LifeLabs GENETICS™

Microdeletion Syndromes

panorama™ natera prenatal screen

Panorama™ now screens for the most common and severe microdeletion syndromes, in addition to its basic screen for trisomies 21, 18, 13, triploidy, and sex chromosome abnormalities.

Why Screen for Microdeletion Syndromes?

- They are common and can be severe
- They carry equal risk across all maternal ages
- They often go undiagnosed
- They may be responsive to early childhood intervention

Scientifically Validated

Microdeletion validation has been completed by Natera with 469 samples, including 110 confirmed positives. Accuracy of performance has been validated at fetal fractions as low as 3.8%.

Limitations of the Test

Panorama does not screen for all microdeletion syndromes. Performance specifications reflect presence or absence of the entire targeted region. Patients who screen high risk should be offered genetic counseling and a follow-up invasive procedure to confirm diagnosis.

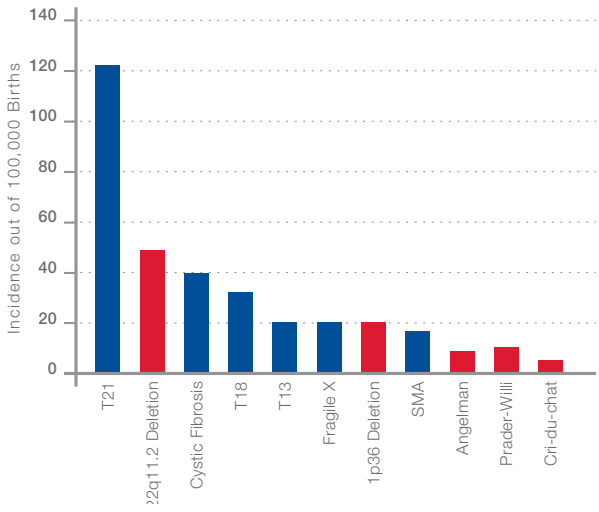
How to Order Panorama's Microdeletion Screening Outside the USA

You may order the Panorama Test alone or with one of these two options where available:

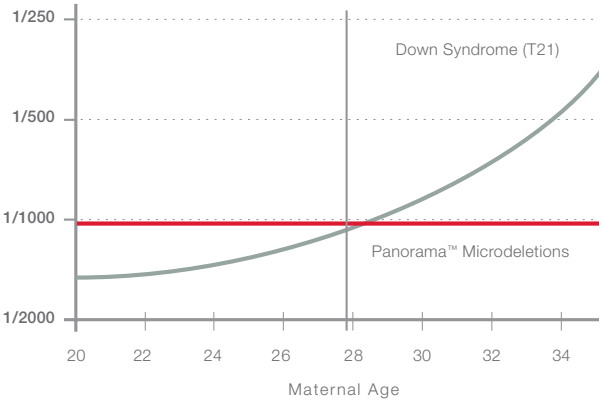
- 22q11.2 Deletion syndrome (also known as DiGeorge syndrome)
- Panorama Extended Panel which includes: 22q11.2 deletion, Prader-Willi, Angelman, Cri-du-chat and 1p36 deletion syndromes

Please Note: Microdeletion screening cannot be ordered separately from Panorama.

22q11.2 IS COMMON



MICRODELETIONS ARE MORE COMMON THAN DOWN SYNDROME IN YOUNGER WOMEN



Microdeletion Syndromes on the Panorama Panel

Syndrome	Incidence	Sensitivity [1,2]	Specificity [1,2]	Location Size of Region # of SNPs	Lifespan	Mental Effects	Heart Defects	Other features
22q11.2 Deletion/ DiGeorge	1 in 2,000 [3,4*]	95.7% (45/47) [9,10] (85.5-99.5%) [10]	>99% (419/422) (97.9-99.9%) [11]	22q11.2 (2.9 MB) 672 SNPs	Reduced	Mild to moderate intellectual disorder & schizophrenia	Yes	Palate and feeding issues, immune problems, low calcium, seizures
Prader-Willi	1 in 10,000 [5]	93.8% (15/16) (69.8-99.8) [11]	>99% (453/453) (99.2-100%) [11]	15q11-q13 Paternal (5.9 MB) 1,152 SNPs	Reduced	Mild to severe intellectual disorder & behavioral problems	No	Hypotonia in babies, insatiable appetite
Angelman	1 in 12,000 [6]	95.5% (21/22) (77.2-99.9%) [11]	>99% (447/447) (99.2-100%) [11]	15q11-q13 Maternal (5.9 MB) 1,152 SNPs	Normal	Severe intellectual disorder	No	"Happy" affect, ataxia, microcephaly, no speech, seizures
Cri-du-chat	1 in 20,000 [7]	>99% (24/24) (85.8-100%) [11]	>99% (444/445) (98.8-99.9%) [11]	5p15.2 (20 MB) 1,152 SNPs	Infancy to adult	Moderate to severe intellectual disorder & behavioral problems	No	Cat-like cry, growth problems, wide set eyes
1p36 Deletion	1 in 5,000 [8]	>99% (1/1) (2.5-100%) [11]	>99% (468/468) (99.2-100%) [11]	1p36 (10 MB) 1,152 SNPs	Normal in most	Severe intellectual disorder & behavioral problems	Yes	Limited/no language, hearing loss, abnormal ears, seizures
Total incidence: approximately 1 in 1,000								

** Recent studies have shown incidence rates as high as 1/992*

1. Performance specifications reflect presence or absence of the complete targeted region
2. Wapner et al. Expanding the scope of noninvasive prenatal testing: detection of fetal microdeletion syndromes. Am J Obstet Gynecol 2015; 212:xxx.
3. Nussbaum et al 2007. Thompson and Thompson Genetics in Medicine (7th edn). Oxford Saunders: Philadelphia
4. Grati FR, et al. Prevalence of recurrent pathogenic microdeletions and microduplications in over 9,500 pregnancies. "http://www.ncbi.nlm.nih.gov/pubmed/25962607" \o "Prenatal diagnosis." Prenat Diagn. 2015 May 11. doi: 10.1002/pd.4613.
5. http://www.ncbi.nlm.nih.gov/books/NBK1330/
6. http://www.ncbi.nlm.nih.gov/books/NBK1144/
7. http://omim.org/entry/123450.
8. http://www.ncbi.nlm.nih.gov/books/NBK1191/
9. Calculated based on the test performance including pregnancy samples
10. Calculated based on the test performance including artificial plasma samples
11. 95% confidence interval

For more information please call 1-844-363-4357 or email us at ask.genetics@lifelabs.com

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