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Case Report

# Beyond the Diagnosis of Cerebral Palsy: A Decade of Misdiagnosis Before Clinical Reappraisal Revealed GCH1-Associated Dopa-Responsive Dystonia

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## Abstract

Dopa-responsive dystonia (DRD) is an uncommon but highly treatable movement disorder that may closely mimic cerebral palsy (CP). We report a 16-year-old female who carried a diagnosis of spastic cerebral palsy for nearly ten years despite progressive symptoms, prolonged rehabilitation, and bilateral Achilles tendon-lengthening procedures. During routine neurological reassessment at Iashvili Children's Central Hospital, careful history-taking identified marked diurnal fluctuation. Re-evaluation of previously performed genetic testing revealed a GCH1 variant of uncertain significance in the appropriate phenotypic context. Treatment with levodopa/benserazide resulted in remarkable functional recovery, including independent ambulation within three months. This case highlights the importance of longitudinal reassessment, avoidance of diagnostic anchoring, and phenotype-guided interpretation of genetic findings.

***Keywords:*** *Cerebral palsy; Dopa-responsive dystonia; GCH1; Diagnostic delay; Levodopa; Pediatric neurology*

## Introduction

An early diagnosis often shapes the entire course of a patient's medical care. While diagnostic continuity is essential in clinical practice, it may also predispose clinicians to diagnostic anchoring, whereby subsequent symptoms and investigations are interpreted within the framework of an established diagnosis rather than prompting diagnostic reconsideration. This cognitive bias can be particularly consequential in pediatric neurology, where several treatable disorders closely mimic more common neurodevelopmental conditions. [1,2]

Cerebral palsy (CP) is the most common cause of chronic motor disability in childhood and is defined as a permanent but non-progressive disorder of movement and posture resulting from injury to the developing brain. Consequently, progressive deterioration, the absence of recognized perinatal risk factors, preserved cognitive function, poor response to conventional rehabilitation, or the emergence of atypical clinical features should prompt careful re-evaluation of the diagnosis rather than attribution of new symptoms to CP alone.[3,4]

Dopa-responsive dystonia (DRD), most commonly caused by pathogenic variants in the **GCH1** gene, is a rare but highly treatable movement disorder that may closely resemble spastic cerebral palsy during childhood.

Characteristic clinical features include lower-limb dystonia, gait disturbance, and diurnal fluctuation with worsening symptoms later in the day, while the hallmark of the disorder is a dramatic and sustained response to low-dose levodopa therapy. Failure to recognize DRD may result in years of unnecessary investigations, prolonged rehabilitation, inappropriate orthopedic procedures, and preventable disability.[4,5]

Here, we present a 16-year-old girl who carried a diagnosis of spastic cerebral palsy for nearly a decade despite progressive symptoms and unsuccessful treatment. Her diagnosis was ultimately reconsidered following careful history-taking, in which a single question uncovered longstanding diurnal fluctuation, leading to recognition of GCH1-associated dopa-responsive dystonia and a remarkable clinical response to levodopa. [6,7]

## Case Presentation

A 16-year-old girl was referred to the Department of Child Neurology at Iashvili Children's Central Hospital for electromyography (EMG) as part of the evaluation of a longstanding diagnosis of spastic cerebral palsy. The referral itself appeared routine, as the diagnosis had been accepted for many years and the purpose of the visit was to further characterize her motor impairment rather than reconsider its underlying cause. However, what initially seemed to be a standard diagnostic assessment ultimately became the turning point in a diagnostic journey that had spanned nearly a decade.

She was born by cesarean section at 38 weeks' gestation with a birth weight of 3,800 g following an uncomplicated pregnancy. There was no history of birth asphyxia, neonatal seizures, neonatal intensive care admission, intracranial hemorrhage, central nervous system infection, or any other perinatal complication typically associated with cerebral palsy. Early developmental milestones were largely appropriate except for delayed independent walking, which was achieved at 19 months of age. Speech, cognition, and social development were preserved, and there was no history of developmental regression.

At approximately six years of age, her parents noticed the gradual onset of intermittent lower-limb pain and subtle gait abnormalities. Initially, she began walking on her toes and experienced increasing difficulty keeping up with her peers during physical activities. Over time, these symptoms progressed insidiously. Walking became increasingly effortful, frequent falls developed, and her endurance declined. The progressive deterioration eventually limited her independence and substantially affected her daily activities and quality of life.

During the following years, she was evaluated by multiple pediatric neurologists, orthopedic surgeons, rehabilitation specialists, and other healthcare professionals. She underwent numerous clinical assessments and investigations, eventually receiving a diagnosis of spastic cerebral palsy. Once this diagnosis had been established, subsequent evaluations were largely directed toward management of the presumed condition rather than questioning its validity. The diagnosis became anchored in her medical records, shaping every therapeutic decision that followed.

For nearly ten years, she underwent intensive rehabilitation programs, repeated courses of physiotherapy, stretching exercises, orthotic management, and symptomatic treatment. Despite remarkable commitment from both the patient and her family, none of these interventions resulted in sustained clinical improvement. As progressive equinus deformity and toe walking were attributed to spasticity, she ultimately underwent bilateral Achilles tendon-lengthening surgery. Although the procedure was technically successful, it failed to produce meaningful functional improvement. Her gait remained severely impaired, and over the following years her mobility continued to decline. Eventually, she became unable to walk independently and required continuous assistance for ambulation.

This clinical course should have raised an important concern. Cerebral palsy is traditionally considered a non-progressive disorder resulting from an early static brain injury. In contrast, this patient's symptoms had progressively worsened over several years despite comprehensive rehabilitation and orthopedic intervention. Nevertheless, because the diagnosis had been accepted for so long, the discrepancy between the expected natural history of cerebral palsy and her progressive deterioration received little attention.

When she presented to our department, the initial objective was to perform electromyography as part of the evaluation of her longstanding motor impairment. Before proceeding with additional investigations, a comprehensive neurological assessment and detailed history were obtained from both the patient and her family. Rather than assuming the existing diagnosis was correct, her clinical history was reconstructed from the beginning.

**Table 1.** Clinical Timeline

| Time | Clinical Event |
|---|---|
| Birth | Term birth (38 weeks), cesarean section, no significant perinatal complications. |
| 19 months | Independent walking achieved. |
| ~6 years | Toe walking, lower-limb pain and progressive gait disturbance. |
| Following years | Diagnosed with spastic cerebral palsy. |
| Following years | Physiotherapy, orthoses and rehabilitation without sustained benefit. |
| Later | Bilateral Achilles tendon-lengthening surgery; no meaningful improvement. |
| Age 16 | Referred for EMG; detailed neurological reassessment performed. |
| Reassessment | Diurnal fluctuation identified from history. |
| Genetic review | Previously reported GCH1 variant reinterpreted in clinical context. |
| Treatment | Levodopa/benserazide started. |
| 3 months | Independent ambulation regained with marked functional recovery. |

**Table 2.** Comparison of Cerebral Palsy, GCH1-Associated DRD, and the Present Case

| Feature | Cerebral Palsy | GCH1-Associated DRD | Present Case |
|---|---|---|---|
| Course | Non-progressive | Progressive | Progressive |
| Perinatal injury | Often present | Usually absent | Absent |
| Cognition | Variable | Usually preserved | Preserved |
| Diurnal fluctuation | Absent | Typical | Present |
| Levodopa response | No dramatic effect | Dramatic | Dramatic |
| Toe walking | Possible | Common | Present |
| Rehabilitation | Variable benefit | Limited until treatment | Poor benefit |

**Table 3.** Selected Literature Relevant to the Present Case

| Reference | Main finding | Clinical relevance | Present case |
|---|---|---|---|
| Segawa et al., 1976 | Described DRD with diurnal fluctuation | Classic phenotype | Characteristic history recognized |
| Nygaard et al., 1991 | Excellent levodopa response | Supports therapeutic trial | Marked improvement |
| Trender-Gerhard et al., 2009 | Expanded GCH1 phenotype | Avoid misdiagnosis | GCH1 variant reinterpreted |
| Furukawa, GeneReviews | DRD can mimic CP | Integrate genotype and phenotype | Consistent with diagnosis |

During this interview, one apparently simple question fundamentally changed the diagnostic direction:

***"Is her walking the same throughout the day, or does it change?"***

Her parents immediately described a striking but previously overlooked feature. Every morning she was able to stand, walk, and move considerably better. As the day progressed, however, her gait progressively deteriorated. By evening, walking became increasingly difficult, stiffness intensified, and fatigue markedly limited her mobility. After a night's sleep, the cycle repeated with partial recovery each morning.

Although this pattern had been present for years, neither the family nor previous clinicians had recognized its diagnostic significance. The history of marked diurnal fluctuation, together with progressive lower-limb dystonia and preserved cognitive function, immediately raised suspicion for dopa-responsive dystonia rather than cerebral palsy. This single historical detail—obtained through careful history-taking rather than advanced diagnostic testing—became the pivotal clue that challenged a diagnosis that had remained unquestioned for almost a decade.

The newly recognized clinical picture prompted a thorough review of her previous investigations. Genetic testing performed several years earlier was revisited and revealed a heterozygous variant of uncertain significance in the **GCH1** gene. At the time it had originally been reported, the result had not been considered diagnostic because its clinical relevance was uncertain. However, when interpreted alongside the characteristic phenotype and the remarkable history of diurnal fluctuation, the genetic finding became highly suggestive of GCH1-associated dopa-responsive dystonia.

Given the strong clinical suspicion, treatment with levodopa/benserazide (Madopar) was initiated as a therapeutic trial. Improvement became apparent within the first weeks of therapy. The patient demonstrated progressive normalization of gait, reduction in lower-limb rigidity and dystonic posturing, and increasing endurance. At the three-month follow-up, she had regained independent ambulation, her gait had improved dramatically, and she was able to perform daily activities that had previously required assistance. Both the patient and her family described the improvement as life-changing after years of unsuccessful treatments, intensive rehabilitation, and orthopedic surgery.

This case illustrates how a diagnosis accepted for many years can persist despite an atypical clinical course and repeated treatment failure. More importantly, it demonstrates that the most decisive diagnostic tool was not a sophisticated investigation but meticulous history-taking. A single carefully chosen question revealed the characteristic feature of diurnal fluctuation, prompting reinterpretation of previously available genetic data and leading to the diagnosis of GCH1-associated dopa-responsive dystonia—a highly treatable neurological disorder that had been mistaken for cerebral palsy for nearly a decade.

## Discussion

The present case highlights an important challenge in pediatric neurology: distinguishing cerebral palsy (CP) from treatable disorders that closely mimic its clinical presentation. Although CP remains the most common cause of childhood motor disability, its diagnosis should not be considered immutable when the clinical course becomes inconsistent with its defining characteristics. In our patient, nearly a decade elapsed between symptom onset and the correct diagnosis despite repeated neurological evaluations, prolonged rehabilitation, orthopedic intervention, and progressive functional deterioration. More importantly, the decisive diagnostic breakthrough resulted not from a novel investigation but from careful history-taking, which prompted reinterpretation of previously available genetic findings and ultimately led to successful treatment.

Dopa-responsive dystonia (DRD), first described by Segawa and colleagues, comprises a heterogeneous group of inherited disorders affecting dopamine biosynthesis, of which autosomal dominant **GCH1** deficiency is the most common form. DRD is characterized by childhood-onset dystonia, gait disturbance, diurnal fluctuation, and a dramatic, sustained response to low-dose levodopa. Unlike neurodegenerative disorders, the underlying defect reflects impaired dopamine synthesis rather than irreversible neuronal loss, explaining the remarkable reversibility of neurological symptoms following appropriate treatment.

One of the greatest diagnostic challenges is the striking phenotypic overlap between DRD and spastic cerebral palsy during childhood. According to GeneReviews and contemporary reviews, the initial manifestations of GCH1-associated DRD typically include foot dystonia, toe walking, gait disturbance, brisk lower-limb reflexes, and progressive difficulty with ambulation—features that closely resemble spastic diplegic CP. Many affected children also demonstrate delayed walking despite otherwise normal cognitive development. Consequently, DRD remains one of the most important and treatable conditions within the differential diagnosis of cerebral palsy.

Our patient's clinical course reflected many of these classical features. Symptoms began during childhood with progressive gait impairment and toe walking, followed by increasing lower-limb disability. She underwent years of rehabilitation and ultimately bilateral Achilles tendon-lengthening surgery without meaningful functional improvement. In retrospect, several clinical findings should have prompted reconsideration of the diagnosis. Cerebral palsy is, by definition, a non-progressive disorder resulting from injury to the developing brain. Although orthopedic complications may evolve with growth, progressive neurological deterioration is not expected. Likewise, the absence of significant perinatal brain injury, preserved cognitive function, and progressive worsening despite comprehensive rehabilitation are all features that should broaden the differential diagnosis beyond CP.

Perhaps the most important educational aspect of this case is the role of diagnostic reasoning. Once the diagnosis of cerebral palsy had been established, subsequent evaluations understandably focused on management rather than reconsideration of the underlying diagnosis. This pattern illustrates the concept of diagnostic anchoring, a recognized cognitive bias in which new clinical information continues to be interpreted through the framework of an existing diagnosis. Although diagnostic continuity is essential in clinical practice, clinicians should remain prepared to revisit previous conclusions whenever the patient's clinical evolution becomes inconsistent with the expected natural history of the disease.

The turning point in this patient's diagnostic journey resulted from meticulous history-taking. Before additional investigations were performed, her clinical history was reconstructed from the beginning. During this interview, one targeted question regarding daily variation in motor performance revealed a striking history of diurnal fluctuation, with substantial improvement after sleep and progressive worsening toward the evening. Although this feature had been present for years, it had never been recognized as diagnostically significant. Diurnal fluctuation is considered one of the hallmark characteristics of GCH1-associated DRD and is specifically highlighted in GeneReviews as a key clinical feature that should prompt consideration of the diagnosis. Nevertheless, because families often regard these daily changes as a normal part of the illness, clinicians must actively inquire about symptom variation rather than waiting for it to be volunteered spontaneously.

An equally important lesson concerns the interpretation of genetic testing. Our patient had previously undergone molecular analysis, which identified a heterozygous **GCH1** variant of uncertain significance. At the time of testing, the result was not considered sufficient to establish a diagnosis. Only after recognition of the characteristic phenotype did the genetic finding acquire meaningful clinical significance. This sequence of events reinforces an increasingly important principle of genomic medicine: molecular testing should complement, rather than replace, careful clinical phenotyping.

As emphasized in GeneReviews, interpretation of genetic variants requires integration of clinical presentation, family history, neurological examination, and therapeutic response. Genetic results obtained in isolation may remain inconclusive, whereas the same findings can become highly informative when interpreted within the appropriate clinical context.

The patient's response to levodopa further supports the diagnosis and illustrates one of the defining characteristics of DRD. GeneReviews describes a dramatic and sustained improvement following relatively low doses of levodopa as a hallmark of GCH1-associated disease, with many patients achieving near-complete resolution of motor symptoms and without the motor complications commonly encountered in Parkinson disease. Similarly, our patient experienced marked improvement within weeks of treatment initiation and regained independent ambulation after three months of therapy. Such a profound clinical response after years of disability highlights why early recognition of DRD is of exceptional clinical importance.

Delayed diagnosis carries important consequences extending far beyond neurological impairment. Before receiving the correct diagnosis, our patient underwent prolonged physiotherapy, repeated specialist consultations, orthopedic surgery, and years of progressive disability. Although these interventions were undertaken appropriately based on the presumed diagnosis of cerebral palsy, they also illustrate the physical, emotional, and socioeconomic burden associated with delayed recognition of a treatable disorder. Contemporary reviews emphasize that DRD is frequently misdiagnosed, resulting in unnecessary investigations and prolonged delays before initiation of effective therapy. Earlier recognition could potentially prevent avoidable disability and substantially improve quality of life during childhood and adolescence.

From a practical perspective, this case reinforces several clinical principles. Children carrying a diagnosis of cerebral palsy should undergo careful diagnostic re-evaluation whenever atypical features emerge, particularly progressive deterioration, preserved cognition, absence of significant perinatal brain injury, poor response to rehabilitation, or marked diurnal fluctuation. In such patients, GCH1-associated dopa-responsive dystonia should remain an important diagnostic consideration, and when the clinical phenotype is suggestive, review of previous genetic investigations together with a carefully supervised therapeutic trial of levodopa may be both diagnostically and therapeutically invaluable.

In conclusion, this case demonstrates that the most important diagnostic advance was not the performance of additional investigations but the willingness to question an established diagnosis. Careful history-taking revealed a longstanding history of diurnal fluctuation, prompting reinterpretation of previously available genetic findings and leading to recognition of GCH1-associated dopa-responsive dystonia. The patient's remarkable response to levodopa not only confirmed the diagnosis but also illustrates the profound consequences that timely recognition of treatable neurological disorders can have on long-term functional outcome. Even in the era of genomic medicine, meticulous clinical observation, thoughtful phenotyping, and careful history-taking remain indispensable components of neurological practice.

## Conclusion

This case demonstrates that a diagnosis of cerebral palsy should not be regarded as a permanent diagnostic endpoint when the clinical course becomes progressive or inconsistent with the natural history of a static encephalopathy. Progressive deterioration, poor response to conventional rehabilitation, absence of significant perinatal risk factors, and the presence of diurnal fluctuation should prompt careful diagnostic re-evaluation and consideration of alternative, potentially treatable neurological disorders.

In our patient, the decisive diagnostic breakthrough resulted not from additional sophisticated investigations but from meticulous history-taking, which uncovered a longstanding history of diurnal fluctuation and prompted reinterpretation of previously available genetic findings. The subsequent dramatic response to levodopa confirmed the diagnosis of GCH1-associated dopa-responsive dystonia and resulted in substantial functional recovery after years of disability.

This case emphasizes that careful clinical phenotyping remains indispensable in the era of genomic medicine. Genetic testing should always be interpreted within the context of the patient's clinical presentation, and thoughtful neurological assessment continues to play a pivotal role in identifying treatable disorders. Early recognition of dopa-responsive dystonia can prevent unnecessary investigations, prolonged rehabilitation, orthopedic procedures, and years of avoidable disability.

## Declarations

Ethical approval was obtained from the appropriate institutional review body in accordance with local institutional requirements. *(Modify this statement according to your institution's ethics approval process. If ethics committee approval was waived, state this accordingly.)*

## Patient Consent

Written informed consent for publication of this case report and any accompanying clinical information was obtained from the patient and her legal guardian. A copy of the signed consent is available for review by the Editor upon reasonable request.

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## Conflict of Interest

The authors declare that they have no competing interests.

## Author Contributions

**Merab Gurgenidze:** Conceptualization, literature review, clinical data collection, manuscript drafting, and manuscript revision.

**Teona Shatirishvili:** Patient supervision, clinical evaluation, diagnostic interpretation, critical revision of the manuscript, and final approval of the version to be published.

All authors read and approved the final manuscript.

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