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Looking at the Other Side of the Coin: Is the Cerebellum Involved in Sudden Unexpected Death in Mitochondrial Disorders?

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Letter to the Editor

We read with great interest the stimulating article by Schreglmann et al. who showed that isolated dystonia so far classified as idiopathic can be a manifestation of a mitochondrial disorder (MID), thus strengthening the link between cerebellar involvement and movement disorders in MIDs[1]. According to these interesting results and new research perspectives on the subject, the cerebellum can be prominently affected in MIDs. In this context, macro-evolutionary studies of the cerebellum contributed to theories on cerebellar functions and connectivity, providing new insights in the evolutionary pathways that underlie cerebellar functions[2]. Furthermore, recent neuroimaging findings demonstrated that the cerebellum is involved in autonomic regulations[3]. Briefly, it was shown that the cerebellum coordinates vestibular stimuli and blood pressure changes, and that cerebellar injury results in substantially altered timing of responses to pressor challenges in sleep-disordered breathing conditions[3]. Interestingly, several cerebellar pathological findings are reported in the sudden infant death syndrome (SIDS) and it has been proposed that cerebellar atrophy may lead to an inability to recover from apnea or apneusis, or failure to recover from extreme hypotension or arrhythmia, ultimately resulting in sudden death in patients with refractory epilepsy[4]. Following this line of reasoning, sudden death in MIDs is well described in the literature and sudden cardiac death (SCD) is an important cause of death in MIDs necessitating the identification of risk factors for SCD in MIDs by appropriate management[5]. In addition, Eom and colleagues investigated the clinical characteristics of risk factors for death in pediatric MID patients[6]. The authors showed that 29% of their MID patients died suddenly at <6 years of age and that early death was associated with lesions in the thalamus and the number of organs involved in the MID[6]. Considering these results together, it is reasonable to establish a connection between cerebellar abnormalities and increased mortality and even sudden death in MID patients.

Overall, there is a need for further studies and a need to establish first hypotheses about the pathogenesis of these phenomena. While answers to the many of the open questions are not extensively discussed in the literature, caution in the management of MID patients is truly necessary.

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