Condition: Trisomy 18 (Edwards syndrome)

Inheritance:

Chromosomal

Genetic etiology:

Trisomy for chromosome 18 due to nondisjunction.

Frequency:

Approximately 1:6,000 livebirths.

Clinical features:

Infants with trisomy 18 are born with multiple congenital anomalies. Most notable are low birth weight, prominent occiput, and tightly clenched fingers. Internal organ malformations, especially of the heart and brain, are common.

Management:

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

Genetic counseling:

Couples who have had an affected child are usually counseled that recurrence risk is around 1%. Trisomy 18 can be detected by prenatal chromosomal analysis.

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