How useful is the diagnosis of ataxic cerebral palsy?

Cerebral palsy (CP) involves a wide range of clinical presentations. Therefore, it is useful to categorize individuals into groups for clinical, research, and service provision purposes. Cerebellar diplegia or congenital cerebellar ataxia was first described in 1903 by Frederick Batten, who differentiated it from hereditary cerebellar ataxia, but the term ‘ataxic CP’ is currently used in various ways. According to the Surveillance of Cerebral Palsy in Europe, individuals with ataxic CP show generalized hypotonia with loss of orderly muscle coordination so that movements are performed with ‘abnormal’ force, rhythm, and accuracy. Other schemes do not require the presence of hypotonia.

Experience (and perhaps my own clinical practice bias) suggests that poor balance and intention tremor are typical, gross motor function is generally less impaired than in other types of bilateral CP, individuals are less likely to develop contractures, and intellectual disability and speech impairment are quite common. However, there is a lack of description of motor and non-motor features in the literature, making the diagnosis problematic. Indeed, early-onset ataxia can signal a host of genetic disorders that are often progressive (thus not CP) with poor outcomes. For example, ataxia-telangiectasia, spinocerebellar ataxia (SCA), and Joubert syndrome can mimic ataxic CP in early life. Developmental impairment of motor coordination as seen in developmental coordination disorder (DCD) is much more prevalent than these conditions, and in many children one cannot distinguish between motor phenotypes of DCD and degenerative ataxias, not to mention ataxic CP. Admittedly, assessing motor disorders in developing children is challenging, and one cannot rely on widely shared experience as in spastic and dyskinetic CP.

Ataxic CP is the least common CP type, accounting for less than 1 in 10 individuals with CP. Moreover, a higher proportion of children see their diagnosis reassigned to non-CP conditions by age 5 years, indicating incorrect initial diagnosis. Thus, more consequentially than in other presentations, it must be stressed that the diagnosis of CP, even when accurate, is essentially descriptive, calling for a comprehensive aetiological work-up.

A genetic origin does not rule out the diagnosis, and it carries counselling implications. Further complexity arises as some responsible genes are better known to cause non-CP disorders. KCNC3 mutations are usually found in SCA13, but some can be associated with an early-onset stable course consistent with ataxic CP. So can SPTBN2 mutations, though they are usually associated with SCA5, or ITPR1 mutations, some of which cause SCA15 and SCA29, and Gillespie syndrome, a congenital hypotonia-ataxia syndrome with aniridia and intellectual disability.

Neuroimaging also fails to show abnormalities much more commonly than in other CP types. Eventual changes are rarely lesional, but rather indicate hypoplasia or other malformations of the cerebellum. Yet, cerebellar malformations do not necessarily manifest themselves with ataxia (or CP). Associations between clinical and neuroimaging features have been far less documented than in spastic and dyskinetic CP. In contrast with those CP types, no causal pathways have been suggested that might improve understanding of the condition and provide at least hypothetical targets for prevention.

In sum, ataxic CP currently appears too heterogeneous and uncertain to be as conceptually useful as other CP types. This may stem from the relatively late development of motor coordination and maturation of the cerebellum, including into the postnatal period. There has also been little intersection between CP, early-onset ataxia, and DCD research, resulting in lack of scrutiny of the overlap between these conditions. The clinical, functional, and pathophysiological picture must be clarified based on studies bringing together phenomenology, neuroimaging, genetics, and epidemiology to improve the definition and understanding of the implications of the diagnosis. Given the low estimated prevalence, such an effort will require worldwide collaboration, ideally involving all major CP registries and groups studying degenerative childhood-onset ataxia, DCD, and typical development.

REFERENCES