Condition: Trisomy 13 (Patau syndrome)

Inheritance:
Chromosomal

Genetic etiology:
Trisomy for chromosome 13 due to nondisjunction.

Frequency:
Approximately 1:12,000 livebirths.

Clinical features:
Infants with trisomy 13 are born with low birthweight and have multiple congenital anomalies. Most notable are facial anomalies, including hypotelorism, and, in many cases, cleft lip and palate. There may be areas of deficient skin in the scalp and "rocker bottom feet." Anomalies of internal organs, including the heart, brain, and kidneys, are common.

Management:
Most affected children die in infancy or childhood. Those who survive tend to have severe developmental impairment.

Genetic counseling:
Couples who have had an affected child are usually counseled that recurrence risk is around 1%. Trisomy 13 can be detected by prenatal chromosomal analysis.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.