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What does an aetiological diagnosis actually mean?

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A diagnosis of cerebral palsy (CP) is based on a clinical description; it is a lifelong disorder of movement and posture that results from atypical development or injury to the developing brain^{1,2} and is aetiologically agnostic. In recent years, new genetic variants have been identified in CP research.³ Here, Aravamuthan et al. explore the views of people with lived experience of CP to understand their preferences for the use of diagnostic labels particularly in regard to CP and genetic aetiologies.⁴

In this study, the participant inclusion criteria did not specify either a genetic aetiology or age limit. The findings therefore reflect participants' recollections of likely historic discussions regarding genetic and non-genetic aetiological risk factors, and imaging findings, paired with their current preferences in relation to diagnostic labels. Throughout the paper, it is frequently unclear as to whether the authors are referring to aetiological diagnostic labels that they suggest a person might 'carry', or aetiological risk factors that should be discussed and documented. Putting these and other methodological issues to one side, the authors conclude that most surveyed people with lived experience of CP, preferred to 'carry' a CP diagnostic label alongside an aetiological one.

Individuals with CP and their families reported that the CP diagnostic label is helpful in assisting them to access services and provides a shared term of reference in the clinic and the community, independent of aetiology. Simultaneously, they valued knowing the aetiological risk factors and brain imaging findings that could help explain their CP.

Encouragingly, most families recalled clinicians sharing information about risk factors and brain imaging. However, these factors were not always recalled as potential causes/contributors of their CP. This disconnect is unsurprising considering the complexity of the causal pathways to CP, the impenetrability of some medical terminology (including those suggested for use by the authors), the reliability of recall after such long periods, and the reality that new

aetiological information can emerge over time. We agree with the participants and authors that ongoing discussions regarding aetiology are an important part of the journey towards understanding and making sense of a CP diagnosis. However, whilst Aravamuthan et al. suggest that aetiological information may alleviate unfounded guilt for some caregivers, it is important to note that this information is not always available, or helpful, to all.

Based upon participants' preferences of 'carrying' both CP and genetic diagnoses, the authors recommend a model for creating a diagnostic label: CP and (genetic) aetiology ± imaging pattern. This builds on previous consensus statements on this topic.³ It is presumed in this case that the genetic variants are those that explain the CP motor disorder. In the case of non-genetic aetiologies, which account for the majority of CP, it is unclear whether Aravamuthan et al. are proposing the same model in terms of adding aetiological details to the CP diagnostic label. As partially acknowledged by the authors, more consultation with people with lived experience of CP is required to determine whether an expanded and possibly lengthy diagnostic label of this kind would be useful or helpful. For example, when communicating with teachers, support workers, or employers, attaching a long, potentially triggering, difficult-to-say list of risk factors and imaging patterns (such as placental abruption, non-accidental injury, and periventricular leukomalacia) to the diagnostic label of CP may be unhelpful and/or redundant. In day-to-day life where emphasis is placed on personal goals, recognizing support needs, and fostering individual strengths and talents,⁵ is the additional lengthy aetiological information actually helpful? On a practical note, it is also unclear how clinicians would determine which risk factors should be included in a proposed aetiological diagnostic label. Should all aetiological risk factors be included or only those most proximal to the brain injury? Who decides and when?

There are significant methodological issues with this study, including a lack of clarity as to what the authors actually mean by an aetiological diagnosis. Aravamuthan et al. should, however, be commended for seeking the opinions of people with lived experience of CP. In the

Both authors of this commentary have lived experience of CP.

This commentary is on the original article by Aravamuthan et al. on pages 723–733 of this issue.

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case of a genetic variant that explains an individual's CP motor disorder, this study shows that some families prefer a diagnostic label of CP plus the genetic variant. Future research with people with lived experience would be required to know whether including non-genetic aetiological risk factors with the CP diagnostic label is preferred, practical, and/or useful.

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Spinal muscular atrophy and the world's most expensive medicines: The price of life

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When our son Rumi was born, there was no sign that he might have a rare disease. But, at 18-months-old, troubling signs of gross motor skill delay surfaced, and after 9 months of testing, he was diagnosed with the degenerative neuromuscular disorder spinal muscular atrophy (SMA). We would wait 3 months to be approved for medical treatment with nusinersen (Spinraza) through a compassionate-use program, as the medicine was not completely approved for public funding in British Columbia.

In retrospect, we see that there were several instances where the lack of knowledge of rare diseases and treatment options limited the health care providers' (HCPs) ability to diagnose SMA. The time we lost in diagnosis still causes deep regret.

It was, therefore, with great interest that I reviewed the article by Carey et al.¹ As a scientific researcher and professor, I have peer-reviewed many papers but never one that spoke to such a personal moment in my family's life, therefore, I will limit my comments to my experiences as an SMA parent.

Rare degenerative neuromuscular diseases often do not have many medical treatment options besides physical

therapy, and what may sometimes seem like a long road of hope-colored palliative care. The only option at the time of our son's diagnosis was 'one of the world's most expensive medicines' (Spinraza). Soon thereafter, a genetic therapy for infants with SMA, onasemnogene abeparvovec (Zolgensma), became a second treatment option and the 'world's most expensive medicine'. As of 2021, risdiplam (Everysdi) became available, and there are several other potential pharmaceutical therapies in the pipeline.

Yet, there remain significant hurdles to accessing medical treatment for SMA, including whether medical treatment is appropriate for a specific type of SMA. For example, when diagnosed and treated early, infantile-onset SMA type 1 (or Werdnig-Hoffmann disease) has better response to Zolgensma than other SMA forms. Further, the relative newness of treatment options and tight timelines for administering treatments for maximum impact require that HCPs reassess diagnostic practices to better incorporate SMA. Another hurdle is that, as of 2022, SMA has not been included in most newborn screening tests. The Canadian CureSMA organization continues to advocate for Canadian