





PEDIATRIC MOLECULAR GENETICS

CYTOSNP-850K BEADCHIP MICROARRAY PEDIATRIC PANEL

TURNAROUND TIME: 10-14 DAYS

This test is an appropriate follow-up test for individuals with:

- Unexplained developmental delay or intellectual disability
- Autism spectrum disorders
- Congenital anomalies with a previously normal conventional chromosome study



Provides the most comprehensive coverage of cytogenomicrelevant genes for constitutional abnormalities. Increased performance and detection sensitivity. High Detection Sensitivity for low-level mosaics.

First tier, postnatal test for individuals with multiple anomalies that are not specific to well-delineated genetic syndromes, nonsyndromic developmental delay or intellectual disability, or autism spectrum disorders.

Determining the size, precise breakpoints, gene content, and any unappreciated complexity of abnormalities detected by other methods such as conventional chromosome and FISH studies.

Determining if apparently balanced abnormalities identified by previous conventional chromosome studies have cryptic imbalances, since a proportion of such rearrangements that appear balanced at the resolution of a chromosome study are actually unbalanced when analyzed by higher resolution chromosomal microarray.

Assessing regions of homozygosity related to uniparental disomy or identity by descent.



IN ADDITION TO PEDIATRIC MICROARRAY, GENESYS DIAGNOSTICS ALSO OFFERS:

CHROMOSOME ANALYSIS

Turnaround time: 3-10 days

- Evaluation of number and structure of all 46 human chromosomes, relating any abnormalities to possible disease
- Useful for determining possible causes for infertility, stillbirth, and multiple spontaneous abortions
- Band-for-band analysis of each chromosome after the final processing step of staining
- High resolution studies available



FRAGILE X TESTING

Turnaround time: 7-14 days

 Used to determine repeat ranges of CGG repeats found in the Fragile X Messenger Ribonucleoprotein 1 (FMR1) gene, where greater than 200 repeats is considered affected.

FLUORESCENCE IN SITU HYBRIDIZATION (FISH)

Turnaround time: 4-6 days

Microdeletions

 DiGeorge, Prader-Willi, Angelman, Williams, SRY, Wolf-Hirschhorn, Cri-du-Chat, Smith-Magenis, Kallman, Miller-Dieker, STS, and XIST

ABOUT US

Genesys Diagnostics provides a range of clinical diagnostic services to physicians, hospitals, and laboratories across the country.

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