Surgical correction of trigonocephaly in Jacobsen syndrome

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Abstract

Jacobsen syndrome is a condition caused by a loss of genetic material from chromosome 11. Because this deletion occurs at the end (terminus) of the long (q) arm of chromosome 11, Jacobsen syndrome is also known as 11q terminal deletion disorder. Patients have a spectrum of malformations of the heart, kidney, gastrointestinal tract, genitalia, central nervous system and skeleton. Trigonocephaly is a typical feature of skull deformity. Due to the complex pattern of the malformations and the wide spectrum of clinical features the surgical correction of trigonocephaly is challenging. The surgical correction technique of trigonocephaly in an 8-month-old infant with Jacobsen syndrome is reported.

Keywords: Craniofacial surgery, Metopic synostosis, Trigonocephaly, Jacobsen syndrome.
Introduction

11q terminal deletion disorder is a rare genetic disorder. It is known as a terminal deletion disorder because it is caused by the loss of genes from the end of chromosome 11. It is also called Jacobsen syndrome (JS) after the Danish researcher who first identified the disorder in 1973.1-3

JS is a condition caused by a loss of genetic material from chromosome 11. Because this deletion occurs at the end (terminus) of the long (q) arm of chromosome 11, the disorder is also known as 11q terminal deletion disorder. The estimated incidence of JS is 1 in 100,000 newborns. One of the craniofacial features in this syndrome is stenosis of the metopic suture.4

The majority of genetic causes are terminal deletions of 11q, and size ranges from 7 to 20 Mb. The breakpoints occur within distal to subband 11q23.3 and the deletion usually extends to the telomere. The deletion appears to be de novo in 85% of reported cases, and in 15% of cases it results from an unbalanced segregation of a familial balanced translocation or from other chromosome rearrangements. Only a few translocations between chromosome 11q and chromosomes 6, 16, 22, Y have been described. Detection of 11q chromosomal rearrangements using routine karyotyping and subtelomeric FISH, precise identification of breakpoints using microarrays and detailed analysis of affected genes lead to a clear, exact genotype-phenotype correlation and clinical determination of patients with JS.5-7

In this article the authors present their experience in the surgical treatment of trigonocephaly in an 8-month-old patient with JS by frontal and orbital bandeau remodeling.

Case Report

The authors report a case of an 8-month old male affected by JS. On physical examination he was quite a sociable child, with dysmorphic craniofacial features such as trigonocephaly with hypertelorism, downward slanting palpebral fissures, epicanthal folds, flat nasal bridge, short
nose with flat philtrum, and thin upper lip. Pre-operative computed tomography (CT) and magnetic resonance imaging (MRI) are necessary to confirm the clinical diagnosis of trigonocephaly. CT scan with 3D reconstruction allows the surgeon to plan the surgical correction. MRI shows soft-tissue alterations, such as triangular brain deformation anteriorly and can be used to detect clinical evidence of increased intracranial pressure.8-10

Surgical Technique. Infiltration with local anesthesia and vasoconstrictors at the level of the frontal temperoparietal orbital and coronal region. Coronal incision, dissection of the frontal tempoparietal naso-orbital region keeping the temporalis muscle in the coronal flap.

Drawing of osteotomic demarcation lines with delimitation of the supraorbital bandeau and two extended side tendons, and delineation also of the frontal flap in front of the fontanelle and coronal suture. Bi-frontal craniotomy with removal of the frontal flap and supraorbital bandeau. Suture of dural tears. Remodeling of the bandeau with its linearization and correction at the level of the metopic suture initially stabilized with metal threads.

Repositioning of the bandeau with advancement of about 1 cm at the level of the nasion and of about 2 cm at the level of the orbital bilateral roofs. The bandeau is secured with plaques and resorbable screws and the metal threads are removed. Bilateral bone grafting at the level of the side wall and of the orbital floor.

Osteotomy of the frontal flap along the metopic suture to obtain two hemiflaps from which two triangular-shaped bone fragments are cut. For each side, three radial temporoparietal osteotomies (barrel stave-type) are done with greenstick fracture of the barrel stave for each hemiflap.

Positioning of bone grafts at the level of the orbital roofs. Fixation of the two hemiflaps with resorbable plaques and screws, and bone grafts at the level of the median line. Tissucol. Drainage. Suture.8-10
Discussion

11q terminal deletion disorder is a rare genetic disorder. It is known as a terminal deletion disorder because it is caused by the loss of genes from the end of chromosome. It is also called Jacobsen syndrome (JS) after the Danish researcher who first identified the disorder in 1973. JS is a condition caused by a loss of genetic material from chromosome 11. Because this deletion occurs at the end (terminus) of the long (q) arm of chromosome 11, JS is also known as 11q terminal deletion disorder. The estimated incidence of the disorder is 1 in 100,000 newborns.

The signs and symptoms of JS vary considerably. Most affected individuals have delayed development, including the development of motor skills (sitting, standing, and walking) and speech. Most also have cognitive impairment and learning difficulties. Behavioral problems have been reported, including compulsive behavior (shredding paper), a short attention span, and easy distractibility. Many people with the condition have been diagnosed with attention deficit-hyperactivity disorder (ADHD).

More than 90 percent of people with JS have a bleeding disorder called Paris-Trousseau syndrome. This condition causes a lifelong risk of abnormal bleeding and easy bruising. Paris- Trousseau syndrome is a disorder of platelets, which are blood cell fragments that are necessary for blood clotting.

Other features of JS can include heart defects, feeding difficulties in infancy, short stature, frequent ear and sinus infections, and skeletal abnormalities. The disorder can also affect the digestive system, kidneys, and genitalia. The life expectancy of people with the disorder is unknown, although affected individuals have lived into adulthood.

JS is also characterized by distinctive facial features. These include small and low-set ears, hypertelorism, ptosis, epicanthal folds, a broad nasal bridge, downturned corners of the
mouth, a thin upper lip, and a small lower jaw. Affected individuals often have macrocephaly and trigonocephaly, which gives the forehead a pointed appearance.\textsuperscript{7,8}

Trigonocephaly is a frontal bone anomaly associated with synostosis of the metopic suture. It is characterized by a triangular appearance of the forehead when viewed from above and is usually associated with ocular hypotelorism.\textsuperscript{6,9}

In patients with the classical phenotype, the diagnosis is suspected on the basis of clinical findings: intellectual disability, facial dysmorphic features and thrombocytopenia. The diagnosis must be confirmed by cytogenetic analysis. The clinical diagnosis may be difficult in patients with less characteristic clinical aspects, borderline mental development, and with no thrombocytopenia. Children with JS share some clinical features (short stature, short, wide, sometimes webbed neck, downsloping palpebral fissures, ptosis, and aortic or pulmonary stenosis) with Turner and Noonan syndromes. Prenatal diagnosis is possible by amniocentesis or chorionic villus sampling and cytogenetic analysis.\textsuperscript{9,10}

Management is multidisciplinary and requires evaluation by a general pediatrician, pediatric cardiologist, neurologist, ophthalmologist, neonatologist and maxillo-facial surgeon.\textsuperscript{8}

One of craniofacial distinctive features in JS is trigonocephaly which is the clinical result of metopic synostosis, with the keel-shaped forehead, hypotelorism and epicanthus, and temporal narrowing with an associated abnormality of the supraorbital rim. Physical examination is the first approach in the diagnosis of metopic suture craniosynostoses.\textsuperscript{8,10}

In conclusion, JS is a complex craniofacial malformation characterized not only by stenosis of the metopic suture but also by other anomalies. The operation performed at the age of 8 months has enabled radical changes in the morphology and cranio-orbito-facial aesthetics with correction of trigonocephaly and hypotelorism.
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Declarations

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Patient Consent: Yes
References


Figure Legends

**Fig. 1.** Schematic drawings of the surgical plan: osteotomies, osteotomies, remodeling, bone grafts, resorbable devices.

**Fig. 2.** The baby before surgery

**Fig. 3.** Post-operative result 1 year after surgery with a radical change of cranio-fronto-orbital morphology and shape
Osteotomies

- Bone graft
- Resorbable devices