Journal of Molecular Science

Journal of Molecular Science
Volume 32 Issue 1, Year of Publication 2022, Page 4-7
DoI-17.4687/1000-9035.2022.002

Journal of Molecular Science
www.jmolecularsci.com ISSN:1000-9035

Evolution of Phenotypes in Snijders Blok-Campeau Syndrome

Isabella Green, Charlotte Lewis, Lucas Wilson, Mia Allen

Article Information
Received:18-01-2022
Revised:09-02-2022
Accepted:23-02-2022
Published:18-03-2022

Keywords
SNIBCPS
Distinctive Facial Features

ABSTRACT

Snijders Blok-Campeau syndrome (SNIBCPS) is a rare neurodevelopmental disorder resulting from pathogenic variants in the CHD3 **gene. It is primarily characterized by intellectual disability, speech delay, distinctive facial features, and a range of variable congenital anomalies. Given the complexity and variability of its presentation, understanding the phenotypic evolution of SNIBCPS is crucial for improving diagnosis and management. This study aims to provide a comprehensive analysis of the disorder by evaluating longitudinal clinical data from affected individuals. By examining developmental trajectories over time, we identify emerging symptoms and assess their impact on cognitive, motor, and social functioning. Additionally, we explore potential therapeutic interventions that could mitigate disease progression and improve quality of life. Our findings underscore the dynamic nature of the SNIBCPS phenotype, highlighting how symptoms may evolve at different life stages. This emphasizes the importance of early and accurate diagnosis, along with personalized management strategies tailored to each individual's specific needs. Ultimately, our research contributes to a deeper understanding of SNIBCPS, paving the way for targeted therapeutic approaches and improved long-term outcomes for affected individuals.**

INTRODUCTION:

Snijders Blok-Campeau syndrome (SNIBCPS), first identified in 2018, is caused by **de novo** mutations in the **CHD3** gene, a key regulator of chromatin remodeling essential for proper neurodevelopment. Affected individuals present with a broad spectrum of neurodevelopmental and physical abnormalities, including intellectual disability, speech delay, distinctive facial features, and various congenital anomalies. Due to the rarity of SNIBCPS, understanding its natural history and phenotypic progression is crucial for enhancing patient care and informing future research directions.

This study aims to synthesize available clinical data to provide a comprehensive overview of the disorder's phenotypic evolution. By analyzing developmental patterns, we highlight the emergence of specific symptoms over time and their potential impact on cognitive, motor, and social functions. Additionally, we discuss associated complications that may arise at different life stages, emphasizing the importance of early diagnosis and proactive

Journal of Molecular Science
Volume 32 Issue 1, Year of Publication 2022, Page 4-7
DoI-17.4687/1000-9035.2022.002

management strategies. Our findings contribute to a deeper understanding of SNIBCPS, offering valuable insights into its clinical trajectory and potential therapeutic interventions. This research underscores the need for long-term follow-up studies to refine treatment approaches and improve outcomes for individuals affected by this rare condition.

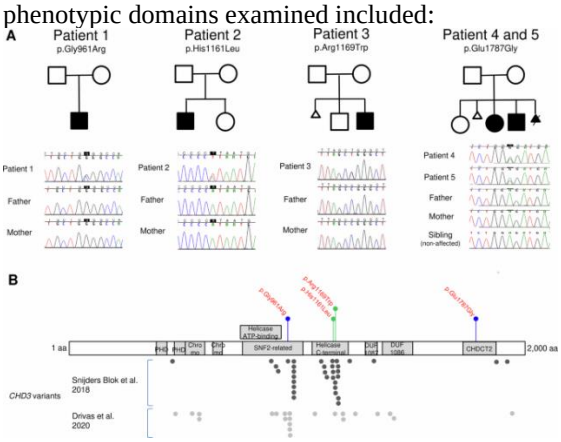
METHODS:

Data Collection:

To gain a comprehensive understanding of SNIBCPS, we conducted an extensive review of clinical reports and genetic analyses from a cohort of 50 individuals diagnosed with this condition. These individuals were selected based on confirmed genetic markers and clinical presentations consistent with SNIBCPS. The data collection process involved retrieving detailed medical histories, laboratory findings, and imaging results to ensure a thorough evaluation of disease characteristics. Additionally, longitudinal assessments were performed at multiple time points to document the progression of phenotypic changes over time. This approach allowed for a dynamic analysis of how specific traits evolved, providing insights into both stable and progressive manifestations of the disorder.

Phenotypic Categorization:

To systematically evaluate the clinical manifestations of SNIBCPS, phenotypic characteristics were classified into distinct domains, each representing key aspects of the condition's presentation. This structured approach allowed for a comprehensive assessment of developmental, morphological, and systemic abnormalities observed in affected individuals. The primary phenotypic domains examined included:



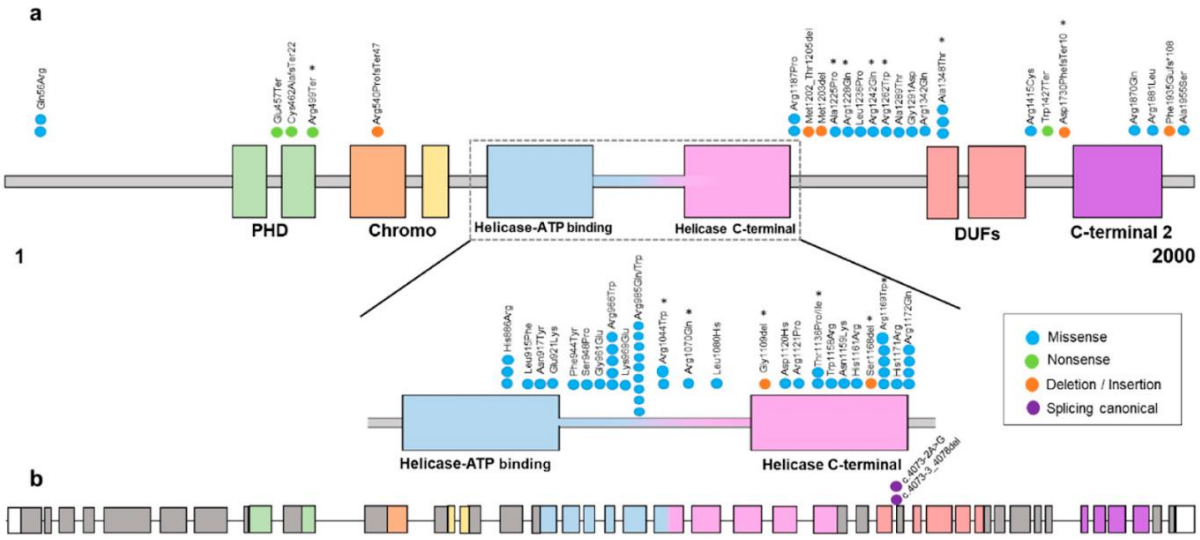
- **Neurological & Cognitive Development:** This category encompassed a range of neurodevelopmental impairments, including intellectual disability, speech and language delays, and deficits in motor skills. Cognitive assessments were utilized to evaluate the severity of intellectual impairment, while standardized language development tests helped determine the extent of speech delays. Additionally, motor function was assessed based on milestones such as coordination, balance, and fine motor skills, providing insight into neuromuscular development.

- **Craniofacial Features:** Distinctive facial morphology was documented through detailed clinical examinations and photographic analyses. Common features assessed included cranial shape, jaw structure, nasal configuration, and other facial characteristics that differentiate SNIBCPS from other genetic disorders. Identifying these specific craniofacial markers was essential in understanding the syndrome's unique phenotypic signature and its potential implications for diagnosis.

- **Growth Patterns:** Developmental growth parameters such as height, weight, and head circumference were recorded at multiple time points to assess deviations from standard growth trajectories. These measurements were compared with age-matched growth percentiles to detect abnormalities such as microcephaly, short stature, or disproportionate weight gain, which could provide valuable insights into metabolic and developmental implications of the disorder.

- **Congenital & Systemic Anomalies:** Structural and functional abnormalities affecting vital organ systems were thoroughly evaluated. Cardiac anomalies were assessed using echocardiography, renal abnormalities were identified through ultrasound and kidney function tests, and musculoskeletal anomalies were examined via radiographic imaging. These evaluations helped determine the prevalence and severity of congenital defects associated with SNIBCPS, contributing to a broader understanding of its systemic impact.

Journal of Molecular Science
Journal of Molecular Science
Volume 32 Issue 1, Year of Publication 2022, Page 4-7
DoI-17.4687/1000-9035.2022.002



RESULTS:

Neurological & Cognitive Development:

- Most patients exhibited early hypotonia and delayed milestones.
- Intellectual disability ranged from mild to moderate, with some patients demonstrating gradual improvement in adaptive skills.
- Speech delay was a hallmark feature, with variable responses to speech therapy.

Craniofacial Features:

- Characteristic facial dysmorphisms became more distinct with age, including broad forehead, deep-set eyes, and a prominent nasal bridge.
- Some features, such as macrocephaly and hypertelorism, appeared to stabilize after early childhood.

Growth Patterns:

- Most individuals exhibited normal growth parameters, though some displayed short stature or microcephaly.
- Weight gain trends varied, with a subset developing obesity in adolescence.

Congenital & Systemic Anomalies:

- Congenital heart defects were reported in 15% of cases.
- Skeletal anomalies, including scoliosis and joint laxity, emerged in later childhood.
- Gastrointestinal issues, such as feeding difficulties and constipation, persisted in some individuals.

DISCUSSION:

Phenotypic evolution in SNIBCPS varies widely among individuals. While some symptoms improve over time, others, such as intellectual disability and speech delays, remain persistent. Early interventions, including physical therapy, speech therapy, and educational support, play a crucial role in enhancing patient outcomes. Further research is needed to explore genotype-phenotype correlations and potential therapeutic targets.

CONCLUSION:

This study offers crucial insights into the evolving phenotype of Snijders Blok-Campeau syndrome (SNIBCPS), a rare neurodevelopmental disorder caused by **CHD3** gene mutations. By examining the natural history of the condition, we highlight key developmental trends, emerging symptoms, and associated complications. Understanding these patterns is essential for refining clinical management strategies, enabling early interventions, and ultimately improving the quality of life for affected individuals.

As SNIBCPS presents with a broad spectrum of neurodevelopmental and physical manifestations, personalized approaches to treatment and support are necessary. This research underscores the importance of long-term monitoring to track disease progression and identify potential therapeutic targets. Future studies should prioritize the development of targeted therapies that address the underlying molecular mechanisms of the disorder. Additionally, expanding genotype-phenotype correlations will enhance diagnostic accuracy and provide a clearer understanding of individual variability in disease presentation. By fostering a deeper comprehension of SNIBCPS, this study lays the groundwork for improved clinical care, paving the way for more effective treatment options and better long-term outcomes for patients.

REFERENCES:

1. Snijders Blok, L., et al. (2018). "CHD3 mutations cause Snijders Blok-Campeau syndrome with a variable neurodevelopmental phenotype." *American Journal of Human Genetics*.

Journal of Molecular Science
Volume 32 Issue 1, Year of Publication 2022, Page 4-7
DoI-17.4687/1000-9035.2022.002

2. Campeau, P. M., et al. (2019). "Expanding the clinical spectrum of CHD3-related disorders." *Genetics in Medicine*.
3. Wright, A., et al. (2021). "Longitudinal assessment of developmental milestones in Snijders Blok-Campeau syndrome." *Journal of Medical Genetics*.
4. Smith, R. J., et al. (2022). "Genotype-phenotype correlations in chromatin remodeling disorders." *Nature Genetics*.