

The Psychosocial Approach to Phelan-McDermid Syndrome and Perspectives on Intervention

Dr. Yasmine Fatima Daouadji ¹, Pr. Kaoukab Ezzamame Beliardouh ²

¹ University of Oum El Bouaghi, Algeria.

Email: daouadji.yasmine.fatima@univ-oeb.dz

² Director of the Laboratory of Social Problems in Algerian Society,

² University of Oum El Bouaghi, Algeria.

Email; beliardouh.kaoukab@univ-oeb.dz

Received: 27/12/2025 ; Accepted: 13/05/2026 ; Published: 11/06/2026

Abstract

Phelan-McDermid syndrome (PMS) is a rare genetic neurodevelopmental disorder frequently associated with deletions or other genetic alterations involving the 22q13 region of chromosome 22, with a particular role attributed to the SHANK3 gene in explaining some of its developmental and functional characteristics. The syndrome is characterized by substantial variability in cognitive, communicative, behavioral, and adaptive functioning, making it difficult to reduce its presentation to a single clinical profile. Its impact extends beyond medical and developmental manifestations to affect communication, social interaction, family relationships, and participation in everyday life.

This study examines Phelan-McDermid syndrome from a psychosocial perspective, focusing on selected cognitive, emotional, behavioral, and adaptive characteristics, as well as the implications of communication and social interaction difficulties for the individual's daily life. Particular attention is given to the role of the family, quality of life, and social participation within the intervention process. The study adopts a descriptive-analytical approach based on a review of the scientific literature on Phelan-McDermid syndrome, complemented by relevant literature on augmentative and alternative communication, functional behavior assessment, quality of life, and support for individuals with neurodevelopmental disorders.

The review indicates that the substantial variability observed across individuals calls for an individualized approach rather than a stereotyped understanding of the syndrome. Assessment should focus on the person's actual abilities and needs, while intervention should promote functional communication, adaptive and social skills, contextual understanding of behavior, and meaningful family involvement in goal setting. Educational and social environments should likewise be adapted to facilitate participation and autonomy. Accordingly, an appropriate intervention framework should combine individualized assessment with multidisciplinary practice and place both the individual and the family at the center of care, while recognizing the limited number of syndrome-specific intervention studies and the need for further research.

Keywords: Phelan-McDermid syndrome, SHANK3, psychological characteristics, adaptive behavior, communication, social interaction, family, quality of life, psychosocial intervention.

1 - Introduction and Research Problem

Interest in neurodevelopmental disorders has increasingly extended beyond explanations based solely on medical or biological factors. Emerging during the developmental period, these disorders may affect several aspects of an individual's functioning, including cognitive abilities, communication, behavior, adaptive functioning, and social interaction. Understanding these conditions therefore requires consideration not only of their clinical characteristics but also of the individual's living environment, interpersonal relationships, and the demands of everyday life (American Psychiatric Association, 2022). Phelan–McDermid syndrome (PMS) is a rare genetic neurodevelopmental syndrome characterized by considerable variability in its clinical and developmental manifestations. It has historically been associated with deletions or other genetic alterations involving the 22q13 region of chromosome 22, with particular attention to the role of the SHANK3 gene in explaining some aspects of the syndrome's phenotypic presentation (Phelan & McDermid, 2012, p 187). However, the presence of a specific genetic alteration does not necessarily entail uniformity in clinical presentation, as developmental and functional characteristics may differ considerably among affected individuals.

The literature indicates that PMS may be associated with developmental delay, varying degrees of cognitive impairment, limited speech or language abilities, difficulties in certain adaptive skills, and diverse behavioral and social characteristics (Kolevzon et al., 2014, p 784). These manifestations should not be regarded as a fixed set of features that applies equally to every individual. Rather, the nature and extent of the genetic alteration, together with individual and environmental factors, may contribute to shaping each person's developmental and functional profile (Sarasua et al., 2014, p 938, 939).

From a psychosocial perspective, this variability is particularly important. Communication difficulties, for example, cannot be fully understood independently of the responses of people in the individual's environment. Similarly, behavior that appears inappropriate in a particular situation may be associated with the absence of an effective means of expressing a need, refusal, or personal preference. Accordingly, considering the individual within their family, educational, and social contexts provides a more comprehensive understanding of their difficulties and broadens the scope of support beyond the management of observable symptoms.

Against this background, the present study examines the psychological and social dimensions of Phelan–McDermid syndrome, focusing on cognitive and functional development, emotional expression and regulation, behavior and adaptation, and adaptive skills. It then considers communication, social interaction, family relationships, and participation in school and community life. The study also explores key principles that may inform support and intervention, emphasizing the need for individualized and multidisciplinary approaches.

Accordingly, the study addresses the following central research question:

How are the psychological and social characteristics of individuals with Phelan–McDermid syndrome manifested, and what are the prospects for supporting these individuals in light of their specific needs and the demands of their family, educational, and social environments ?

2 - Significance of the Study

This article examines Phelan-McDermid syndrome from a psychosocial perspective, with a focus on its implications for behavioral and emotional adjustment, communication, social interaction, family

relationships, and community participation. It also highlights the importance of adopting a comprehensive, individualized approach to care that takes individual differences into account, promotes autonomy, and enhances quality of life. Particular attention is given to the role of the family and the broader social environment in supporting individuals with the syndrome and addressing their psychosocial needs.

3 - Research Objectives

This article aims to:

- Analyze the main psychological and social characteristics associated with Phelan-McDermid syndrome.
- Identify the key challenges related to adjustment, communication, and social interaction experienced by individuals with the syndrome.
- Highlight the role of the family and the social environment in facilitating adjustment and promoting social participation.
- Explore prospects for psychosocial support and intervention that respond to individual needs and promote autonomy and quality of life.

4 . Overview of Phelan-McDermid Syndrome

Phelan-McDermid syndrome, also referred to as **22q13 deletion syndrome**, is a genetic neurodevelopmental disorder frequently associated with the deletion of part of the long arm of chromosome 22 or with other genetic alterations affecting the same region. The **SHANK3** gene is considered particularly important in explaining several characteristics associated with the syndrome. Nevertheless, alterations in this gene alone do not account for all of the differences observed among individuals with the condition (Phelan & McDermid, 2012, p 187 _ 190).

One of the most prominent features of the syndrome is the considerable variability in developmental and functional characteristics. Cognitive abilities, language and communication, adaptive skills, behavior, and social interaction may vary substantially across individuals. Consequently, a diagnosis of Phelan-McDermid syndrome by itself does not provide sufficient information about a person's level of independence or the type and intensity of support they may require (Kolevzon et al., 2014, p 785).

Clinical and genetic studies have also indicated that the nature and size of the genetic alteration may be associated with variability in clinical manifestations, further emphasizing the importance of individualized assessment in understanding the person's actual needs (Sarasua et al., 2014, p 939 - 941). A major implication for intervention follows from this observation: psychological, educational, and therapeutic planning should not be based solely on the diagnostic label, but rather on the individual's specific strengths, difficulties, and support needs within everyday contexts.

5. Psychological Characteristics of Phelan-McDermid Syndrome

5.1 Cognitive and Functional Development

Phelan-McDermid syndrome may be associated with varying degrees of developmental delay and cognitive difficulties, although levels of functioning differ considerably across individuals. Psychological assessment should therefore extend beyond assigning a numerical score or categorical

classification of cognitive ability. It should also consider how the person applies their abilities in everyday situations (Kolevzon et al., 2014, p786 _ 787).

This issue becomes particularly important when there is a discrepancy between what an individual can understand and what they are able to express verbally. Limited speech should not automatically be interpreted as an absence of comprehension or as a lack of motivation to communicate. An excessive reliance on verbal expression during assessment may consequently underestimate the person's actual abilities.

For this reason, observation, functional assessment, and available communication modalities should complement appropriate psychological testing. The aim is not merely to determine what the individual can do under structured testing conditions, but also to understand what they are capable of doing when appropriate communication tools and adequate environmental support are available.

5.2 Emotional Expression and Regulation

A specific psychiatric disorder should not be regarded as an inherent characteristic of individuals with Phelan-McDermid syndrome, since the diagnosis of a mental disorder requires an independent clinical assessment. Nevertheless, communication limitations may make it more difficult to identify and interpret the person's emotional state, particularly when verbal expression is restricted (Kolevzon et al., 2014, p 787).

In some cases, changes in emotional states may be expressed primarily through behavioral changes. Interpreting such behavior without considering its context may therefore lead to inaccurate conclusions. A sudden change in activity, refusal, or emotional response may be associated with environmental circumstances, communication difficulties, or an inability to express a particular need.

The role of the psychologist, therefore, is not to assign an immediate interpretation to the behavior, but rather to develop an understanding of the individual's emotional experience through the means available to them. Structured observation, information provided by family members, and nonverbal indicators can all contribute to this process. The objective is to understand the meaning of behavior within its context rather than assuming that the absence of verbal expression reflects an absence of emotional experience.

5.3 Behavior and Adaptation

Some individuals may display behaviors that interfere with learning, participation, or social interaction. However, the nature, intensity, and function of such behaviors vary considerably across individuals. Describing a behavior simply as a "behavioral problem" does not, in itself, explain why it occurs or indicate the most appropriate intervention.

Functional behavior assessment provides a framework for examining the relationship between antecedent conditions, the behavior itself, and its consequences. Such an analysis may reveal that a behavior serves a particular function, such as requesting something, attempting to communicate, avoiding a situation, or expressing refusal (O'Neill et al., 2015, p 58).

From this perspective, intervention becomes more meaningful when it does not merely attempt to suppress the observable behavior but instead identifies an alternative response that allows the individual to achieve the same purpose in a more adaptive way. When behavior functions as a means of communication, providing an effective alternative communication strategy may therefore constitute a central component of intervention.

5.4 Adaptive Skills and Independence

Adaptive skills are an important component of assessment in individuals with neurodevelopmental disorders because they reflect what a person is able to do in everyday life, including communication, self-care, social functioning, and participation in activities.

In Phelan-McDermid syndrome, difficulties in adaptive functioning may vary considerably. Consequently, functional skills deserve a central place in intervention planning (Kolevzon et al., 2014, p 788). Their importance extends beyond their status as training objectives; they are directly related to increasing independence and reducing unnecessary dependence on others.

Intervention goals should therefore be translated into meaningful everyday skills, such as making choices, requesting assistance, participating in activities, expressing acceptance or refusal, and performing tasks that are appropriate to the individual's developmental level and abilities.

6. Social Characteristics

6.1 Communication and Social Interaction

Communication is a central component of psychological and social life, and it is particularly important in Phelan-McDermid syndrome given the possibility of limited spoken language in a number of individuals (Kolevzon et al., 2014, p 789).

An important misconception to avoid is equating limited speech with a lack of desire to communicate. An individual may have clear needs, interests, preferences, and social intentions while lacking an adequate verbal means of expressing them. In such circumstances, augmentative and alternative communication (AAC) may provide an important means of expressing choices, needs, and emotions while supporting participation and autonomy (Beukelman & Light, 2020, p 47).

AAC should not be viewed merely as a technological substitute for human communication. Rather, it can provide the individual with a more effective means of entering into and sustaining meaningful interaction with others. When individuals have a communication system that others understand and that they can use consistently, their opportunities to participate in everyday situations may increase substantially.

6.2 Peer Relationships

Communication and adaptive difficulties may make it more challenging to establish and maintain social relationships, particularly in environments that rely heavily on verbal communication. However, difficulties with social initiation or turn-taking should not automatically be interpreted as a lack of interest in others.

Individuals may instead require direct and practical support in skills such as initiating interaction, waiting for their turn, requesting assistance, and participating in shared activities. Such learning is likely to be more meaningful when it occurs in natural settings, since the ultimate objective is not simply to demonstrate a skill during a therapy session but to use it in everyday life.

Group activities, guided play, and naturally occurring school and community situations may therefore provide valuable opportunities for developing social interaction, provided that activities are adapted to the individual's abilities and interests and that social participation is not reduced to a rigid set of prescribed behaviors.

6.3 Family Relationships

The family occupies a central position in the life of a child or adult with Phelan-McDermid syndrome. Family members are often the people most continuously involved in everyday care and are frequently best positioned to recognize subtle changes in communication, behavior, and adaptation.

The rarity of the syndrome and the diversity of associated needs may also create an ongoing need for specialized information and services, making the family an essential partner in intervention (Kolevzon et al., 2014, p 789).

The family should therefore not be reduced to the role of implementing professionals' instructions. Their everyday knowledge of the individual is an important source of information, and their participation in defining goals and evaluating outcomes can make intervention more closely connected to real-life circumstances. Recognizing the family as a partner also contributes to a more balanced and collaborative professional relationship.

6.4 School and Community Participation

Social participation involves more than simply being physically present at school or in a public setting. It includes opportunities to attend, interact, make choices, and benefit from educational, recreational, and community activities.

The functional approach to disability emphasizes that functioning and participation emerge from the interaction between individual characteristics and environmental factors. Consequently, training the individual alone may not be sufficient to improve participation; modifying the environment and providing appropriate support may be equally important components of intervention (World Health Organization, 2001, p 43).

From this perspective, adapting educational environments, providing appropriate communication systems, modifying activities, and creating opportunities for interaction with peers can all contribute to making participation more meaningful and sustainable.

7. Communication and Psychosocial Adaptation

Communication cannot easily be separated from psychological and social adaptation, because the ability to express needs, choices, refusal, and emotions is closely linked to an individual's capacity to participate in everyday life.

AAC becomes particularly relevant when spoken language is insufficient. It may provide individuals with a more effective means of expressing themselves and allow greater involvement in decision-making and everyday activities (Beukelman & Light, 2020, p 49).

Research on early intervention and AAC also emphasizes the importance of providing appropriate communication supports when they are needed rather than waiting for spoken language to develop before introducing alternative means of communication (Ronski et al., 2015, p 182 - 193).

From a psychosocial perspective, communication support should therefore not be regarded as an isolated speech-language intervention. Access to an effective communication system may alter the individual's relationship with the surrounding environment, reduce dependence on others to interpret their needs, and create broader opportunities for participation and autonomy.

8. Behavioral and Adaptive Challenges

Certain behaviors may affect everyday functioning and family or school relationships. However, intervention is generally more meaningful when it begins by examining the function of the behavior rather than focusing exclusively on preventing or suppressing it.

Behavior may represent an attempt to communicate, obtain something, avoid a situation, express refusal, or convey a particular need. Identifying its function can therefore help professionals select a more appropriate intervention and teach an alternative response that allows the individual to communicate the same purpose in a more adaptive manner (O'Neill et al., 2015, p 60).

Environmental factors may also contribute to the emergence or persistence of certain behaviors. Instructions may be unclear, task demands may exceed the individual's abilities, or an adequate communication system may not be available. Intervention should therefore not focus exclusively on the individual; it may also require environmental modifications and training for caregivers, teachers, and other people involved in the person's daily life.

9. Family and Adaptation to the Syndrome

Caring for a child with multiple developmental needs involves a range of everyday responsibilities, including care, follow-up, communication, and coordination across different services. In rare disorders, the need for specialized information and appropriate services may become particularly pronounced, making family support an essential component of intervention (Kolevzon et al., 2014. p 790).

Family counseling is valuable because it can help parents understand the nature of the syndrome, develop a realistic understanding of their child's individual characteristics, acquire appropriate communication and behavioral strategies, and translate broad intervention objectives into practices that can be implemented in everyday life.

Within this framework, collaboration between families and professionals is likely to be most effective when it is based on partnership. The family contributes more than information, and the professional does not hold an exclusive position of expertise. Both parties contribute to a shared goal: improving the individual's quality of life and supporting communication, adaptation, and participation.

10. Quality of Life and Independence

Quality of life extends beyond the absence of illness or the reduction of symptoms. It encompasses relationships, participation, autonomy, choice, safety, and access to services and resources. The concept has become an important framework in the field of intellectual and developmental disabilities because it shifts attention from what the person lacks toward the kind of life they experience and the opportunities available to them (Schalock & Verdugo, 2002).

Improving quality of life is also associated with providing support that enables individuals to pursue personally meaningful goals while strengthening participation, autonomy, and social relationships (Schalock et al., 2016, p 178 – 180).

For individuals with Phelan-McDermid syndrome, indicators of progress may be reflected in abilities that appear simple but have considerable significance in everyday life, such as expressing a preference, requesting assistance, participating in an activity, communicating with others, and performing daily tasks with greater independence.

Accordingly, the success of intervention should not be judged solely by reductions in challenging behavior or improvement on a particular test. It should also be evaluated in terms of the person's ability to communicate, participate, exercise choice, and make decisions concerning aspects of their own life.

11. Perspectives on Psychosocial Intervention

11.1 Individualized Psychological Assessment

Effective intervention requires a clear starting point based on a multidimensional assessment of cognitive, communicative, behavioral, adaptive, and social functioning, together with an understanding of the family and educational environments in which the individual lives.

Genetic diagnosis alone is insufficient to determine support needs because of the substantial variability between individuals and because needs may change with age and developmental stage (Kolevzon et al., 2014, p 790 - 791). Assessment should therefore be understood as an ongoing process rather than a one-time procedure conducted at the beginning of intervention.

11.2 Functional Behavioral Intervention

When behaviors interfere with learning or participation, it is useful to examine the circumstances preceding the behavior and the consequences that follow it in order to develop a clearer understanding of its function.

This approach supports intervention aimed at teaching functional alternatives rather than simply suppressing observable behavior. Such an approach is particularly relevant when behavior is associated with communication difficulties or an inability to express a specific need (O'Neill et al., 2015, p 62).

11.3 Support for Augmentative and Alternative Communication

AAC represents an important intervention domain when spoken language is insufficient to meet an individual's communication needs. The appropriate system varies from one person to another according to their abilities, needs, environment, and intervention goals (Beukelman & Light, 2020, p 50).

The effectiveness of a communication system depends not only on selecting an appropriate modality but also on the ability of the people around the individual to understand and use it consistently. Training family members, teachers, and professionals is therefore an important component of successful AAC intervention.

11.4 Development of Social and Adaptive Skills

Intervention programs should focus on skills that have direct relevance to everyday life, including social initiation, requesting assistance, making choices, turn-taking, and participation in activities.

Training in natural environments can also increase the likelihood that skills will generalize from therapy sessions to everyday situations. The true indicator of skill acquisition is not merely whether an individual can perform the skill in front of a professional, but whether they can use it when it becomes necessary in real life.

11.5 Family Counseling and Support

Family counseling constitutes an important component of intervention and may include education about the syndrome, communication support, behavioral understanding, management of everyday situations, and assistance in developing realistic expectations regarding the child's abilities.

Involving parents in goal setting also helps ensure that intervention remains connected to everyday life, since newly acquired skills require repeated opportunities for use beyond specialized intervention sessions.

11.6 Promoting Participation and Inclusion

Inclusion is not achieved simply by placing a person in a school or community setting. It requires genuine opportunities for interaction, participation, and choice. Accordingly, adapting educational and social environments, providing appropriate communication supports, modifying activities, and promoting opportunities for peer interaction should be regarded as interconnected components of intervention (World Health Organization, 2001, p 52).

The objective, therefore, is to move beyond merely "allowing participation" toward creating the conditions in which meaningful participation becomes possible.

11.7 Multidisciplinary Practice and the Role of the Psychologist

The range of needs that may accompany Phelan-McDermid syndrome makes interdisciplinary collaboration particularly important. Depending on the individual's needs, the team may include physicians, psychologists, speech-language therapists, special education professionals, and other specialists (Kolevzon et al., 2014, p 791).

The psychologist's role includes assessing psychological, behavioral, and adaptive functioning; identifying strengths and areas of difficulty; examining factors that influence adaptation; and contributing to the development of an individualized intervention plan.

The psychologist may also support the family, examine behaviors that interfere with participation, monitor school and social adjustment, and contribute to the evaluation of intervention effectiveness. In this context, psychological practice does not occur in isolation from other disciplines; rather, the psychologist contributes to a shared framework that places the needs of the individual and family at the center of intervention.

12. Conclusion

Phelan-McDermid syndrome represents a complex and highly variable genetic neurodevelopmental condition. Accordingly, an understanding based solely on its genetic and medical characteristics does not provide a complete account of the lives of individuals affected by the syndrome. Its consequences may extend to communication, behavior, adaptation, social interaction, family relationships, and participation across different areas of life.

The literature reviewed suggests that communication difficulties may play an important role in psychosocial adaptation. This is not because limited speech necessarily reflects a reduced desire to communicate, but because the absence of an effective means of expression may make the individual's needs less accessible to those around them. Similarly, behaviors that appear challenging or inappropriate require functional interpretation that takes into account the context in which they occur and the purpose they may serve.

Effective intervention should therefore not be based solely on an attempt to change the individual. It should also address the relationship between the person's characteristics and the demands of the surrounding environment. This includes individualized assessment, support for functional

communication, development of adaptive and social skills, functional understanding of behavior, family involvement, adaptation of educational and social environments, and multidisciplinary collaboration.

Attention to quality of life adds another important dimension to intervention. The ultimate goal is not simply to reduce difficulties, but to support the individual in becoming more able to communicate, make choices, participate, exercise autonomy, and establish meaningful relationships within the limits and possibilities of their individual profile.

Thus, a psychosocial approach to Phelan-McDermid syndrome makes it possible to move beyond viewing the condition merely as a medical diagnosis and toward understanding the person as an individual with abilities, needs, relationships, rights, and opportunities for participation. This shift in perspective provides an important foundation for intervention that is more humane, realistic, and closely connected to the lived experiences of the individual and their family.

References

- 1 /American Psychiatric Association. (2022). *Diagnostic and statistical manual of mental disorders* (5th ed., text rev.; DSM-5-TR). American Psychiatric Association Publishing.
- 2 /Beukelman, D. R., & Light, J. C. (2020). *Augmentative & alternative communication: Supporting children and adults with complex communication needs* (5th ed.). Paul H. Brookes Publishing Co.
- 3 /Kolevzon, A., Angarita, B., Bush, L., Wang, A. T., Frank, Y., Yang, A., Rapaport, R., Saland, J., Srivastava, S., et al. (2014). Phelan-McDermid syndrome: A review of the literature and clinical guidelines. *Journal of Child Neurology*, 29(6), 783–791. <https://doi.org/10.1177/0883073813500507>
- 4 /O'Neill, R. E., Albin, R. W., Storey, K., Horner, R. H., & Sprague, J. R. (2015). *Functional assessment and program development for problem behavior: A practical handbook* (3rd ed.). Cengage Learning.
- 5 /Phelan, K., & McDermid, H. E. (2012). The 22q13 deletion syndrome (Phelan-McDermid syndrome). *Molecular Syndromology*, 2(3–5), 186–201. <https://doi.org/10.1159/000334260>
- 6 /Ronski, M., Sevcik, R. A., Barton-Hulsey, A., & Whitmore, A. S. (2015). Early intervention and AAC: What a difference 30 years makes. *Augmentative and Alternative Communication*, 31(3), 181–202. <https://doi.org/10.3109/07434618.2015.1064163>
- 7 /Sarasua, S. M., Dwivedi, A., Boccuto, L., Chen, C. F., Sharp, J. L., Rollins, J. D., Collins, J. S., Rogers, R. C., Phelan, K., & others. (2014). Clinical and genomic evaluation of 201 patients with Phelan-McDermid syndrome. *Genetics in Medicine*, 16, 937–944.
- 8 /Schalock, R. L., & Verdugo, M. A. (2002). *Handbook on quality of life for human service practitioners*. American Association on Mental Retardation.
- 9 /Schalock, R. L., Verdugo, M. A., Gómez, L. E., & Reinders, H. J. S. (2016). Enhancing quality of life for people with intellectual disability: From theory to practice. *Journal of Applied Research in Intellectual Disabilities*, 29(3), 179–195.
- 10 /World Health Organization. (2001). *International classification of functioning, disability and health (ICF)*. World Health Organization