

HYPERKINETIC MOVEMENT DISORDERS IN A BELGIAN TERTIARY CARE CENTRE

AN EPIDEMIOLOGICAL STUDY

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INTRODUCTION

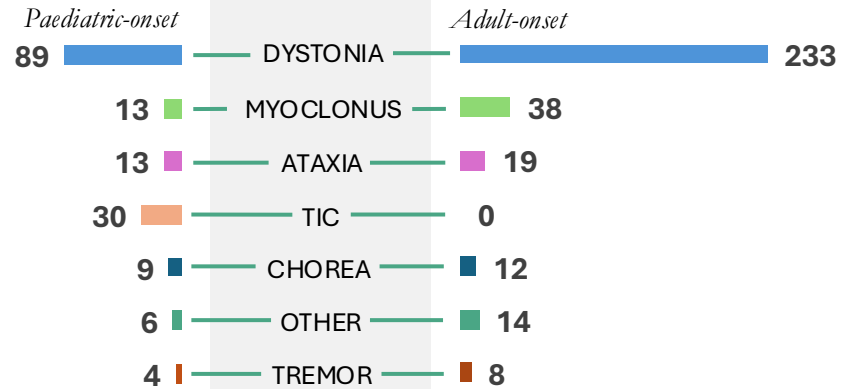
Hyperkinetic MD are rare disorders with involuntary movements, often genetic (1,2). Belgian epidemiology data are scarce but vital for care and policy (3).

METHODS

- Two-year observational study at Liege MD clinic
- Clinical and demographic data collected
- Genetic testing: NGS panel, WES, CGH as needed

Figure 1

Distribution of 488 patients by dominant hyperkinetic MD subtype and age at onset, with paediatric-onset cases on the left and adult-onset cases on the right.



RESULTS

- 488 patients (n paediatric/adult = 164/324) with hyperkinetic MD included
- Dystonia: most frequent subtype (66%)
- Among dystonia patients (n=323):
 - Genetic diagnosis in 14.6%
 - Paediatric-onset: 40.4% (36/89), Adult-onset: 5.1% (12/234)
- Diagnostic yield:
 - NGS panels: 10.3% overall, 27.7% in paediatric cases
 - WES & CGH (selected patients): >75% yield
 - Most frequent genes: *SGCE*, *THAP1*, *MYH3*, *PANK2*, *TOR1A*, *KMT2B*, *ATP7B*, *FA2H*

CONCLUSION

- Dystonia predominates in our tertiary referral centre with dedicated botulinum toxin clinic
- Systematic NGS + WES + CGH improves diagnosis, especially in paediatrics
- Findings inform care strategies and health policy planning

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