Rethinking dyspraxia diagnoses in children: the example of DYT-*THAP1*.

David Aktan[1,2], MD; Charlotte Mouraux[1,3], MD; Frédérique Depierreux[1,2], MD, PhD

Affiliations:

[1] GIGA – CRC – Rare Movement Disorders Research Group, University of Liège, Liège, Belgium

[2] Department of Neurology, University Hospital of Liège, Liège, Belgium

[3] Department of Human Genetics, University Hospital of Liège, Liège, Belgium

Corresponding author:

Dr AKTAN David
Department of Neurology
University Hospital of Liege
1 Avenue de l'Hôpital, 4000 Liege, Belgium

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Abstract

Introduction: DYT-*THAP1* is a monogenic form of autosomal dominant isolated dystonia caused by heterozygous pathogenic variants in the *THAP1* gene [1]. It typically presents in childhood or adolescence with segmental or generalised dystonia, often affecting the upper limbs, cervical region, and craniofacial muscles [2]. Diagnosis may be delayed due to misphenotyping and inappropriate genetic testing [3].

Case report: We report the case of a progressive motor limb dysfunction in an 11-year-old patient, initially misdiagnosed as dyspraxia and dysgraphia. Symptoms began at the age of 5 and were later recognized as task-specific hand dystonia. The condition evolved into generalised dystonia with brachiocranial predominance, including cervical dystonia, but also involvement of the right foot. Neuroimaging revealed T2 hyperintensities in the basal ganglia and cerebellar dentate nuclei. Genetic testing identified a *de novo* heterozygous frameshift variant in *THAP1* (c.4delG, p.Val2Cysfs*71), classified as likely pathogenic.

Conclusion: This case illustrates a typical presentation of DYT-*THAP1* and underscores the importance of early referral to specialised movement disorder centres [4]. Recognising dystonic features behind apparent developmental motor disorders is crucial for timely diagnosis [5]. Genetic testing plays a key role in establishing a definite diagnosis and guiding personalised management strategies, including botulinum toxin therapy and deep brain stimulation [3,6].

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