

Conclusions: We draw attention to the necessary debate between sensible and problematic use of new technologies and the need for longitudinal prevention in schools.

Disclosure of Interest: None Declared

EPV0187

The psychological impact on parents of children with pyridoxine-dependent epilepsy

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Introduction: Pyridoxine-dependent epilepsy (PDE) is a rare autosomal recessive disease usually associated with neonatal seizures that are sensitive to pyridoxine (vitamin B6). This disease can have a significant impact on family functioning, with significant psychological distress in parents. Post-traumatic stress disorder (PTSD), depression, and anxiety are the most common psychiatric outcomes in parents of children with PDE.

Objectives: To investigate the prevalence of significant symptoms of depression, anxiety, and stress in parents of children with PDE.

Methods: The study consisted of a survey of parents accompanying their children diagnosed with PDE. The diagnosis was already confirmed by objectifying a homozygous or a compound heterozygous mutation in the *ALDH7A1* gene in all siblings with heterozygous carrier parents. The Impact of Event Scale-Revised (IES-R) was used to assess parental post-traumatic stress, and the Hospital Anxiety and Depression Scale was used to screen for parental depression and anxiety.

Results: Our study included eight unrelated families with one infant presenting a confirmed PDE disease. The average age of the children with epilepsy was 4.18 years (8 months to 12 years) with equal representation of both sexes.

Half of parents surveyed had depressive symptoms and about two thirds reported anxious symptomatology. These troubles are mainly related to the uncertain prognosis of the disease, even with vitamin B6 supplementation, and the high risk of recurrence in siblings, which led some parents to not have other children. A higher anxiety scores was reported in parents who claimed to have difficulties in providing the necessary vitamin supplements to their affected children on a regular basis. PTSD was diagnosed in three parents: most parents reported difficulties in dealing with stress, specifically in relation to the unpredictability of seizures and the unavailability of medical care for their child, which taxed their financial resources and made it difficult for them to perform their roles effectively.

Besides, being an autosomal recessive transmission disease, the notion of responsibility/guilt was not reported by either parent, and both parents are equally involved in the care of their child.

Conclusions: A significant proportion of children's parents with pyridoxine-dependent epilepsy are suffering from depression, anxiety, and post-traumatic stress. A deeper understanding of the clinical expressions of these troubles could help practitioners to develop prevention and intervention strategies for these parents.

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Psychological impact of motor impairment in tow forms of congenital muscular dystrophy

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Introduction: Congenital muscular dystrophies (CMDs) represent a heterogeneous group of early-onset muscle disorders presenting primarily with hypotonia and delayed motor development. Several genes are known to be responsible for CMDs, including the *LAMA2* gene, involved in merosin-deficient type 1A (MDC1A), and the *FKRP* gene involved in muscular dystrophy-dystroglycanopathy type B5 (MDDGB5). These two forms of CMD are autosomal recessive and are each characterized by the presence of a mutation with a founder effect in South Tunisia. Cognitive development associated with the founder mutation in the *LAMA2* gene (c.8007delT) is often conserved, whereas in the founder mutation of the *FKRP* gene (c.1364 C>A), motor impairment is associated with intellectual disability (ID).

Objectives: To compare the psychological impact of motor impairment in children presenting these two forms of CMD and their families.

Methods: The study consisted of a survey of parents of children with a confirmed diagnosis of MDC1A (5 from 3 unrelated families) or MDDGB5 (3 from 3 unrelated families). The correspondent founder mutation was already identified in the homozygous state by targeted sequencing. Participants' parents completed the Parent Strengths and Difficulties Questionnaire (SDQ), a behavioral screening tool designed for children aged from 2 to 17 years. The SDQ assesses emotional symptoms, behavior problems, hyperactivity, and peer relationships; The SDQ Impact Supplement assesses the impact of all these children's difficulties on their families.

Results: The average age of the children was 4.95±3.92 with two children who were not assessable by the SDQ (age< 2 years). Unlike children with MDC1A, ID has been reported in all children with MDDGB5. The mean SDQ total score for children with MDC1A was 11, whereas the mean score for children with MDDGB5 was 14.875, reflecting greater difficulty for children with MDDGB5. The family impact score was higher in families with children with MDDGB5 than in children with MDC1A (10,5 vs 7), which may be due to the burden of management of the ID associated with the motor impairment. The more pronounced difficulties associated with MDDGB5 are likely to be related to the associated ID. Whereas in MDC1A, the difficulties observed are related to the direct impact of the motor impairment. The presence of cognitive disorders associated with a motor deficit aggravates behavioral adaptation and makes the management of these children more difficult.

Conclusions: In the absence of a comparable study in the literature, the present is conducting future studies on the behavioral profile of children with CMD to obtain a better understanding of their difficulties in everyday life and to develop interventions adapted to their families

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