J Med Allied Sci 2026; 16 (2): 109-116
www.jmas.in
Print ISSN: 2231 1696 Online ISSN: 2231 170X
Journal of Medical & Allied Sciences

Case series

Brown-Vialetto-Von Laere syndrome: A case series with review of literature

Mohammed Zoheb, Singireddy Chandra Reddy

Department of Neurology, Deccan College of Medical Sciences, DMRL 'X' Road, Kanchanbagh, Hyderabad-500058, Telangana, India.

Article history:

Received 14 March 2026
Revised 31 March 2026
Accepted 31 March 2026
Online 31 July 2026
Print 31 July 2026

Corresponding author

Mohammed Zoheb
Professor
Department of Neurology,
Deccan College of Medical Sciences,
DMRL 'X' Road, Kanchanbagh,
Hyderabad-500058, Telangana, India.
Phone: +91-9989166906
Email: zoheb.mohammed@gmail.com

Abstract

Brown-Vialetto-Van Laere syndrome (BVVL) is an exceptionally rare juvenile-onset motor neuron disease characterized by progressive ponto-bulbar palsy, sensorineural hearing loss, and multiple cranial nerve involvement, caused by mutations in riboflavin transporter genes SLC52A1, SLC52A2, and SLC52A3. Fewer than 90 cases have been documented in the literature to date. We report a familial case series of three siblings of Indian descent, born to non-consanguineous parents, presenting with bilateral lower motor neuron palsies involving cranial nerves VII, VIII, IX, X, and XII, progressive sensorineural hearing loss, dysarthria, dysphonia, and bilateral vocal cord paralysis. Notably, all three siblings reported a preceding febrile illness prior to symptom onset, a pattern previously described in the literature, though its pathophysiological significance remains uncertain. Markedly reduced plasma riboflavin levels were documented in all three patients. Clinical exome sequencing identified a heterozygous missense mutation in exon 2 of the SLC52A3 gene (chr20:g.746030A>T, p.Met130Lys) in all three siblings — a single allelic heterozygous mutation, which is an atypical and rarely symptomatic genotype. This case series is significant as it highlights that heterozygous single allelic SLC52A3 mutations can produce significant clinical manifestations, expands the phenotypic spectrum of BVVL in an Indian familial context, and underscores the critical importance of early genetic diagnosis. All three patients were initiated on high-dose riboflavin supplementation (200 mg twice daily), with the eldest reporting meaningful functional improvement. This report reinforces that timely recognition and treatment of BVVL can substantially alter disease trajectory.

Key words: Brown-Vialetto-Van Laere syndrome, Juvenile onset MND, Riboflavin transporter deficiency, Ponto-bulbar palsy, SLC52A3

DOI: 10.5455/jmas.313210

Motor neuron diseases are among the most challenging diagnoses in neurology, both in diagnosis and management. Affecting approximately 4.5 per 100,000 people worldwide each year, these disorders carry significant morbidity. Brown-Vialetto-Von Laere syndrome (BVVL) is

Juvenile onset Anterior horn cell disorder, which is an extremely rare condition, first described in 1894 by Dr. Charles Henry Brown[1]. To date, fewer than 90 cases have been documented. Characteristic clinical features include bilateral, progressive ponto-bulbar palsy with degeneration of both upper and lower motor neurons. The primary cause of mortality is respiratory failure due to diaphragmatic involvement.

BVVL is distinguished from Fazio-Londe syndrome primarily by the presence of sensorineural hearing loss, though both are considered to fall within the same disease spectrum. Madras Motor neuron disease (MMND) classically presents in second decade, however rarely can present as early as 5 years of age with sensorineural hearing loss, cranial nerve involvement and lower motor neuron features.

Reported symptoms in the literature include ocular findings such as nystagmus, ptosis, opsoclonus, macular hyperpigmentation, and optic atrophy, as well as motor symptoms like early tremors, proximal muscle weakness, and pure motor quadriparesis. Ataxia has also been described in some cases. The disorder demonstrates heterogeneity, with both sporadic and familial occurrences. Most cases follow an autosomal recessive inheritance pattern, but autosomal dominant and X-linked forms have also been observed. The onset can range from infancy to the third decade of life, with earlier, more rapidly progressive disease associated with poorer outcomes.

Although the female-to-male ratio is approximately 3:1, males are often noted to have more severe disease progression compared to females, as reported in multiple case series.

Mutations in genes encoding intestinal riboflavin transporters—specifically SLC52A1, SLC52A3, and SLC52A2, which produce the proteins hRFT1, hRFT2, and hRFT3—are responsible for BVVL. Recognition of these genetic defects has been pivotal in transforming patient outcomes. The disorder is notable for its rapid progression and, importantly, for its often-dramatic improvement with early initiation of high-dose riboflavin supplementation. Awareness and timely diagnosis are crucial, as prompt treatment can significantly reduce both morbidity and mortality.

Here, we present a case series involving three siblings of Indian descent, born to non-consanguineous parents, who exhibited multiple bilateral cranial neuropathies affecting the VII, VIII, IX, X and XII nerves, along with sensorineural hearing loss. The eldest sibling also demonstrated features of pure motor quadriparesis.

The siblings initially presented to the Otorhinolaryngology outpatient department for audiometric evaluation and were subsequently referred to Neurology Department for further assessment. Their distinctive neurological findings and family history raised suspicion for Brown-Vialetto-Von Laere syndrome after other common etiologies were excluded. Clinical exome sequencing confirmed a heterozygous mutation in the SLC52A3 gene, consistent with autosomal recessive inheritance.

Case report 1

A 20-year-old female, the eldest of three siblings from non-consanguineous parents, presented with progressive facial weakness, speech difficulties, hoarseness and hearing impairment, all evolving over six years.

Her symptoms began at the age of 14 years with right-sided facial weakness following a febrile illness. Over the next year, the weakness extended bilaterally, accompanied by progressive speech impairment, hoarseness, and hearing loss. The subsequent year, she developed marked tongue atrophy, exacerbating her dysarthria and causing involuntary flickering movements of the tongue and facial muscles. She had no prior medical issues, and both her birth and developmental history were unremarkable.

There was no similar history in her parents or extended family, except in her two younger siblings.

General examination revealed a conscious, coherent, and cooperative young woman with normal vital signs, moderate build and nutrition, and no neurocutaneous stigmata or peripheral nerve thickening.

Neurologically, she was alert, oriented and cognitively intact. Examinations of cranial nerves I–VI were unremarkable. She exhibited bilateral lower motor neuron facial palsy. Audiometry indicated right-sided sensorineural hearing loss and mixed hearing loss on the left. Examination revealed uvular atrophy with absent gag reflex, no palatal deviation and notable bilateral tongue atrophy with fasciculations.

Motor assessment showed mild hypotonia with variable decline in muscle strength both proximally and distally of all limbs. Superficial and deep tendon reflexes were preserved, and plantar responses were normal. Bilateral facial fasciculations were present. Sensory and cerebellar systems were intact.

Routine laboratory investigations and ophthalmological assessment were unremarkable. Video laryngoscopy revealed bilateral vocal cord paralysis. Brain MRI was normal, while spinal imaging showed right thoracic scoliosis. Electroneuromyography (ENMG) demonstrated a chronic neurogenic pattern involving bulbar, cervical and lumbar regions. Given the atypical features and family history, hereditary causes were pursued.

Her riboflavin level was markedly low at 0.5 mcg/dl (reference: 4–24 mcg/dl). Clinical exome sequencing was subsequently performed. This identified a heterozygous missense mutation in exon 2 of the SLC52A3 gene (chr20:g.746030A>T, p.Met130Lys; ENST00000217254.7).

Figure 1 shows bilateral loss of wrinkling in the forehead and nasolabial folds. Bilateral facial weakness, asymmetrical in nature, with the left being weaker than the right; hence, a slight deviation of the mouth angle is seen towards the right. The tongue shows bilateral significant atrophy.

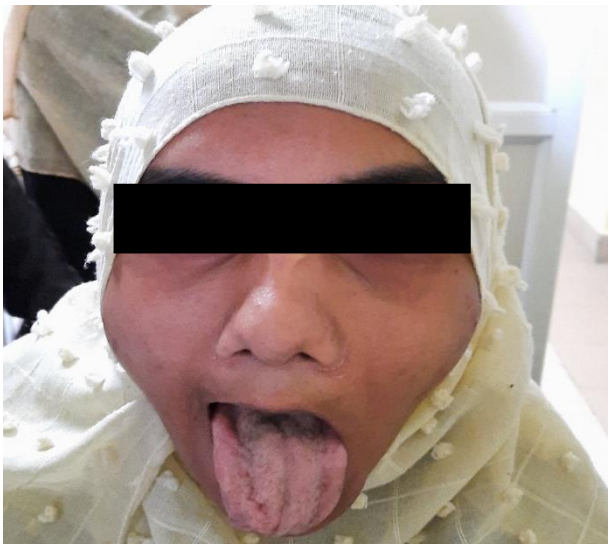

Figure 1. Bilateral loss of wrinkling in the forehead and nasolabial folds. The tongue shows bilateral significant atrophy

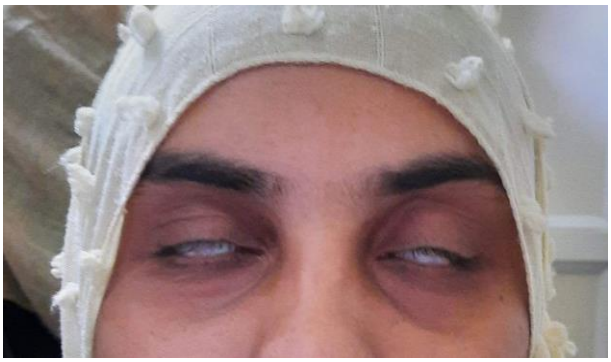

Figure 2. Shows bilateral Bell's phenomenon, indicating bilateral LMN facial palsy

Case report 2

An 18-year-old male, the younger brother of Case 1, exhibited a similar clinical course. His symptoms began after a febrile illness at the age of 12 years, presenting as bilateral facial palsy, and subsequently progressed to hearing loss, dysarthria and dysphonia. He also had a history of childhood-onset epilepsy. Birth, developmental and cognitive histories were normal. General and neurological examinations revealed bilateral LMN palsies of cranial nerves VII, VIII, IX, X and XII, with spontaneous facial and tongue fasciculations similar to those of his elder sibling. The remaining motor, sensory and cerebellar examinations were unremarkable. Audiological assessment showed bilateral mixed hearing loss. Video laryngoscopy confirmed bilateral vocal cord palsy. MRI of the brain and spine was normal. Plasma riboflavin was low at 1.3 mcg/dl (reference: 4–24 mcg/dl). Genetic testing revealed a heterozygous missense mutation in exon 2 of SLC52A3 (chr20:g.746030A>T, p.Met130Lys; ENST00000217254.7).

Case report 3

A 15-year-old female, the youngest sibling, experienced a similar initial febrile illness followed by bilateral facial palsy, progressively worsening hearing loss, dysphonia and dysarthria over six years. Neurological examination showed bilateral LMN palsies of cranial nerves VII, VIII, IX, X and XII, with no additional motor, sensory or cerebellar deficits. Audiometry revealed bilateral sensorineural hearing loss. Video laryngoscopy indicated bilateral vocal cord paresis. MRI of the brain and spine was unremarkable. Her plasma riboflavin was decreased at 2.2 mcg/dl (reference: 4–24 mcg/dl). Genetic analysis revealed the same heterozygous missense mutation in exon 2 of SLC52A3 (chr20:g.746030A>T, p.Met130Lys; ENST00000217254.7) as seen in her siblings.

Discussion

Brown-Vialetto-Von Laere syndrome (BVVL) (MIM 211530, ORPHA97229) is a rare clinical entity. The earliest documented case involved a 12-year-old boy exhibiting multiple cranial nerve palsies—bilateral VII, VIII, IX, X and XII—along with proximal muscle weakness and upper limb motor paresis. Dr. Brown emphasized the rarity of such symptoms in children and described it as "Infantile Amyotrophic Lateral Sclerosis of the Family Type"[1].

Subsequent reports by Vialetto in 1936[2] and Von Laere in 1966[3] added to the initial literature. As of 2008, only 58 cases had been described[4]. Common presentations include sensorineural hearing

loss, bulbar palsy, and respiratory failure, with onset ranging from infancy to adulthood. The female-to-male ratio is about 3:1, but males generally have more severe disease and higher mortality. Sathasivam provided a comprehensive review in 2008 in the Orphanet Journal of Rare Diseases[4].

Major progress in understanding BVVL occurred in 2010, when Green et al analyzed 9 cases from 7 families, 3 of which involved consanguinity. A notably consanguineous Arabic speaking family with several affected children who died in infancy strongly supported an autosomal recessive inheritance pattern[5]. They identified mutations in the gene C20orf54, now recognized as SLC52A3 (also known as BVVLS, BVVLS1, RFT2, RFVT3, bA371L19.1, hRFT2), as the cause of the disease[5].

In 2011, biochemical abnormalities in the acylcarnitine profile were reported by Bosch and colleagues, who described three cases with low riboflavin levels despite normal dietary intake, leading to the hypothesis that a riboflavin transporter defect underlies the disorder. They demonstrated that SLC52A3 encodes a protein closely related to the rat rRFT2, a known intestinal riboflavin transporter[6]. Patients showed marked improvement with riboflavin supplementation, supporting this theory. The following year, Haack et al identified mutations in SLC52A2, encoding riboflavin transporter 3 (hRFT3), another family member[7]. While SLC52A2 mutations did not lower plasma riboflavin, consistent with its role in tissue uptake, SLC52A3 mutations caused decreased plasma levels, reinforcing the rationale for high-dose riboflavin supplementation[7].

Furthermore, Ho et al reported a newborn who presented with clinical features highly suggestive of multiple acyl-CoA dehydrogenase deficiency (MADD) resulting from maternal riboflavin deficiency[8]. The mother was found to have a hemizygous deletion spanning exons 2 and 3 of the SLC52A1 gene (other aliases: GPCR42, GPR172B, PAR2, RFT1, RFVT1, hRFT1), which encodes another riboflavin transporter—the infant fully recovered after riboflavin treatment[8]. Koy et al described a case of a 3-year-old girl with early-onset Brown-Vialetto-Van Laere syndrome and a novel mutation in the C20orf54 gene (c.989G>T)[9]. It is now clear that proper diagnosis of BVVL requires mutation analysis of all three transporter genes, namely SLC52A1, SLC52A2 and SLC52A3 encoding for RFVT 1, 2 and 3 transporters, respectively[10].

Riboflavin, also known as vitamin B2, is a water-soluble aromatic substance that has to be absorbed from the diet. In humans, the riboflavin transporters strongly expressed in small intestine epithelial cells, hRFT1 and hRFT2 and hRFT3 encoded by SLC52A1, SLC52A3, SLC52A2, respectively, cooperatively function in riboflavin uptake and subsequent distribution into the blood[11].

Uptake into the target cells from the blood might occur via passive diffusion, but is more likely to depend on specific transport systems[12]. Inside the cells, riboflavin is converted into its bioactive coenzymes FMN and FAD. The latter functions as an electron acceptor for numerous dehydrogenases involved, e.g. in mitochondrial fatty acid oxidation and branched-chain amino acid catabolism. In rats, severe riboflavin deficiency mimics the metabolic signature of a multiple acyl-CoA dehydrogenase deficiency (MADD)[13]. Both decreased levels of riboflavin and its metabolites and biochemical findings suggestive of mild MADD have recently been reported in hRFT2 mutant BVVLS individuals (Bosch et al, 2011)[6]. Plasma acylcarnitine levels and urine organic acid levels normalized upon oral riboflavin supplementation, and clinical features such as muscle weakness improved within weeks[6]. Inheritance is mainly autosomal recessive[10,14,15]. However, Hawkins et al also described a case with autosomal dominant or X-linked inheritance[16]. In Fazio-Londe syndrome (FL) (MIM 211500, ORPHA56965), the clinical presentation is the same, except without the hearing loss. FL is considered to be the same disease entity as BVVL[17]. BVVL classically described in SLC52A2 and SLC52A3. Homozygous mutation are typically seen in consanguineous families with early-onset disease and rapid progression. Heterozygous mutations can either be compound variant or single allelic. Heterozygous single allelic mutation are rarely symptomatic, however in our case the clinical manifestation even though presented late, but were significant. Mutations in the SLC52A1 gene that cause disease lead to a temporary deficiency of riboflavin, starting in new-born whose mothers carry one heterozygous mutation in SLC52A1 (OMIM 615026). The clinical manifestations are atypical for the Riboflavin transporter deficiency (RTD) phenotype and typically resolve by the age of 2 years. The most common clinical features include progressive bilateral ponto-bulbar palsy involving VII – XII cranial nerves, progressive sensorineural hearing loss, facial weakness, and respiratory compromise. Sensorineural hearing loss appears to precede neurological signs[1,2,3,4,16]. However, this is not always the case, as seen in our case series, where all 3 siblings developed hearing loss after the appearance of neurological signs. In very rare

cases, affected individuals do not appear to develop deafness, presumably because they die before the hearing impairment develops[17]. Motor symptoms include proximal muscle weakness of the neck and shoulder muscles, and pure motor quadriparesis with upper and lower motor neuron signs.

The patients in our case series are similar to the classical presentation of ponto-bulbar palsy with sensorineural hearing loss. Case number 1 also has pure motor quadriparesis of LMN type. Respiratory compromise has not been observed in any of our cases, even 6 years after symptom onset. Sensory involvement is rare, with only one reported case of subjective blunting of pinprick sensation below the knees[18]. There are also case reports of eye involvement, including ptosis[19], nystagmus[20], optic atrophy [21], opsoclonus[22], decreased corneal sensation in both eyes[23] and external ophthalmoplegia[5]. In addition, ataxia leading to clumsiness and frequent falls[5,20], as well as tremors, has been reported.

Seizures were also noted in rare cases[19]. Case 2 of our series had epilepsy since childhood, controlled well with medication. Personality changes were also seen in a case reported by Allison et al[24]. A review of literature published by Sathasivam et al lists the following rare symptoms: retinitis pigmentosa, macular hyperpigmentation, autonomic dysfunction, intellectual disability and reduced horizontal eye movements[4]. In some cases, the trigger for presentation was a preceding infection, i.e. an upper respiratory tract infection[18,25]. All 3 cases of our series reported having a short-lived febrile illness before their symptoms started; the significance of this is still unknown. In one instance, the symptoms developed a few days after HBV vaccination[26], suggesting a neuroimmune trigger. Allison et al studied 3 cases presenting initially as neuroimmune disorders but later diagnosed as BVVL after failure of steroids and intravenous immunoglobulin[24].

Before the onset of explicit neurological signs and symptoms, patients may also have subtle early signs such as repeated falls, clumsiness, recurrent infections in early childhood and dyspnea on exertion[5,15,16]. Among the non-neurological features, respiratory compromise is the most common in BVVL. Diaphragmatic weakness and respiratory compromise lead to recurrent chest infections and respiratory failure, which are often the cause of patients' demise. Other non-neurological features that have been reported include auditory hallucinations, behavioral change, color blindness, diabetes insipidus, delayed puberty and hypogonadism, dysmorphic features, gynecomastia and hypertension[4].

Two unique cases, one of cor pulmonale secondary to hypoventilation-induced chronic hypoxia and hypercapnia, described by da Silva et al[27], and another reporting a case of sleep disordered breathing in BVVL by Miao et al[28], have also been studied. Our case 1 shows spinal scoliosis, as previously reported once[14].

BVVL is largely a clinical diagnosis characterized by a distinct pattern of cranial nerve involvement, sensorineural hearing loss and a family history. Neuroradiology is generally unremarkable in this disease; however, MRI in a case series by Koul et al showed hyperintense lesions in the floor of the 4th ventricle and Ponto-medullary junction, suggestive of degeneration of cranial nerve neurons[29]. In another case report by Ciccolella et al, brain MRI showed hyperintense signals on T2- and FLAIR-weighted sequences corresponding to the CST in the posterior arm of the internal capsule and the cerebral peduncles[30]. T2 hyperintensities in the middle cerebellar peduncles on T2-weighted images were also identified in a study[26].

Table 1: Distinguishing features between BVVL and Fazio-Londe disease

Feature	BVVL	Fazio-Londe disease
Age of onset	Infancy to Adulthood (8 years mean)	First decade of life
Causative gene	SLC52A1, SLC52A2, SLC52A3	SLC52A3
Sex predilection	Two-thirds female	Both sexes equally
Sensorineural hearing loss	Present	Absent
Bulbar features	Present	Present
Riboflavin levels	Reduced	Not Affected

An electroencephalogram may show an excess of theta activity or slow waves. Cerebrospinal fluid examination in BVVL may show mildly elevated protein content. Other investigations are usually performed to exclude other causes or to confirm the clinical signs in patients. Neurophysiological studies show changes consistent with chronic or active denervation in muscles. Motor nerve conduction velocities are usually normal. Sensory action potentials are rarely reduced. Visual evoked potentials performed in 15 cases showed normal latencies in 7 patients and prolonged latencies in the remaining 8. Brainstem auditory evoked potentials were abnormal when performed in 17 cases. When performed, audiometry consistently showed sensorineural deafness[4]. The electrocardiogram showed an incomplete right bundle branch block in one case[27].

Polysomnography demonstrated predominantly central sleep apnea with minimal obstructive sleep apnea in one case[28].

In patients with SLC52A3 mutations, both riboflavin and its metabolites are decreased, with accompanying biochemical evidence of mild MADD. Oral riboflavin supplementation rapidly normalizes acylcarnitine and organic acid profiles, accompanied by clinical improvement[6].

Other conditions which can be considered in differential diagnosis include Fazio-Londe disease, MADD, riboflavin responsive lipid storage myopathy (predominant motor symptoms, no cranial nerve involvement) (Table 1).

Clinical response and treatment

Before the discoveries by Green et al[5] and Bosch et al[6], the underlying mechanism of BVVL remained unknown, leading to numerous unsuccessful treatments attempts with steroids, intravenous immunoglobulin (IVIg) and immunosuppressive therapies. The recognition of riboflavin transporter defects as the cause of BVVL has revolutionized management, significantly improving patients' quality of life.

Anand et al were the first to document clear clinical improvement following early genetic diagnosis and initiation of high-dose riboflavin therapy in a BVVL patient. Their case received 150 mg of riboflavin twice daily (25 mg/kg/day). Within one week, the patient's motor abilities improved dramatically—from requiring support to sit, to regaining full mobility and even climbing stairs[31].

Many other case reports have described similarly remarkable recoveries with riboflavin supplementation[6,19,20,21,22,23,24,26]. Although the optimal dosage remains uncertain, initial regimens of 10 mg/kg/day in children and up to 1500 mg daily in adults have been utilized.

In our series, all three patients were started on riboflavin tablets at a dose of 200 mg twice daily, with ongoing clinical monitoring. Notably, Case 1 has reported subjective improvement in symptoms, particularly motor strength—she is now able to perform daily activities independently and execute fine-motor tasks, such as sewing clothes, to assist her family. While facial weakness and cranial nerve palsies remain, we continue to observe her and her siblings for further changes closely. Other two cases are being closely followed.

Conflict of interest: None

Acknowledgments: None

References

1. Brown CH. Infantile amyotrophic lateral sclerosis of the family type. The Journal of Nervous and Mental Disease. 1894 Nov; 19(11):707-716.
2. Vialetto E. Contributo alla forma ereditaria della paralisi bulbare progressiva. Riv Sper Freniat. 1936; 40:1–24.
3. Van Laere J. Paralysie bulbo-pontine chronique progressive familiale avec surdité. Un cas de syndrome de Klippel-Trenaunay dans la même fratrie. Problèmes diagnostiques et génétiques [Familial progressive chronic bulbopontine paralysis with deafness. A case of Klippel-Trenaunay syndrome in siblings of the same family. Diagnostic and genetic problems]. Rev Neurol (Paris). 1966 Aug;115(2):289-95. French. PMID: 5969547.
4. Sathasivam S. Brown-Vialetto-Van Laere syndrome. Orphanet J Rare Dis. 2008 Apr 17;3:9. doi: 10.1186/1750-1172-3-9. PMID: 18416855; PMCID: PMC2346457.
5. Green P, Wiseman M, Crow YJ, Houlden H, Riphagen S, Lin JP, Raymond FL, Childs AM, Sheridan E, Edwards S, Josifova DJ. Brown-Vialetto-Van Laere syndrome, a ponto-bulbar palsy with deafness, is caused by mutations in c20orf54. Am J Hum Genet. 2010 Mar 12;86(3):485-9. doi: 10.1016/j.ajhg.2010.02.006. Epub 2010 Mar 4. PMID: 20206331; PMCID: PMC2833371.
6. Bosch AM, Abeling NG, Ijlst L, Knoester H, van der Pol WL, Stroomer AE, Wanders RJ, Visser G, Wijburg FA, Duran M, Waterham HR. Brown-Vialetto-Van Laere and Fazio Londe syndrome is associated with a riboflavin transporter defect mimicking mild MADD: a new inborn error of metabolism with potential treatment. J Inherit Metab Dis. 2011 Feb;34(1):159-64. doi: 10.1007/s10545-010-9242-z. Epub 2010 Nov 26. PMID: 21110228; PMCID: PMC3026695.
7. Haack TB, Makowski C, Yao Y, Graf E, Hempel M, Wieland T, Tauer U, Ahting U, Mayr JA, Freisinger P, Yoshimatsu H, Inui K, Strom TM, Meitinger T, Yonezawa A, Prokisch H. Impaired riboflavin transport due to missense mutations in SLC52A2 causes Brown-Vialetto-Van Laere syndrome. J Inherit Metab Dis. 2012 Nov;35(6):943-8. doi: 10.1007/s10545-012-9513-y. Epub 2012 Aug 3. PMID: 22864630; PMCID: PMC3470687.
8. Ho G, Yonezawa A, Masuda S, Inui K, Sim KG, Carpenter K, Olsen RK, Mitchell JJ, Rhead WJ, Peters G, Christodoulou J. Maternal riboflavin deficiency, resulting in transient neonatal-onset glutaric aciduria Type 2, is caused by

a microdeletion in the riboflavin transporter gene GPR172B. Hum Mutat. 2011 Jan;32(1):E1976-84. doi: 10.1002/humu.21399. PMID: 21089064.
9. Koy A, Pillekamp F, Hoehn T, Waterham H, Klee D, Mayatepek E, Assmann B. Brown-Vialetto-Van Laere syndrome: a riboflavin-unresponsive patient with a novel mutation in the C20orf54 gene. Pediatr Neurol. 2012 Jun;46(6):407-9. doi: 10.1016/j.pediatrneurol.2012.03.008. PMID: 22633641.
10. Bosch AM, Stroek K, Abeling NG, Waterham HR, Ijlst L, Wanders RJ. The Brown-Vialetto-Van Laere and Fazio Londe syndrome revisited: natural history, genetics, treatment and future perspectives. Orphanet J Rare Dis. 2012 Oct 29;7:83. doi: 10.1186/1750-1172-7-83. PMID: 23107375; PMCID: PMC3517535.
11. Yao Y, Yonezawa A, Yoshimatsu H, Masuda S, Katsura T, Inui K. Identification and comparative functional characterization of a new human riboflavin transporter hRFT3 expressed in the brain. J Nutr. 2010 Jul;140(7):1220-6. doi: 10.3945/jn.110.122911. Epub 2010 May 12. PMID: 20463145.
12. Foraker AB, Khantwal CM, Swaan PW. Current perspectives on the cellular uptake and trafficking of riboflavin. Adv Drug Deliv Rev. 2003 Nov 14;55(11):1467-83. doi: 10.1016/j.addr.2003.07.005. PMID: 14597141.
13. Goodman SI. Organic aciduria in the riboflavin-deficient rat. Am J Clin Nutr. 1981 Nov;34(11):2434-7. doi: 10.1093/ajcn/34.11.2434. PMID: 6895440.
14. Mégarbané A, Desguerres I, Rizkallah E, Delague V, Nabbout R, Barois A, Urtizberea A. Brown-Vialetto-Van Laere syndrome in a large inbred Lebanese family: confirmation of autosomal recessive inheritance? Am J Med Genet. 2000 May 15;92(2):117-21. doi: 10.1002/(sici)1096-8628(20000515)92:2<117::aid-ajmg7>3.0.co;2-c. PMID: 10797435.
15. Malheiros JA, Camargos ST, Oliveira JT, Cardoso FE. A Brazilian family with Brown-Vialetto-van Laere syndrome with autosomal recessive inheritance. Arq Neuropsiquiatr. 2007 Mar;65(1):32-5. doi: 10.1590/s0004-282x2007000100008. PMID: 17420823.
16. Hawkins SA, Nevin NC, Harding AE. Pontobulbar palsy and neurosensory deafness (Brown-Vialetto-Van Laere syndrome) with possible autosomal dominant inheritance. J Med Genet. 1990 Mar;27(3):176-9. doi: 10.1136/jmg.27.3.176. PMID: 2325091; PMCID: PMC1017000.
17. Dipti S, Childs AM, Livingston JH, Aggarwal AK, Miller M, Williams C, Crow YJ. Brown-Vialetto-Van Laere syndrome; variability in age at onset and disease progression highlighting the phenotypic overlap with Fazio-Londe disease. Brain Dev. 2005 Sep;27(6):443-6. doi: 10.1016/j.braindev.2004.10.003. Epub 2004 Dec 15. PMID: 16122634.
18. Sathasivam S, O'Sullivan S, Nicolson A, Tilley PJ, Shaw PJ. Brown-Vialetto-Van Laere syndrome: case report and literature review. Amyotroph Lateral Scler Other Motor Neuron Disord. 2000 Sep;1(4):277-81. doi: 10.1080/14660820050515106. PMID: 11465021.
19. Horoz OO, Mungan NO, Yildizdas D, Hergüner Ö, Ceylaner S, Kör D, Waterham H, Coskun T. Brown-Vialetto-Van Laere syndrome: two siblings with a new mutation and dramatic therapeutic effect of high-dose riboflavin. J Pediatr Endocrinol Metab. 2016 Feb;29(2):227-31. doi: 10.1515/jpem-2015-0198. PMID: 26444347.
20. Shashi V, Petrovski S, Schoch K, Crimian R, Case LE, Khalid R, El-Dairi MA, Jiang YH, Mikati MA, Goldstein DB. Sustained therapeutic response to riboflavin in a child with a progressive neurological condition, diagnosed by whole-exome sequencing. Cold Spring Harb Mol Case Stud. 2015 Oct;1(1):a000265. doi: 10.1101/mcs.a000265. PMID: 27148562; PMCID: PMC4850881.
21. Bamaga AK, Maamari RN, Culican SM, Shinawi M, Golumbek PT. Child Neurology: Brown-Vialetto-Van Laere syndrome: Dramatic visual recovery after delayed riboflavin therapy. Neurology. 2018 Nov 13;91(20):938-941. doi: 10.1212/WNL.0000000000006498. PMID: 30420458; PMCID: PMC6260193.
22. Cosgrove J, Datta S, Busby M. Adult onset Brown-Vialetto-Van Laere syndrome with opsoclonus and a novel heterozygous mutation: a case report. Clin Neurol Neurosurg. 2015 Jan;128:1-3. doi: 10.1016/j.clineuro.2014.10.016. Epub 2014 Nov 4. PMID: 25462087.
23. Koul R, Jain R, Chacko A, Alfutaisi A, Hashim J, Chacko J. Pontobulbar palsy and neurosensory deafness (Brown-Vialetto-van Laere syndrome) with hyperintense brainstem nuclei on magnetic resonance imaging: new finding in three siblings. J Child Neurol. 2006 Jun;21(6):523-5. doi: 10.1177/08830738060210060501. PMID: 16948939.
24. Allison T, Roncero I, Forsyth R, Coffman K, Pichon JL. Brown-Vialetto-Van Laere Syndrome as a Mimic of Neuroimmune Disorders: 3 Cases From the Clinic and Review of the Literature. J Child Neurol. 2017 May;32(6):528-532. doi: 10.1177/0883073816689517. Epub 2017 Jan 24. PMID: 28116953.
25. Ciccolella M, Catteruccia M, Benedetti S, Moroni I, Uziel G, Pantaleoni C, Chiapparini L, Bizzi A, D'Amico A, Fattori F, Salsano ML, Pastore A, Tozzi G, Piemonte F, Bertini E. Brown-Vialetto-van Laere and Fazio-Londe overlap syndromes: a clinical, biochemical and genetic study. Neuromuscul Disord. 2012 Dec;22(12):1075-82. doi: 10.1016/j.nmd.2012.05.007. Epub 2012 Jul 21. PMID: 22824638.
26. Bandettini Di Poggio M, Monti Bragadin M, Reni L, Doria-Lamba L, Cereda C, Pardini M, Roccatagliata L, Rossi A, Schenone A. Brown-Vialetto-Van Laere syndrome: clinical and neuroradiological findings of a genetically proven patient. Amyotroph Lateral Scler Frontotemporal Degener. 2014 Mar;15(1-2):141-4. doi: 10.3109/21678421.2013.837931. Epub 2013 Oct 1. PMID: 24079556.
27. da Silva-Júnior FP, Moura Rde D, Rosemberg S, Marchiori PE, Castro LH. Cor pulmonale in a patient with Brown-Vialetto-Van Laere syndrome: a case report. J Neurol Sci. 2011 Jan 15;300(1-2):155-6. doi: 10.1016/j.jns.2010.10.010. Epub 2010 Nov 5. PMID: 21055769.
28. Miao J, Li H, Lin H, Su C, Liu Y, Lei G, Yang T, Li Z. Severe sleep-disordered breathing in a patient with Brown-Vialetto-Van Laere syndrome: polysomnographic findings. J Neurol Sci. 2007 Dec 15;263(1-2):214-7. doi: 10.1016/j.jns.2007.06.032. Epub 2007 Jul 31. PMID: 17669429.
29. Koul R, Jain R, Chacko A, Alfutaisi A, Hashim J, Chacko J. Pontobulbar palsy and neurosensory deafness (Brown-Vialetto-van Laere syndrome) with hyperintense brainstem nuclei on magnetic resonance imaging: new finding in three siblings. J Child Neurol. 2006 Jun;21(6):523-5. doi: 10.1177/08830738060210060501. PMID: 16948939.
30. Ciccolella M, Catteruccia M, Benedetti S, Moroni I, Uziel G, Pantaleoni C, Chiapparini L, Bizzi A, D'Amico A, Fattori F, Salsano ML, Pastore A, Tozzi G, Piemonte F, Bertini E. Brown-Vialetto-van Laere and Fazio-Londe overlap syndromes: a clinical, biochemical and genetic study. Neuromuscul Disord. 2012 Dec;22(12):1075-82. doi: 10.1016/j.nmd.2012.05.007. Epub 2012 Jul 21. PMID: 22824638.
31. Anand G, Hasan N, Jayapal S, Huma Z, Ali T, Hull J, Blair E, McShane T, Jayawant S. Early use of high-dose riboflavin in a case of Brown-Vialetto-Van Laere syndrome. Dev

Med Child Neurol. 2012 Feb;54(2):187-9. doi: 10.1111/j.1469-8749.2011.04142.x. Epub 2011 Nov 18. PMID: 22098162.

32. Johnson JO, Gibbs JR, Van Maldergem L, Houlden H, Singleton AB. Exome sequencing in Brown-Vialetto-van Laere syndrome. Am J Hum Genet. 2010 Oct 8;87(4):567-9; author reply 569-70. doi: 10.1016/j.ajhg.2010.05.021. PMID: 20920669; PMCID: PMC2948797.