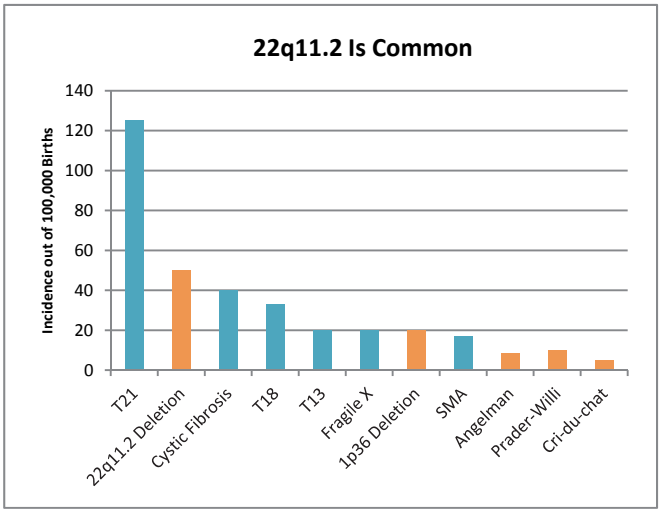
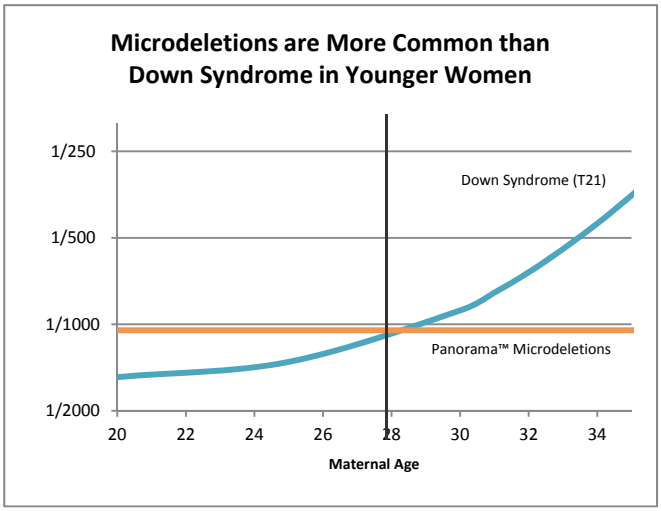
MICRODELETION SYNDROMES

Panorama™ now screens for the most common and severe microdeletion syndromes, in addition to its basic screen for T21, T18, T13, triploidy, and sex chromosome abnormalities.





Why Screen for Microdeletion Syndromes?

- ARE COMMON AND CAN BE SEVERE
- CARRY EQUAL RISK ACROSS ALL MATERNAL AGES
- OFTEN GO UNDIAGNOSED
- ARE 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 positive should be offered a follow-up invasive procedure to confirm diagnosis. Natera has a team of highly trained genetic counselors on staff who are available to discuss results with clinicians or directly with patients.

How to Order Panorama's Microdeletion Screening

When you order the "Panorama Prenatal Panel", 22q11.2 Deletion syndrome (also known as DiGeorge syndrome) will automatically be included. However you can opt out of 22q11.2 if you choose. If you would like to screen for the other 4 microdeletions, be sure to check "Panorama XP" on the requisition form.

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

For more information or to order Panorama kits, please contact the Client Services department of ARUP Laboratories at (800) 522-2787 or www.aruplab.com.

Syndrome	Incidence	Sensitivity [1]	Specificity [1]	Location Size of Region # of SNPs	Lifespan	Mental Effects	Heart Defects	Other features
22q11.2 Deletion/ DiGeorge	1 in 2,000 [2]	95.7% (45/47) [5, 6] (85.5-99.5%) [7]	>99% (419/422) (97.9-99.9%) [7]	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 [3]	93.8% (15/16) (69.8-99.8) [6]	>99% (453/453) (99.2-100%) [7]	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 [3]	95.5% (21/22) (77.2-99.9%) [6]	>99% (447/447) (99.2-100%) [7]	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 [4]	>99% (24/24) (85.8-100%) [7]	>99% (444/445) (98.8-99.9%) [7]	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 [3]	>99% (1/1) (2.5-100%) [7]	>99% (468/468) (99.2-100%) [7]	1p36 (10 MB) 1,152 SNPs	Normal in most	Severe intellectual disorder & behavioral problems	Yes	Limited/no language, hearing loss, abnormal ears, seizures, 2:1 M:F
Total incidence: approximately 1 in 1,000								

[1] Performance specifications reflect presence or absence of the complete targeted region

[2] Nussbaum et al 2007. *Thompson and Thompson Genetics in Medicine* (7th edn). Oxford Saunders: Philadelphia

[3] http://www.genetests.org.

[4] http://ncbi.nlm.nih.gov/entrez/disponim.cgi?id=123450

[5] Calculated based on the test performance including pregnancy samples

[6] Calculated based on the test performance including artificial plasma samples

[7] 95% confidence interval

These tests were developed by Natera, Inc., a laboratory certified under the Clinical Laboratory Improvement Amendments (CLIA). These tests have not been cleared by the Food and Drug Administration (FDA).