symptoms. It has been recommended that rhombencephalosynapsis + alopecia + 1 major craniofacial criterion [brachycephaly, turribrachycephaly (shortening of the anteroposterior dimention and an elongation of the vertical dimention of the skull), midface retrusion] or alopecia + two or more neurological [motor delay, ataxia, hypotonia, intellectual disability, head shaking or other stereotypic movement] / minor craniofacial criteria [strabismus, widely spaced eyes, plagiocephaly (asymmetric shape of head due to unilateral flattering), lambdoid craniosynostosis (fusion of one of the lambdoid sutures before birth and flatting in the back of the head)] can be used instead of triad as current diagnostic criteria. Considering the current literature, it is underlined that trigeminal anesthesia is a rare finding seen in genetic syndromes. Therefore, this particular symptom can be a spesific sign in diagnosis of Gómez-López-Hernández Syndrome. It is claimed that its presence together with rhombencephalosynapsis, local alopecia, brachycephaly, turribrachycephaly or midface retrusion may be sufficient for the diagnosis of Gómez-López-Hernández Syndrome. Therefore, it is suggested that the neurological examination of the trigeminal nerve should be done carefully and its anatomy should be examined in detail with imaging methods (Perrone et al., 2021).

The alopecia seen in this syndrome is bilateral and nonscarring, usually seen in the parietal or occipital regions of scalp. Although it is rarer, it is also observed in different distributions such as temporal or frontal areas, and there are case reports in which it is distinguished unilaterally. The absence of congenital alopecia does not exclude the possibility of diagnosis, hair loss may also occur in later ages (Brocks et al., 2000; Fernández-Jaén et al., 2009; Purvis et al., 2007).

Approximately 57 cases have been described worldwide to date. Patients are mostly diagnosed in childhood and the latest age of diagnosis reported in the literature is 41 (Perrone et al., 2021; Perrone et al., 2020). Therefore, the case described in this article is of particular importance since he was diagnosed at the age of 58 years.

Even though more than 30 years have passed since the definition of the syndrome, the etiology of the disease is still unknown. It is not certain whether Gómez-López-Hernández Syndrome and isolated rhombencephalosynapsis are different entities, or they represent a disease spectrum (Sukhudyan et al., 2010). It has been claimed that genetic, teratogenic and mixed factors may be involved in the pathogenesis of the disease. While partial deletions and duplications were detected in case reports or case series-based genetic studies, no consistent candidate gene, single nucleotide polymorphism or copy number variations was stated. The low rate of familial history in terms of the syndrome and the rare presence of consanguineous marriages in the families of the patients indicate the possibility of de novo mutation more on genetic background (Aldinger et al., 2018; Gomy et al., 2008; Schell-Apacik et al., 2008; Bowdin et al., 2007). For this reason, it is suggested that whole genome sequencing or DNA and/or RNA sequencing of the alopesia tissue for the investigation of non-coding causative variants and somatic mosaic causative variants can provide more detailed information on the genetic origin of the disease in the future (Perrone et al., 2021; Choudhary et al., 2017).

Both the trigeminal nerve and the cerebellum develop from rhombomeres, which are subunits of the rhombencephalon. These structures, which have a common ectodermal origin, are seen in the 3–4th week of gastrulation (O'Rahilly and Müller, 2007). During this period, factors such as trauma, alcohol or illegal substance use, drugs, infections are considered to be important in the development of the syndrome. In the retrospective inquiries made with the mothers of some patients with rhombencephalosynapsis, no serious exposure to fetal development during pregnancy was detected for Gómez-López-Hernández Syndrome, except for diabetes and the use of assisted reproductive techniques. However, misoprostol exposure in the intrauterine period was reported in a case with Gómez-López-Hernández Syndrome (Perrone et al., 2020), and a history of multiple illicit drugs use was reported in the mother of another case (Choudhri et al., 2015). The fact that the symptoms of the syndrome are observed only in ectoderm-derived structures indicates the possibility of somatic mosaicism, which is characterized by only some cells carry the genetic disease and exhibit its symptoms. In addition, considering the probability of somatic mosaicism, prenatal diagnosis methods may be important in the early diagnosis of the disease.

There is limited data in the literature on psychiatric symptoms in both isolated cases of rhombencephalosynapsis and in patients with Gómez-López-Hernández Syndrome. Psychiatric disorders such as attention deficit and hyperactivity disorder, depression, bipolar disorder, cognitive impairment, intellectual disability etc. can be observed in patients with cerebellar anomalies (Schmahmann et al., 2007). In a report of a typical case of Gómez-López-Hernández Syndrome published in 2000, it was underlined that the case was diagnosed with bipolar disorder at the age of 13, and the importance of being careful in terms of early-onset psychiatric diseases in the follow-up of other cases was emphasized (Brocks et al., 2000). Attention problems were reported as the most common behavioural disturbance in a series of 5 cases in which cognitive evaluation was performed in children with rhombencephalosynapsis (Poretti et al., 2009). In a case series of 9 children, it was stated that rhombencephalosynapsis accompanied by supratentorial anomaly and hydrocephalus may have an important role on long-term cognitive functions. Cognitive evaluations made in patients with rhombencephalosynapsis diagnosed with MRI in different studies between 1991 and 2001 were presented as a table in this case series and it was stated that cognitive functions were frequently impaired (Toelle et al., 2002). But a typical cognitive profile for rhombencephalosynapsis could not be detected (Poretti et al., 2009). It is thought that this situation occurred as a result of the affected limbic cerebellar structures, which has recently come to the fore with cerebellar cognitive affective syndrome. However, it is not known yet how the long-term cognitive effects in rhombencephalosynapsis will be (Ahmadian et al., 2019; Schmahmann, 2019). In current studies, the cerebellum is frequently discussed in terms of its place in the etiology of psychiatric diseases. Neuropsychiatric symptoms in people with cerebellar disorders are examined in 5 different categories: attention control, emotion control, autism spectrum, psychosis spectrum and social skills. Apart from these 5 areas, there are functions of cerebellar structures in executive functions, linguistic processing and visiospatial awareness. It is suggested that the midline structures including the vermis may be involved in the formation of these symptoms (Schmahmann et al., 2007). The influence of the cortico-cerebellar-thalamic-cortical circuit may be associated with psychosis. Cerebellar hypoactivity and vermis atrophy are among the findings detected so far in schizophrenia. In addition, the most determining area in the cerebellum in terms of the severity of psychotic symptoms is the posterior cerebellum. Data on the cerebellum contain inconsistent results for depression. The most common finding in bipolar disorder is decreased blood flow in the vermis. An increase in cerebellar metabolism was reported in treatment-resistant bipolar disorder (Konarski et al., 2005). Our case is similar to the cases in the literature due to early onset bipolar disorder diagnosis, treatment-resistant course of illness and impaired cognitive functions. Considering all these findings, rhombencephalosynapsis, which occurred as a part of Gómez-López-Hernández Syndrome in our patient, could be a decisive factor in the patient's early onset and treatment-resistant symptoms, more susceptibility for the side effects from medications and deterioration of cognitive functions. However, there is no clear information in the literature related to these conditions. In severe psychiatric diseases resistant to treatment, detailed brain imaging (esp. MRI), physical and neurological examination should be done. Also psychiatric evaluation of all family members, genetic and neurological counseling of the patient could be recommended. Indeed, if the symptoms of this syndrome are mild, the diagnosis can be made only in adulthood, whereas the severest cases stand out in childhood.

Informed consent

Written informed consent was obtained from the patient to report his case in this case study.

Declaration of Competing Interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

References

Ahmadian, N., van Baarsen, K., van Zandvoort, M., Robe, P.A., 2019. The cerebellar cognitive affective syndrome-a meta-analysis. Cerebellum 18, 941–950. https://doi.org/10.1007/s12311-019-01060-2.

Aldinger, K.A., Dempsey, J.C., Tully, H.M., Grout, M.E., Mehaffey, M.G., Dobyns, W.B., 2018. Rhombencephalosynapsis: fused cerebellum, confused geneticists. Am. J. Med. Genet. C Semin. Med. Genet. 178, 432–439. https://doi.org/10.1002/ajmg.c.31666.

Bowdin, S., Phelan, E., Watson, R., McCreery, K.M., Reardon, W., 2007. Rhombencephalosynapsis presenting antenatally with ventriculomegaly/hydrocephalus in a likely case of Gomez-López-Hernández syndrome. Clin. Dysmorphol. 16, 21–25. https://doi.org/10.1097/01.mcd.0000228426.97284.ff.

Brocks, D., Irons, M., Sadeghi-Najad, A., McCauley, R., Wheeler, P., 2000. Gomez-Lopez-Hernandez syndrome: expansion of the phenotype. Am. J. Med. Genet. 94, 405–408. https://doi.org/10.1002/1096-8628(20001023)94:5.405::aid-ajmg12.3.0.co;2-8.

Choudhary, A., Minocha, P., Sitaraman, S., 2017. Gomez-Lopez-Hernández syndrome: first reported case from the Indian subcontinent. Intractable Rare Dis. Res. 6, 58–60. https://doi.org/10.5582/irdr.2016.01074.

Choudhri, A.F., Patel, R.M., Wilroy, R.S., Pivnick, E.K., Whitehead, M.T., 2015. Trigeminal nerve agenesis with absence of foramina rotunda in Gómez-López-Hernández syndrome. Am. J. Med. Genet. A 167, 238–242. https://doi.org/10.1002/ajmg.a.36830.

Fernández-Jaén, A., Fernández-Mayoralas, D.M., Calleja-Pérez, B., Muñoz-Jareño, N., Moreno, N., 2009. Gomez-Lopez-Hernandez syndrome: two new cases and review of the literature. Pediatr. Neurol. 40, 58–62. https://doi.org/10.1016/j.pediatrneurol.2008.10.001.

Fouda, M.A., Kim, T.Y., Cohen, A.R., 2022. Rhomboencephalosynapsis: review of the literature. World Neurosurg. 159, 48–53. https://doi.org/10.1016/j.wneu.2021.12.062.

Gomez, M.R., 1979. Cerebellotrigeminal and focal dermal dysplasia: a newly recognized neurocutaneous syndrome. Brain Dev. 1, 253–256. https://doi.org/10.1016/S0387-7604(79)80039-X.

Gomy, I., Heck, B., Santos, A.C., Figueiredo, M.S.L., Martinelli, C.E., Nogueira, M.P.C., 2008. Two new Brazilian patients with Gómez-López-Hernández syndrome: reviewing the expanded phenotype with molecular insights. Am. J. Med. Genet. A 146A, 649–657. https://doi.org/10.1002/ajmg.a.32173.

Ishak, G.E., Dempsey, J.C., Shaw, D.W.W., Tully, H., Adam, M.P., Sanchez-Lara, P.A., 2012. Rhombencephalosynapsis: a hindbrain malformation associated with incomplete separation of midbrain and forebrain, hydrocephalus and a broad spectrum of severity. Brain 135 (5), 1370–1386. https://doi.org/10.1093/brain/aws065. Pt.

Konarski, J.Z., McIntyre, R.S., Grupp, L.A., Kennedy, S.H., 2005. Is the cerebellum relevant in the circuitry of neuropsychiatric disorders? J. Psychiatry Neurosci. 30, 178–186. http://www.ncbi.nlm.nih.gov/pubmed/15944742.

López-Hernández, A., 1982. Craniosynostosis, ataxia, trigeminal anaesthesia and parietal alopecia with pons-vermis fusion anomaly (atresia of the fourth ventricle). report of two cases. Neuropediatrics 13, 99–102. https://doi.org/10.1055/s-2008-1059606.

Lindsay, F., Anderson, I., Wentzensen, I.M., Suhrbier, D., Stevens, C.A., 2020. Genetic evaluation including exome sequencing of two patients with Gomez-Lopez-Hernandez syndrome: case reports and review of the literature. Am. J. Med. Genet. A 182, 623–627. https://doi.org/10.1002/ajmg.a.61496.

Muñoz, R.M.V, Santos, A.C., Graziadio, C., Pina-Neto, J.M, 1997. Cerebello-trigeminal-dermal dysplasia (Gómez-López-Hernández syndrome): description of three new cases and review. Am. J. Med. Genet. 72, 34–39. https://doi.org/10.1002/(SICI)1096-8628(19971003)72:1.34::AID-AJMG7.3.0.CO;2-V.

O'Rahilly, R., Müller, F., 2007. The development of the neural crest in the human. J. Anat. 211, 335–351. https://doi.org/10.1111/j.1469-7580.2007.00773.x.

Perrone, E., D'Almeida, V., de Macena Sobreira, N.L., de Mello, C.B., de Oliveira, A.C., Burlin, S., 2020. Gomez-López-Hernández syndrome: a case report with clinical and molecular evaluation and literature review. Am. J. Med. Genet. A 182, 1761–1766. https://doi.org/10.1002/ajmg.a.61594.

Perrone, E., Perez, A.B.A., D'Almeida, V., Mello, C.B., Jacobina, M.A.A., Loureiro, R.M., 2021. Clinical and molecular evaluation of 13 Brazilian patients with Gomez-López-Hernández syndrome. Am. J. Med. Genet. A 185, 1047–1058. https://doi.org/10.1002/ajmg.a.62059.

Poretti, A., Alber, F.D., Bürki, S., Toelle, S.P., Boltshauser, E., 2009. Cognitive outcome in children with rhombencephalosynapsis. Eur. J. Paediatr. Neurol. 13, 28–33. https://doi.org/10.1016/j.ejpn.2008.02.005.

Purvis, D.J., Ramirez, A., Roberts, N., Harper, J.I., 2007. Gomez-Lopez-Hernandez syndrome: another consideration in focal congenital alopecia. Br. J. Dermatol. 157, 196–198. https://doi.org/10.1111/j.1365-2133.2007.07952.x.

Rush, E.T., Adam, M.P., Clark, R.D., Curry, C., Hartmann, J.E., Dobyns, W.B., 2013. Four new patients with Gomez-Lopez-Hernandez syndrome and proposed diagnostic criteria. Am. J. Med. Genet. A 161A, 320–326. https://doi.org/10.1002/ajmg.a.35817.

Schell-Apacik, C.C., Cohen, M., Vojta, S., Ertl-Wagner, B., Klopocki, E., Heinrich, U., 2008. Gomez-Lopez-Hernandez syndrome (cerebello-trigeminal-dermal dysplasia): description of an additional case and review of the literature. Eur. J. Pediatr. 167, 123–126. https://doi.org/10.1007/s00431-007-0478-z.

Schmahmann, J.D., Weilburg, J.B., Sherman, J.C., 2007. The neuropsychiatry of the cerebellum - insights from the clinic. Cerebellum 6, 254–267. https://doi.org/10.1080/14734220701490995.

Schmahmann, J.D., 2019. The cerebellum and cognition. Neurosci. Lett. 688, 62–75. https://doi.org/10.1016/j.neulet.2018.07.005.

Sukhudyan, B., Jaladyan, V., Melikyan, G., Schlump, J.U., Boltshauser, E., Poretti, A., 2010. Gómez–López-Hernández syndrome: reappraisal of the diagnostic criteria. Eur. J. Pediatr. 169, 1523–1528. https://doi.org/10.1007/s00431-010-1259-7.

Toelle, S.P., Yalcinkaya, C., Kocer, N., Deonna, T., Overweg-Plandsoen, W.C.G., Bast, T., 2002. Rhombencephalosynapsis: clinical findings and neuroimaging in 9 children. Neuropediatrics 33, 209–214. https://doi.org/10.1055/s-2002-34498.