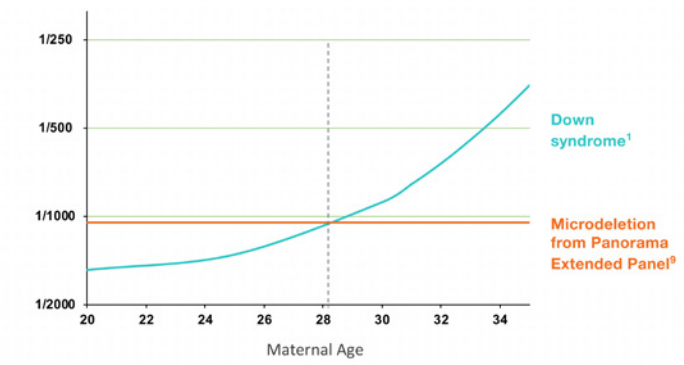
Microdeletion syndromes

Panorama screens for five microdeletions that are clinically relevant and have a combined incidence of 1 in 1000 live births, in addition to its basic screen for trisomies 21, 18, 13, triploidy, and sex chromosome abnormalities.

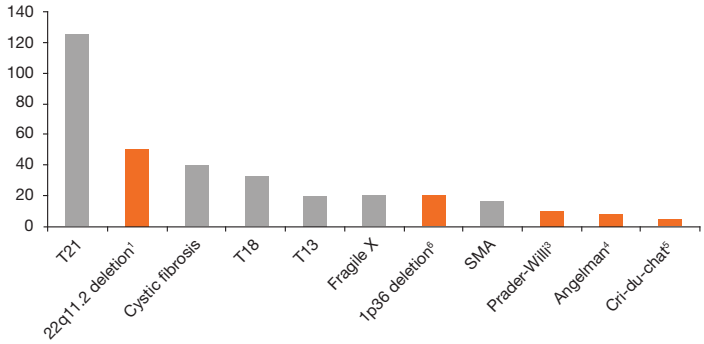
Why screen for microdeletion syndromes?

- They are common and can be severe.
- They carry equal likelihood across all maternal ages.
- They may be difficult to diagnose at birth.
- Early recognition enables early intervention, which may improve prognosis.

Microdeletions are more common than Down syndrome in younger women



Incidence of microdeletion syndromes screened by Panorama



Scientifically validated and clinically proven

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 2.8%. Panorama is validated for the microdeletions screened and offers the highest commercially available sensitivity for 22q11.2 deletion syndrome.[7,8]

Get started with Panorama's microdeletion screening

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

- 22q11.2 deletion syndrome (DiGeorge syndrome/VCFS/22q)
- Panorama Extended Panel which includes: 22q11.2 deletion syndrome, 1p36 deletion syndrome, Cri-du-chat syndrome, Angelman syndrome, and Prader-Willi syndrome

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

If you want to learn more about Panorama, visit **https://www.natera.com/panorama-test** or contact your Natera representative. You may also reach out to Natera's genetic counselors at niptgc@natera.com.

Panorama Extended Microdeletion Panel

Syndrome	Incidence	Sensitivity[7,8]	Specificity[8]	Location size and number of SNPs[1]	Clinical features of the syndrome may include:
22q11.2 deletion/ DiGeorge	1 in 2,000[2]	90%	>99%	22q11.2 (2.9Mb) 1358 SNPs	Mild to moderate intellectual disability, schizophrenia, feeding difficulties, immune disorders, low calcium, seizures.
Prader-Willi	1 in 10–30,000[3]	93.8%	>99%	15q11-q13 Maternal (5.9Mb) 1152 SNPs	Severe hypotonia and feeding difficulties in infancy, then gradual development of obesity; developmental delay; mild to severe intellectual disorder and behavioural problems; hypogonadism.
Angelman	1 in 12–24,000[4]	95.5%	>99%	15q11-q13 Paternal (5.9Mb) 1152 SNPs	Severe intellectual disability, ataxia, microcephaly and seizures.
Cri-du-chat	1 in 15–50,000[5]	>99%	>99%	5p15.2 (20Mb) 1152 SNPs	Cat-like cry, microcephaly, severe psychomotor and intellectual disability.
1p36 deletion	1 in 5–10,000[6]	>99%	>99%	1p36 (10Mb) 1152 SNPs	Developmental delay or intellectual disability, hypotonia, seizures, congenital heart defects, abnormalities of skeleton, kidneys, external genitalia, vision deficits.

Total incidence: Approximately 1 in 1,000

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