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Beta-propeller Protein-associated Neurodegeneration

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Editorial

Beta-propeller protein-associated neurodegeneration (BPAN) is a disorder that damages the nervous system and is progressive, which means that it gradually gets worse. Affected individuals develop a build-up of iron in the brain that can be seen with medical imaging. For this reason, BPAN is classified as a type of disorder called neurodegeneration with brain iron accumulation (NBIA), although the iron accumulation may not occur until late in the disease.

Children with BPAN also have intellectual disability, delayed development including significant problems with vocabulary and producing speech (expressive language), and difficulty coordinating movements (ataxia). Ataxia can affect the ability to walk and perform fine motor skills such as using utensils. Affected individuals can have behavioral changes that are often compared to features of a disorder called Rett syndrome. These features include repeated hand wringing or clasping (stereotypic hand movements); teeth grinding (bruxism); sleep disturbances; and problems with communication and social interaction characteristic of autism spectrum disorder. In most X-linked dominant disorders, males experience more severe symptoms than females. While this is not always the case in BPAN, most individuals with the disorder are females, likely because a smaller number of affected males survive until birth.

Almost all cases of BPAN result from new mutations in the gene and occur in people with no history of the disorder in their family. Rarely, an affected person inherits the mutation from a mildly affected mother. Among reported cases, males with BPAN and most females with BPAN have not had children.

Beta-propeller protein-associated neurodegenerationis a form of neurodegeneration with brain iron accumulation, which was initially described as static encephalopathy of childhood with neurodegeneration in adulthood (SENDA). BPAN is very rare, with fewer than 50 cases reported to date. The disease typically starts in early childhood with intellectual impairment and seizures. The disease progresses during adolescence or early adulthood with the emergence of dementia and movement disorders, especially dystonia and Parkinsonism. Rett syndrome-like behaviors, including hand stereotypies, may be observed. The vast majority of cases are female and sporadic, resulting from a de novo mutation in the WD repeat-containing protein 45 (WDR45) gene located at Xp11.23. The gene product, WIPI4, is a 7-bladed beta-propeller protein believed to be involved in autophagy, and there is preliminary evidence suggesting that BPAN could be a tauopathy.

Seizure management, tailored to the individual, may include antiepileptic drugs, ketogenic diet, and/or vagus nerve stimulation. Developmental delays and intellectual disability are managed in infancy and early childhood with early intervention programs and in school-age children with individual education programs. Consultation with a developmental pediatrician to ensure appropriate services is recommended. Discussion about transition plans including financial, vocation/employment, and medical arrangements should begin at age 12 years. Consider alternative means of communication, such as an Augmentative and Alternative Communication program, for individuals who have expressive language difficulties. Motor dysfunction in childhood is managed with physical therapy to maximize mobility and to reduce the risk for later-onset orthopedic complications.

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