



Important Message from Our Medical Advisory Committee

January 11, 2021

From: Alex Kolevzon, MD, PMS MAC chairperson; William Bennett, Jr, MD, MS; Elizabeth Berry-Kravis, MD, PhD; Ann Neumeyer, MD; Curtis Rogers, MD

Re: Therapeutic interventions in Phelan-McDermid syndrome

Dear families affected by Phelan-McDermid syndrome (PMS),

It has come to the attention of the PMS Medical Advisory Committee that some individuals with PMS have challenges sustaining funding for behavioral therapy and related services if children are deemed to not be making adequate gains. For this reason, we wanted to provide the following guidance that we hope will inform discussions of appropriate interventions.

PMS results from deletion or mutation on chromosome 22 involving the *SHANK3* gene and causing significant impairments in behavior, communication, learning, and motor skills. PMS is one of the most common causes of autism spectrum disorder (ASD). Therapeutic interventions include Applied Behavioral Analysis (ABA), speech, physical, and occupational therapies that are critical and beneficial for many individuals.

An intensive behavioral program based on the principles of ABA is medically necessary for the treatment of PMS and/or ASD. Effectiveness of ABA has been clearly established through well-controlled scientific studies in similar populations with developmental disability in order to address the behaviors, communication impairment, and social difficulties that children with PMS exhibit.

In PMS, progress in therapy may take longer to meet goals as compared to other recipients of ABA services and, as with any ABA program, goals will need to be highly individualized. Because of profound challenges in learning and memory, individuals require intensive intervention and constant repetition of skills in order for gains to take hold and to reach their full potential. Importantly, most children with PMS require therapies in addition to what they may be receiving at school. Further, without constant training and repetition, individuals with PMS are at risk for regression and loss of skills. The attached addendum provides some specific clinical considerations.

It is strongly recommended and medically necessary that children with PMS receive ABA with speech, occupational, and physical therapy for a minimum of 40 hours weekly to improve functioning in both the school, home, and community setting.

Please feel free to share this letter with your providers.

ADDENDUM

Challenges with long-term retention of skills in PMS may require flipping the focus of ABA goals so that more time is spent on maintenance sessions. Good ABA programs always have maintenance sessions but emphasize new skills over maintenance - especially during early and middle childhood.

The level of intellectual disability (ID) in PMS suggests using a blended ABA + structured teaching approach that may be more effective than traditional discrete trial or naturalistic behavioral interventions. Most ABA therapists use environmental supports (e.g., picture schedules), but fewer use structured teaching approaches to sequence life skills and adaptive skills (e.g., work systems).

We strongly recommend that individuals with PMS receive intensive, comprehensive behavioral programming based on the principles of Applied Behavior Analysis (ABA) (educational and therapeutic intervention). While this intervention is sometimes mistakenly linked exclusively to ASD diagnoses, this evidence-based intervention is designed for individuals with a range of learning and intellectual disabilities.

The link below provides an overview of ABA and citations highlighting the application of ABA to individuals with a range of intellectual and developmental disabilities.

<https://www.kennedykrieger.org/sites/default/files/library/documents/patient-care/centers-and-programs/own-syndrome-clinic-and-research-center/aba-scientific-support-9-2015.pdf>

In a seminal paper by Baer, Wolf, & Risley (1968) describing ABA, the intervention is described as “the process of systematically applying interventions based upon the principles of learning theory to improve socially significant behaviors to a meaningful degree, and to demonstrate that the interventions employed are responsible for the improvement in behavior. This definition is agnostic to diagnosis and in fact, Baer and colleagues’ original studies of ABA were with participants with moderate to severe intellectual disability and presentations similar to those with PMS.