This 1-day-old baby boy was born with the following anomalies: bilateral cleft lip and palate, hypotelorism, scalp defect, bulbous nose, patent urachus, cryptorchidism, hydrocele, simian crease of right hand, and overlapping fingers.

He was born to a 33-year-old woman (gravida 1) at 35 weeks gestational age via emergency cesarean delivery secondary to decreased fetal movement. The mother had a healthy pregnancy. Her serum triple screen for fetal abnormalities showed normal results. Fetal ultrasonography at 20 weeks was also normal.

At delivery, oligohydramnios was noted. Apgar scores were 0, 0, and 0 at 1, 5, and 10 minutes, respectively. The baby was immediately intubated and successfully resuscitated.

**What condition is this? What rapid diagnostic test would you order?**

**ANSWER: TRISOMY 13**

Trisomy 13 was clinically suspected. Because of the patient's critical condition, a fluorescent in situ hybridization (FISH) test was done to determine the diagnosis quickly. Results showed trisomy 13 (Figure 1). Because this syndrome is associated with early death and severe mental retardation, the parents opted to withdraw support for their baby. He died shortly thereafter.

Autopsy showed extensive cortical hemorrhages, atrial septal defect, patent ductus arteriosus, and multiple external anomalies.

**WHAT IS TRISOMY 13?**

In 1960, Klaus Patau first reported trisomy 13 in a full-term female infant with a cerebral defect, anophthalmia, cleft lip and palate, simian creases, polydactyly, capillary hemangioma, and a heart defect. Subsequent cytogenetic analysis confirmed trisomy 13 in that patient.

The frequency of trisomy 13 in spontaneously aborted fetuses is estimated to be about 8%. The incidence of trisomy 13 is about 1 in 5000 live births. Although this condition is considered to be lethal, relatively long-term survival has been reported. About 28% of newborns die during the first week of life; 44% succumb within the first month, and 86% die within the first year. The most common reported causes of death are cardiopulmonary arrest (69%), congenital heart disease (13%), and pneumonia (4%).

About 5% to 10% survive beyond the first year. Race and gender seem to affect survival in this condition. Females and African Americans have higher median ages at death. Survivors often have profound mental retardation, seizures, apneic spells, deafness, and reduced vision or blindness.

**CYTOGENETICS**

Trisomy 13 occurs when there are 3 representatives of chromosome 13 (as seen in our patient) instead of the usual 2 (Figure 2). Meiotic nondisjunction—the failure of a chromosome pair to separate—is the mechanism for the chromosome error in most cases. Occasionally, cases result from translocation or post-conception mosaicism.

Primary trisomy 13 is present in approximately 75% of cases; 4% are mosaics, 10% have an unbalanced (13q or 13q14q) translocation (Figure 3). At least half of cases involving (13q14q) translocations but fewer than 10% of cases with (13q13q) translocations are familial.

Maternal age in trisomy 13 is bimodally distributed, which is consistent with 2 distinct mechanisms. The presence of an extra chromosome 13 is a maternal age-dependent phenomenon. In contrast, trisomy 13 associated with translocation is independent of the age of the mother.

There is a 1% chance that trisomy 13 will recur in subsequent siblings. However, for mothers 35 years and older, the recurrence risk is the same as that for trisomy 21. If chromosomal analysis shows a translocation type of trisomy 13, then chromosomal analysis should also be done on the parents. The reproductive risk to a (13q13q) translocation carrier parent is 100%, and for a (13q14q) translocation carrier parent it is estimated to be 1% for a carrier female and less than 1% for a carrier male. Trisomy 13 can also be diagnosed prenatally using amniotic fluid cells, blood, or placental tissues for chromosomal analysis.

**CLINICAL FEATURES**

Common features seen in most of these patients are.
Facial features: Microcephaly, sloping forehead, hypotelorism, bulbous nose, low-set malformed ears, microphthalmia/anophthalmia, coloboma of eye structures, cleft lip (often midline or bilateral), low-set ears, and abnormal helices.

CNS abnormalities: Holoprosencephaly-type defect with varying degrees of incomplete development of forebrain and olfactory and optic nerves; minor motor seizures and deafness from defect of the Corti organ.

Cardiac: Ventricular septal defect (80%), patent ductus arteriosus, atrial septal defect, and dextrocardia. Other cardiac defects include anomalous venous return, overriding aorta, pulmonary stenosis, atretic mitral/aortic valves, and bicuspid aortic valve (less common).

Skin: Cutis aplasia of scalp, capillary hemangioma, and loose skin in the posterior neck.

Skeletal: Thin posterior ribs with or without missing ribs and hypoplasia of pelvis with shallow acetabular angle.

Hands and feet: Simian crease, hyperconvex narrow fingernails, flexion contractures of fingers with or without overlapping, ulnar and fibular polydactyly, posterior prominence of heel, hypoplastic toenails, syndactyly, equinovarus, and (less commonly) radial aplasia.

Genitourinary: Cystic kidneys, duplicated ureters (31%); in males, cryptorchidism and abnormal scrotum. In females, bicornuate uterus. A patent urachus (as in this case) was present in the first reported case of trisomy 13.¹

Abdominal (less common): Malrotation of colon, omphalocele, heterotopic pancreatic or splenic tissue, and Meckel diverticulum.

Hematologic: Increased frequency of nuclear projections in neutrophils and unusual persistence of fetal-type hemoglobin.

**DIAGNOSIS**

A standard chromosomal analysis usually takes about 2 to 3 days. FISH can provide the diagnosis of trisomy 13 in 6 to 8 hours. FISH involves the use of fluorescent-labeled DNA probes of known chromosomal regions to detect the presence of the hybridized fluorescent signal and, hence, the presence of the chromosomal material. FISH should be followed by a standard chromosomal analysis to rule out a translocation for genetic counseling.

**MANAGEMENT**

Trisomy 13 is associated with high mortality. Surgical or orthopedic corrective procedures should be withheld in early infancy to await the outcome of the first few months. The use of extraordinary medical means to prolong the life of babies with this syndrome is not recommended.⁹ Therefore, a rapid diagnosis is essential to planning further management and discussing the prognosis with the parents. It is also important that the full spectrum of the natural history of this condition be presented to the parents to enable them to make the best possible decisions for their family. Finally, it should be emphasized that each case must be taken on an individual basis and the personal feelings of the parents must be taken into consideration.⁵

**References:**


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