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EARLY-ONSET FAHR SYNDROME IN SIBLINGS: NEURODEVELOPMENTAL DISSOCIATION BETWEEN COGNITIVE IMPAIRMENT AND BEHAVIORAL DYSREGULATION

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ABSTRACT

Fahr syndrome (FS), also known as primary familial brain calcification (PFBC), is a rare and heterogeneous neurodegenerative disorder characterized by bilateral intracranial calcifications, most commonly affecting the basal nuclei. Although the condition is typically associated with movement disorders in adults, increasing attention has been given to early-onset cases, particularly in children, where the clinical presentation often differs from the classical phenotype.

This study examines cognitive, emotional, and behavioral functioning in two siblings diagnosed with early-onset Fahr syndrome. By combining neurological assessment, developmental psychopathology, and neuropsychological evaluation, the analysis provides insight into variability observed within the same family.

The results indicate marked differences between the siblings despite a shared genetic and environmental background. Notably, cognitive deficits did not consistently correspond with emotional or behavioral disturbances. This suggests that the extent of calcifications alone may not fully explain functional outcomes. Instead, factors such as the timing of neurodevelopmental disruption, environmental influences, and possible genetic modifiers may play an important role.

These findings highlight the importance of early neuropsychological assessment and support a multidisciplinary approach to diagnosis and care. They also point to the possibility that the clinical spectrum may extend beyond classical PFBC, underlining the need for broader genetic and metabolic evaluation.

KEYWORDS

Fahr Syndrome, Basal Nuclei Calcification, Pediatric Neuropsychology, Cognitive Impairment, Psychopathology, Rare Neurological Diseases

CITATION

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Introduction

Fahr syndrome represents a diagnostically and pathophysiologically complex neurological entity, situated at the intersection of neurodegeneration, metabolic dysregulation, and genetic vulnerability[1]. The hallmark feature of the disorder—bilateral, symmetrical calcifications within subcortical brain structures—has been well documented since its initial description by Karl Theodor Fahr in 1930[2]. However, despite nearly a century of investigation, the precise mechanisms underlying these calcifications remain only partially elucidated.

From a contemporary perspective, Fahr syndrome is best conceptualized not as a singular disease entity but as a spectrum disorder encompassing both idiopathic and secondary forms of intracranial calcification. The idiopathic variant, now commonly termed primary familial brain calcification (PFBC), is genetically mediated and most often inherited in an autosomal dominant manner. Mutations in genes such as *SLC20A2*, *PDGFB*, *PDGFRB*, and *XPR1* implicate disturbances in phosphate transport, vascular integrity, and blood-brain barrier function [3-6].

Importantly, these molecular pathways suggest that calcification is not merely a passive degenerative process but rather the result of dysregulated mineral homeostasis within the neurovascular unit. This has profound implications for understanding the variability in clinical expression.

While adult-onset FS typically presents with movement disorders, psychiatric symptoms, and cognitive decline, pediatric cases remain poorly characterized. Early-onset presentations often deviate from the classical phenotype, instead manifesting as:

- developmental delays,
- language impairments,
- behavioral dysregulation,
- intellectual disability.

This divergence underscores the importance of adopting a developmental neuropsychology framework when analyzing pediatric FS. Brain calcifications occurring during critical periods of neurodevelopment may disrupt network maturation, synaptic pruning, and functional specialization, leading to atypical developmental trajectories rather than simple neurodegeneration [7-9].

The current study contributes to this emerging field by providing an in-depth, comparative analysis of two siblings, thereby offering a unique opportunity to explore intra-familial variability under shared genetic and environmental conditions.

Materials and Methods

The present research adopts a qualitative case study design, which is particularly appropriate for rare disorders characterized by low prevalence and high phenotypic variability. While such a design limits statistical generalizability, it enables rich, context-sensitive analysis and hypothesis generation.

Multimodal Data Integration

A key strength of the study lies in its multimodal data collection strategy, integrating:

- **Neurological data** – motor signs, coordination, neuroimaging
- **Psychiatric evaluation** – emotional regulation, behavioral patterns
- **Neuropsychological testing** – cognitive domains
- **Developmental history** – milestones, early functioning
- **Family context** – caregiving environment, relational dynamics

This integrative approach aligns with contemporary models of neurodevelopmental disorders, which emphasize the interaction between biological and environmental factors.

Neuropsychological Framework

The neuropsychological assessment was guided by a domain-specific approach, focusing on:

- executive functions (inhibition, flexibility, planning),
- memory systems (working memory, episodic memory),
- language processing,
- visuospatial abilities,
- attentional control.

Such a framework allows for the identification of specific cognitive profiles rather than global impairment alone.

Results

Case 1

The first case concerns a 9-year-old girl with a complex and internally varied neurodevelopmental profile. Her overall level of intellectual functioning remained relatively preserved; however, clear difficulties were observed in the domains of emotional regulation and behavioral control. This discrepancy represents an important aspect of her clinical picture and may help to better understand the neurofunctional consequences associated with early-onset Fahr syndrome. Computed tomography (CT) imaging demonstrated irregular areas measuring 21 × 7 mm on the right side and 14 × 5 mm on the left, located within the subcortical nuclei (thalamus and globus pallidus), consistent with calcifications [Fig.1].

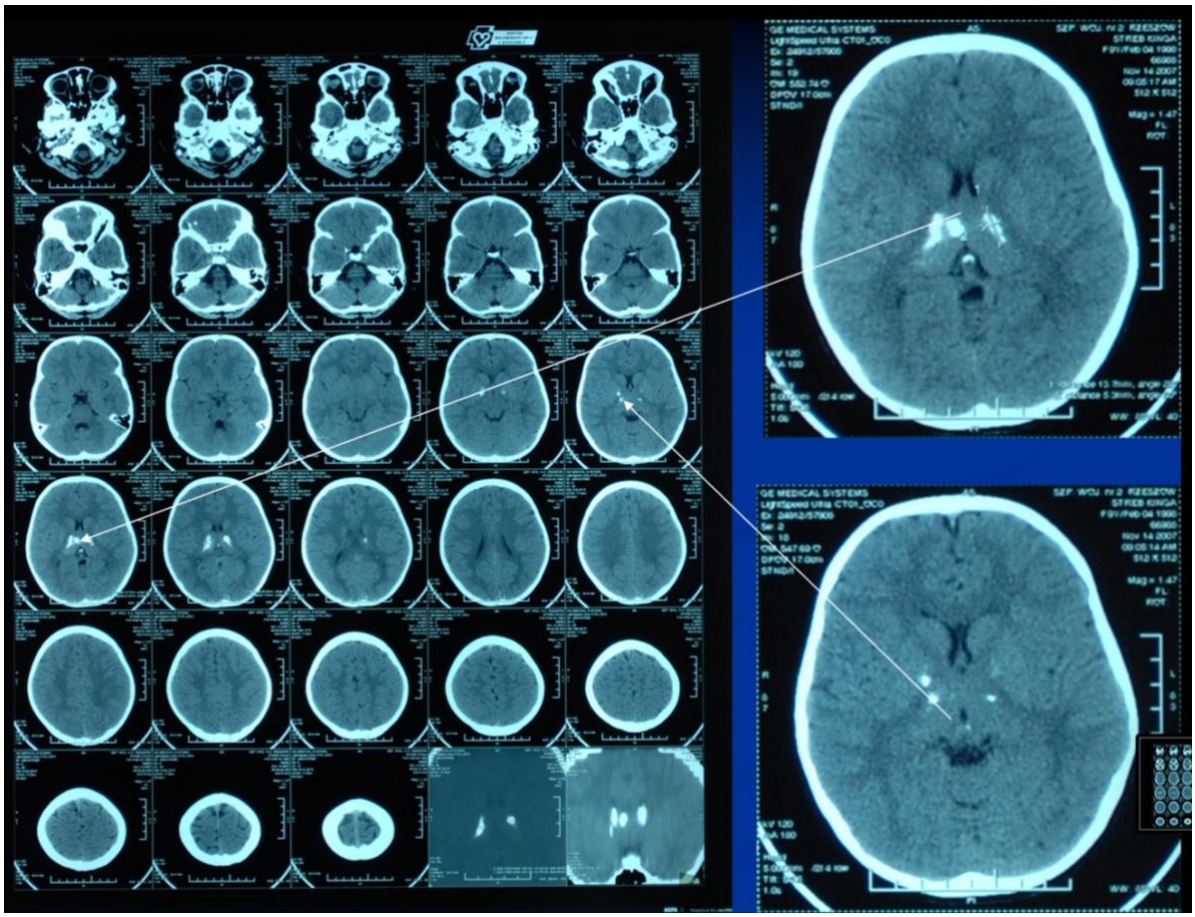

Fig. 1. *A CT scan of the 9-year-old patient revealed bilateral calcifications in the thalamic region*

Developmental milestones were reported as being achieved within expected timeframes during infancy and early childhood, with no evident delays in gross motor development. Despite this, more subtle irregularities became noticeable during the preschool years, particularly in areas related to self-regulation and sustained attention. Caregivers described the child as increasingly prone to irritability, with reduced tolerance to frustration and episodes of emotional outbursts that often appeared disproportionate to the triggering situations. Although language development progressed within normative limits in terms of vocabulary acquisition and grammatical structure, some difficulties were observed in the pragmatic aspects of communication. These included problems with maintaining conversational turn-taking, reduced sensitivity to social cues, and challenges in appropriately modulating tone and affect in social interactions.

Neuropsychological assessment indicated borderline-to-average intellectual functioning, although the cognitive profile was clearly uneven across domains. Executive functions emerged as the most affected area. In particular, the patient demonstrated significant difficulties with inhibitory control, cognitive flexibility, and planning abilities. She had noticeable problems suppressing impulsive responses, both verbal and behavioral, and showed limited adaptability when task demands changed. In addition, impairments in planning and organization were evident, especially in tasks requiring sequencing of actions or anticipating potential outcomes. Overall, this pattern of difficulties may be indicative of dysfunction within frontostriatal networks, particularly involving the dorsolateral prefrontal cortex and its connections with subcortical structures.

Attention and working memory were also clearly affected. The patient's ability to sustain attention was inconsistent, with frequent lapses and a pronounced vulnerability to distraction from environmental stimuli. Working memory capacity appeared limited, particularly in situations that required the simultaneous storage and processing of information or the ongoing updating of mental representations. Altogether, these findings point toward disruption within prefrontal–parietal networks, possibly further influenced by subcortical calcifications that interfere with efficient integration of neural signals.

In contrast to these difficulties, basic language abilities remained relatively well preserved. The patient demonstrated an adequate range of vocabulary and appropriate grammatical structures; however, inefficiencies

became apparent in more complex aspects of language functioning. These included reduced verbal fluency, difficulties in organizing narratives, and challenges in pragmatic communication. Such a pattern suggests that while core linguistic systems remain intact, language functions relying on executive control are compromised. At the same time, visuospatial and perceptual abilities appeared comparatively preserved, with satisfactory performance in tasks involving visual construction, spatial processing, and pattern recognition. This relative strength further supports the domain-specific nature of the observed cognitive profile.

The most prominent and clinically significant aspect of the patient's presentation concerns severe emotional and behavioral dysregulation, which markedly interferes with functioning both at home and in the school environment. The behavioral pattern is characterized by impulsivity, reduced inhibitory control, oppositional tendencies, and rapid escalation of emotional responses. The patient frequently shows difficulty complying with external demands, particularly in interactions involving authority figures, and exhibits a low tolerance for frustration. Emotional reactions tend to be intense, insufficiently modulated, and often disproportionate to the situational context, reflecting deficits in both emotional awareness and regulatory capacity. In addition, recovery following emotional arousal is prolonged, which contributes to ongoing interpersonal conflict and reduced overall functioning.

Neuroimaging findings revealed bilateral calcifications primarily located in the globus pallidus and thalamus. These regions form essential components of corticostriatal circuits that play a key role in behavioral inhibition, emotional regulation, and the selection of actions. Disruption within these pathways offers a plausible neurobiological explanation for the observed disinhibition and affective instability. In particular, the globus pallidus has been linked to motivational and affective processes, suggesting that calcifications in this structure may influence reward processing and the assignment of emotional significance.

From a diagnostic standpoint, the clinical picture may initially resemble several primary psychiatric or neurodevelopmental disorders, such as attention-deficit/hyperactivity disorder, oppositional defiant disorder, or disruptive mood dysregulation disorder. However, the presence of structural abnormalities in the brain, together with an atypical developmental course and a clear dissociation between cognitive functioning and behavioral dysregulation, distinguishes this case from primary psychiatric conditions. These characteristics emphasize the importance of considering underlying neurological causes, particularly in presentations that are atypical or resistant to standard treatment approaches.

The severity and manifestation of symptoms appear to be influenced by environmental factors. Settings that provide clear structure, consistent expectations, and predictable routines tend to reduce the level of behavioral dysregulation. In contrast, unstructured environments, increased cognitive demands, and emotionally charged situations often intensify symptom expression. Caregiver responses also play a significant role. Inconsistent discipline or heightened emotional reactivity within the caregiving environment may exacerbate dysregulation, whereas stable behavioral frameworks, emotion-focused guidance, and structured reinforcement strategies can help to mitigate symptom severity.

From a functional perspective, the patient's difficulties translate into notable impairments in academic performance, particularly in tasks requiring sustained attention, planning, and organization. Social functioning is also affected, as impulsivity and emotional instability contribute to peer conflicts and reduced acceptance within peer groups. Within the family context, behavioral challenges are associated with increased caregiver stress and strain in relationships. Nevertheless, the relatively preserved level of intellectual functioning suggests that the prognosis may be favorable if appropriate and targeted interventions are implemented.

Clinical management should therefore adopt a multimodal approach that integrates neuropsychological, behavioral, and educational strategies. Interventions aimed at improving executive functions—particularly inhibition, cognitive flexibility, and planning—are of central importance. In addition, therapeutic approaches focusing on emotional regulation, including adapted cognitive-behavioral techniques, may support the development of more effective coping strategies. Behavioral interventions, such as consistent reinforcement systems and structured parent training, are also essential. Educational support should include individualized learning plans and external organizational aids to facilitate academic functioning. Given the neurological basis of the disorder, regular neurological follow-up remains necessary in order to monitor potential progression and the emergence of additional symptoms.

In summary, this case represents a distinct and informative presentation of early-onset Fahr syndrome, characterized by relatively preserved general cognitive abilities alongside pronounced deficits in executive functioning and emotional regulation. The findings emphasize the role of network-specific dysfunction, particularly within frontostriatal circuits, in shaping behavioral outcomes. At the same time, they challenge the assumption that global cognitive impairment is the primary determinant of functional disability, instead highlighting the importance of regulatory mechanisms in neurodevelopmental disorders.

Case 2

The second case concerns a 7-year-old boy whose neurodevelopmental profile is marked by globally reduced cognitive functioning consistent with moderate intellectual disability, accompanied by clear delays in language development. The clinical presentation reflects a broad disruption of cognitive processes affecting reasoning, communication, and adaptive functioning. At the same time, emotional and social functioning appear relatively preserved, offering an important perspective on the organization and relative independence of neurodevelopmental domains.

Computed tomography (CT) imaging revealed bilateral calcifications measuring 11 × 6 mm in the thalamic region. In addition, the patient was diagnosed with thrombocytopenia and psoriasis, suggesting possible involvement of systemic processes beyond the central nervous system [Fig.2].

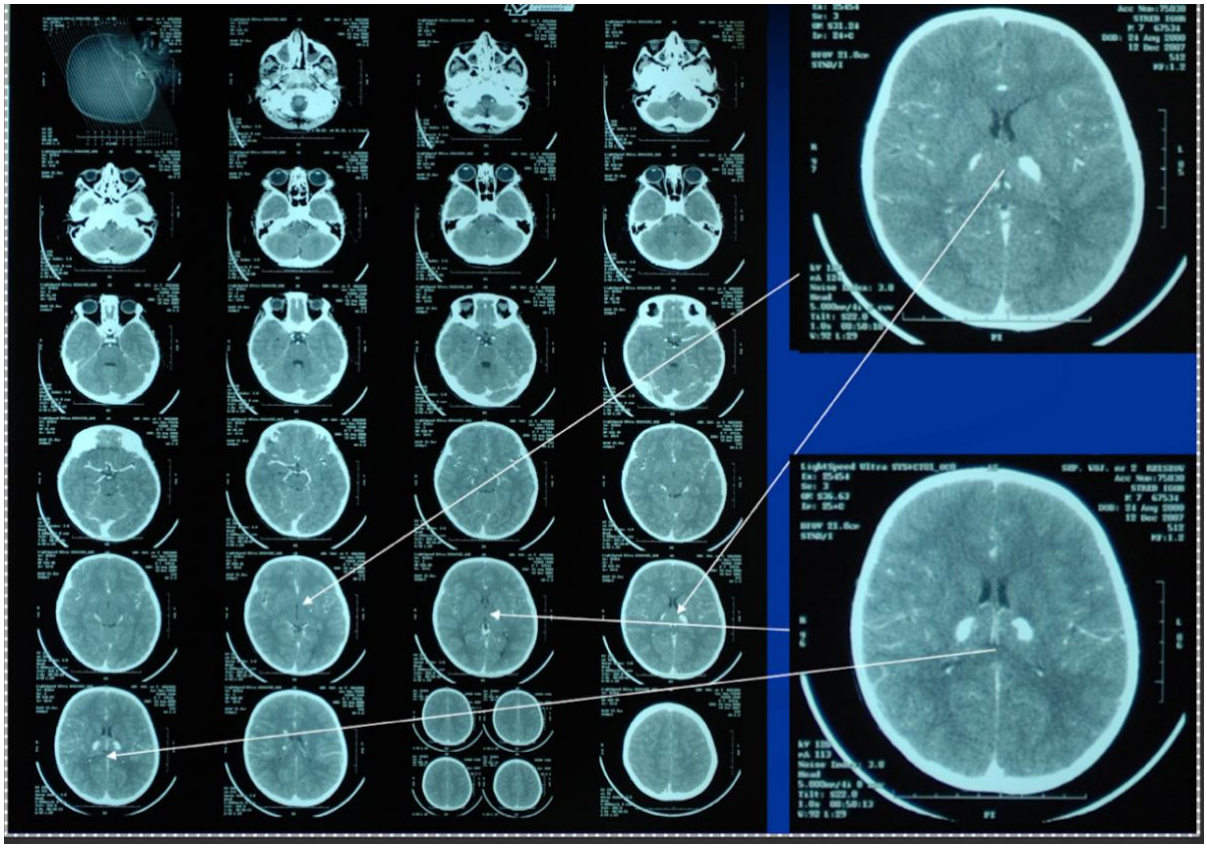

Fig. 2. *A CT scan of the patient's 7-year-old brother revealed bilateral calcifications measuring 11 × 6 mm in the thalamic region*

Developmental history indicates early deviations from expected milestones, particularly in the domain of speech and language. Caregivers reported delayed onset of first words and slower progression toward phrase speech. The child produced limited verbal output and demonstrated reduced complexity in communication. Although nonverbal behaviors such as gestures and eye contact were present, they were not sufficient to compensate for expressive language deficits. Over time, these language difficulties became increasingly pronounced and began to interfere with both learning and social participation.

Language impairment represents a central component of the clinical profile. Expressive language is characterized by a restricted vocabulary, simplified grammatical structures, and a reliance on concrete forms of expression. The child has difficulty producing longer utterances and organizing coherent narratives. Receptive language is also affected, particularly in the comprehension of more complex instructions and abstract content, often necessitating repetition and simplified input.

Neuropsychological evaluation indicates a globally reduced level of intellectual functioning, with deficits observed across major cognitive domains. Abstract reasoning is limited, and the child experiences difficulty identifying patterns, forming conceptual relationships, and generalizing knowledge across contexts.

Problem-solving tends to be concrete and inefficient, frequently relying on repetition rather than flexible strategies.

Deficits in working memory are also evident, particularly in tasks requiring both retention and manipulation of information. Processing speed is reduced, contributing to slower task execution and increased cognitive effort. Difficulties with attention appear to be primarily related to limited cognitive capacity rather than impulsivity, as improved engagement is observed in structured and simplified conditions.

Despite these cognitive challenges, emotional and social functioning remain relatively intact. The child demonstrates an appropriate level of social interest, actively engages in interactions, and responds adequately to basic social cues. Emotional expression is generally appropriate to context, and the child is capable of participating in shared activities and maintaining interpersonal contact.

From a behavioral perspective, the child is typically cooperative, compliant, and responsive to guidance. There is no clear pattern of impulsivity or significant emotional dysregulation, and emotional reactions are generally proportionate to situational demands. This profile suggests relatively preserved emotional regulation despite broader cognitive limitations.

The observed dissociation between cognitive impairment and socio-emotional functioning suggests that these domains may depend on partially distinct neural systems. While higher-order cognitive processes are broadly affected, neural circuits underlying emotional processing and social interaction may remain relatively preserved. In addition, temperamental factors, such as a calm and adaptable disposition, may further contribute to this pattern.

Adaptive functioning is nevertheless affected, particularly in tasks that require planning, independence, and organization. The child benefits from structured environments, predictable routines, and consistent external support. In the educational setting, significant learning difficulties are evident, especially in tasks involving language and abstract reasoning, making individualized instruction, visual supports, and repetition necessary.

Importantly, the child's preserved social engagement represents a key strength that can be effectively utilized in intervention. His cooperative behavior facilitates participation in therapeutic and educational activities.

Clinical management should focus on speech and language therapy, structured cognitive support, and educational accommodations tailored to the child's needs. Interventions aimed at improving adaptive skills, along with active involvement of caregivers, are essential. Continuous monitoring is recommended in order to track developmental progress and adjust intervention strategies as needed.

In summary, this case illustrates a neurodevelopmental profile characterized by moderate intellectual disability and language delay, accompanied by relatively preserved emotional and social functioning. The dissociation observed across domains highlights the complexity of brain–behavior relationships and underscores the importance of individualized, multidimensional approaches to intervention.

Comparative Analysis

The divergence between the siblings represents a critical finding of the study. Despite shared genetic background and similar neuroimaging findings, their functional outcomes differ markedly.

This intra-familial variability can be conceptualized through several frameworks:

1. Neurodevelopmental Timing Hypothesis

The timing of calcification onset relative to critical developmental windows may influence which neural systems are most affected.

2. Network Vulnerability Model

Different individuals may exhibit varying susceptibility across neural networks, leading to distinct phenotypic profiles.

3. Environmental Modulation

Subtle differences in environmental experiences, caregiver interactions, and educational support may amplify or mitigate symptom expression.

Discussion

The findings of the present study support the view that early-onset Fahr syndrome (FS) can be understood not only as a neurodegenerative condition, but also as a disorder with a substantial neurodevelopmental component. While FS has traditionally been associated with progressive neurological deterioration in adulthood, observations from pediatric cases suggest a different perspective. When calcifications emerge during critical periods of brain maturation, they may interfere with developmental processes, leading to atypical developmental trajectories rather than exclusively degenerative changes.

The timing of these pathological processes appears to play a key role in shaping the clinical presentation. Calcifications that develop early in life may disrupt synaptic organization, pruning mechanisms, and the formation of functional neural networks. As a result, connectivity may be altered and overall network efficiency reduced. Such disturbances are more likely to produce domain-specific impairments than uniform deficits, which aligns FS with other neurodevelopmental conditions characterized by heterogeneous cognitive and behavioral profiles.

At the same time, the findings are consistent with a model in which FS involves both developmental disruption and the potential for later progression. In early-onset cases, the disorder may initially manifest through atypical neurodevelopment, while neurodegenerative features may become more evident over time. This perspective helps account for the variability observed across cases and highlights the importance of longitudinal monitoring in understanding the course of the disorder.

An important observation concerns the dissociation between cognitive impairment and emotional or behavioral dysregulation. This challenges simplified models that assume a direct correspondence between structural brain pathology and functional outcome. Instead, it suggests that different domains of functioning may rely on partially independent neural systems. Emotional regulation and behavioral control appear to depend strongly on frontostriatal circuits, whereas global cognitive functioning may be affected in a more diffuse manner.

This dissociation has clear diagnostic implications. Symptoms such as impulsivity, irritability, or oppositional behavior may initially lead to diagnoses within the spectrum of psychiatric or neurodevelopmental disorders, including attention-deficit/hyperactivity disorder, mood disorders, or conduct-related conditions. However, in cases that present atypically or show limited response to standard interventions, the possibility of an underlying neurological condition should be considered. Without appropriate neuroimaging, intracranial calcifications may remain undetected, delaying accurate diagnosis and appropriate management.

The co-occurrence of thrombocytopenia and psoriasis in one of the patients suggests that, in some cases, FS may involve processes extending beyond the central nervous system. This observation raises the possibility of shared underlying mechanisms, such as immune dysregulation or overlapping genetic pathways. Although currently identified PFBC-related genes are primarily associated with phosphate metabolism and vascular integrity, additional biological mechanisms may contribute to more complex or systemic clinical presentations.

These considerations underline the importance of comprehensive diagnostic evaluation. Advanced genetic testing, metabolic assessment, and immunological profiling may improve diagnostic precision and contribute to a more complete understanding of the underlying pathophysiology. A multidimensional diagnostic approach is particularly relevant in cases where clinical presentation extends beyond the classical neurological phenotype.

From a clinical perspective, the findings emphasize the need for a multidisciplinary approach to management. Effective care should integrate expertise from neurology, psychiatry, psychology, and clinical genetics. Given the heterogeneity of presentations, interventions should be tailored to the individual cognitive and behavioral profile of each patient. Early intervention is especially important, as it allows clinicians to make use of neuroplasticity during key developmental periods. Targeted therapeutic strategies addressing cognitive, language, and emotional domains may improve functional outcomes over time.

The role of the family environment should also be taken into account. Providing caregivers with appropriate education, structured guidance, and psychological support can positively influence both the child's functioning and overall family well-being. Interventions that actively involve the family system are likely to be more effective and sustainable in the long term.

Taken together, these observations support the view that FS should be conceptualized as a multidimensional condition involving interacting neurodevelopmental, neuropsychiatric, and potentially systemic components. Further research is needed to clarify the mechanisms underlying phenotypic variability, including the roles of genetic, environmental, and developmental factors.

Limitations

Several limitations should be acknowledged. The small sample size limits generalizability, and findings should be interpreted with caution. The cross-sectional design prevents conclusions about developmental trajectories and long-term outcomes, highlighting the need for longitudinal studies.

The lack of comprehensive genetic, metabolic, and immunological data limits the ability to fully characterize the underlying etiology and potential systemic involvement. Additionally, the qualitative nature of the assessment introduces the possibility of observer bias.

Future research should address these limitations through larger samples, longitudinal designs, and more comprehensive diagnostic evaluations, including genetic and metabolic analyses, to improve understanding and clinical management of early-onset FS.

Conclusions

Early-onset Fahr syndrome represents a complex and heterogeneous neurological condition with significant implications for cognitive, emotional, and social development in children. Its clinical presentation is multidimensional and often difficult to interpret in a straightforward manner, which can complicate the diagnostic process and delay appropriate intervention.

The findings of the present study indicate that clinical expression may vary considerably, even among individuals within the same family. This variability suggests that, in addition to shared genetic factors, environmental influences and individual developmental trajectories contribute to shaping the phenotype of the disorder.

It was also observed that cognitive impairment and behavioral dysregulation do not necessarily occur in parallel. These domains may show relative independence, which highlights the importance of assessing them separately and designing interventions that address specific areas of difficulty rather than relying on a global characterization of functioning.

The study further emphasizes the importance of early and comprehensive neuropsychological assessment. Such evaluation plays a crucial role in identifying both deficits and preserved abilities, thereby supporting the development of targeted and effective intervention strategies that can enhance developmental outcomes.

In addition, the presence of co-occurring conditions suggests that the clinical picture may extend beyond the classical definition of Fahr syndrome. This observation points to the need for a broader and more integrative diagnostic approach, including ongoing monitoring and consideration of systemic involvement.

The course of the disorder may also be dynamic, with changes emerging over time. This highlights the importance of long-term follow-up and the need to adapt therapeutic strategies in response to evolving clinical needs. Attention should also be given to the psychosocial context of the child's functioning, including the impact of symptoms on family relationships, educational experiences, and peer interactions.

Overall, these conclusions contribute to a more nuanced understanding of pediatric Fahr syndrome. They underscore the importance of continued interdisciplinary research that integrates perspectives from neurology, psychiatry, neuropsychology, and clinical genetics in order to improve both diagnostic accuracy and the effectiveness of clinical management.

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