

Clinical Features, Genetic Basis, and Management of Jacobsen Syndrome in Pediatric Populations

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DESCRIPTION

Jacobsen Syndrome (JS), also known as 11q terminal deletion disorder, is a rare chromosomal disorder caused by deletions at the distal end of chromosome 11. First described in 1973 by Petrea Jacobsen, the syndrome is characterized by a wide spectrum of phenotypic features, including congenital heart defects, craniofacial dysmorphism, hematologic abnormalities, developmental delays, and behavioral challenges. Despite its rarity, with an estimated incidence of 1 in 100,000 live births, understanding the clinical presentation and management of JS is critical for pediatric care providers, geneticists, and families affected by the condition.

The genetic basis of Jacobsen syndrome involves partial deletion of the long arm of chromosome 11, most commonly spanning 7 megabases-20 megabases. This deletion affects multiple genes, contributing to the variable phenotype observed in patients. Craniofacial anomalies are among the most recognizable features of JS. Affected children often present with trigonocephaly, broad forehead, hypertelorism, down-slanting palpebral fissures, ptosis, and a small, triangular-shaped nose. Additionally, low-set ears, micrognathia, and palate anomalies such as cleft palate or high-arched palate are common. These features may influence feeding, speech, and dental development, requiring multidisciplinary evaluation and intervention.

Congenital heart defects are observed in approximately 50%-60% of children with Jacobsen syndrome. The most frequent abnormalities include ventricular septal defects, atrial septal defects, tetralogy of Fallot, and hypoplastic left heart syndrome. Early cardiac evaluation using echocardiography and ongoing cardiology follow-up are essential, as timely surgical or medical intervention can improve survival and long-term cardiac function. Hematologic abnormalities are hallmark features of JS, most notably Paris-Trousseau thrombocytopenia, characterized by reduced platelet count and platelet dysfunction. This condition increases bleeding risk, particularly during infancy and childhood. Hematologic monitoring, supportive care, and prophylactic measures are critical. Some children may require

platelet transfusions or specialized hematologic interventions in cases of severe bleeding.

Neurodevelopmental outcomes in Jacobsen syndrome are variable, ranging from mild learning difficulties to significant intellectual disability. Cognitive impairments often correlate with the size of the chromosomal deletion. Speech and language development are frequently delayed, with expressive language more affected than receptive language. Early intervention, including speech therapy, occupational therapy, and special education support, is essential to maximize developmental potential. Behavioral characteristics associated with JS may include Attention-Deficit/Hyperactivity Disorder (ADHD), autism spectrum traits, anxiety, and emotional dysregulation. Behavioral therapy, structured routines, and individualized educational planning are recommended to support social adaptation and functional independence. Multidisciplinary collaboration between psychologists, educators, and caregivers is essential for optimizing behavioral outcomes.

Growth delays are also commonly observed. Many children exhibit short stature, low weight, and delayed bone age. Endocrine evaluation may be warranted to assess growth hormone levels, thyroid function, and nutritional status. Nutritional support, monitoring of caloric intake, and early intervention for feeding difficulties contribute to improved growth outcomes. Musculoskeletal anomalies in Jacobsen syndrome may include joint laxity, scoliosis, clinodactyly, and foot deformities. Physical therapy and orthopedic interventions help improve motor function, posture, and mobility. Early assessment and intervention enhance participation in daily activities and prevent secondary complications associated with musculoskeletal dysfunction. Ophthalmologic and auditory evaluations are recommended, as strabismus, refractive errors, and hearing loss are more prevalent in this population. Corrective interventions, such as glasses, surgery for strabismus, or hearing aids, contribute to improved sensory function, communication, and overall quality of life.

Long-term management requires a multidisciplinary approach, including pediatricians, geneticists, cardiologists, hematologists,

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neurologists, endocrinologists, therapists, and educators. Regular monitoring, early interventions, and individualized care plans are essential to optimize health outcomes, functional abilities, and psychosocial wellbeing. Coordination of care between home, school, and medical environments ensures consistent support and improves quality of life. Emerging research on genotype-phenotype correlations, molecular pathways affected by 11q deletions, and potential therapeutic targets offers hope for more personalized management strategies. Ongoing studies aim to identify biomarkers for prognosis, explore gene therapy approaches, and develop interventions to mitigate cognitive and behavioral challenges.

CONCLUSION

Jacobsen syndrome is a complex chromosomal disorder with multisystem involvement. Early diagnosis, careful monitoring, and comprehensive multidisciplinary management are essential for optimizing outcomes. Attention to cardiac, hematologic, neurodevelopmental, musculoskeletal, sensory, and behavioral aspects ensures holistic care. Family education, support, and involvement are central to effective management. Advances in molecular genetics and research into targeted therapies hold promise for improving the long-term health, development, and quality of life for children affected by Jacobsen syndrome.