

INDICATIONS FOR CHROMOSOME STUDIES

1. Advanced Maternal Age
2. Previous Child with Chromosome Anomaly
3. Abnormal AFP
4. Ultrasound Anomalies
5. Ambiguous Genitalia and Undescended Testes
6. Multiple Congenital Anomalies
7. Dysmorphic Features in a Child or Adult
8. Mental Disability of Unknown Cause
9. Amenorrhea (Primary and Secondary)
10. Gynecomastia and Oligospermia
11. Infertility (Male and Female)
12. Recurrent Pregnancy Loss
13. Balanced Translocations
14. Leukemia; Lymphoma; Myeloproliferative and Myelodysplastic Disorders; Cytopenia and Tumors

Most insurance companies provide coverage for all of the previously mentioned disorders.

If you have any questions regarding anything mentioned in the pamphlet, ask your physician or contact Genetics Associates, Inc.
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CHROMOSOME ANALYSIS



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COMMON CHROMOSOMAL ANOMALIES

TRISOMY 21 (DOWN SYNDROME)

Trisomy 21 is the most common chromosomal anomaly causing moderate to severe mental disability. Typically, 25 to 30% die within the first year, 50% before five years. Occurrence is 1 in 700 births.

TRISOMY 18 (EDWARDS SYNDROME)

Severity is profound. Ninety-five percent have heart malformations (almost always the cause of death). Average survival is 2 to 3 months for males and 10 months for females. Rarely do patients survive childhood. Occurrence is 1 in 8,000 births.

TRISOMY 13 (PATAU SYNDROME)

Malformations can be very profound, affecting several major organ systems. The majority of patients die within the first month. Occurrence is 1 in 10,000 births.

KLINEFELTER SYNDROME 47, XXY

Patients are sterile with very few dysmorphic features. Gynecomastia (development of breast tissue in males) may occur at puberty. Klinefelter syndrome is generally recognizable at that time. Occurrence is 1 in 600 births.

TURNER SYNDROME 45, X

Patients are short in stature and infertile. Affected individuals may experience a webbing of the neck and/or swelling of the hands and feet at birth. Life expectancy is normal. Occurrence is 1 in 2,000 births.

FRAGILE X SYNDROME

Fragile X is the second most common chromosomal form of mental disability following Down syndrome. Affected individuals are predominantly male. This disorder is diagnosed via molecular genetic analysis. Life expectancy is normal.

TESTS PERFORMED

AMNIOCENTESIS

An amniocentesis is a procedure in which a small amount of amniotic fluid is removed for testing purposes. Normally performed between 14 and 18 weeks of gestation, an amniocentesis yields definitive results concerning chromosome normality and fetal sex. In addition, biochemical analysis of enzymes such as alpha-fetoprotein (AFP) and acetylcholinesterase (AChE) can be performed to help diagnose neural tube defects. The procedure related risk of miscarriage is 1 in 200. Final results can be expected within 7 to 10 days.

CHORIONIC VILLUS SAMPLING

Chorionic villus sampling involves removing a small sample from the placenta at the point where it attaches to the uterine wall. The sample can be collected transcervically or transabdominally (similar to an amniocentesis). This procedure is performed between 10 and 12 weeks of gestation. Like the amniocentesis, this test yields decisive results regarding chromosome normality and fetal sex. Unlike the amniocentesis, AFP/AChE testing cannot be performed on chorionic villi. The procedure related risk of miscarriage is 1 in 100. Preliminary results may be available in 2-3 days, with final results following within 7-10 days.

PERCUTANEOUS UMBILICAL BLOOD SAMPLE (PUBS)

Also known as cordocentesis, PUBS testing entails drawing a sample of fetal blood from the umbilical cord after 20 weeks of gestation following the discovery of fetal anomalies. Preliminary results can be expected in 1 day, with a final result following in 3 days.

PERIPHERAL BLOOD

A blood sample is collected for newborns with multiple congenital anomalies, children and adults with developmental delay and/or mental disability, and patients experiencing infertility and/or recurrent miscarriages. Newborn preliminary results can be expected in 1 day. Final results are available within 3 to 7 days.