## The largest prospective NIPT study





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## **Study Objective**

**Panorama**<sup>™</sup>

Next-generation NIPT

To measure the performance of single nucleotide polymorphism (SNP)-based, non-invasive prenatal testing (NIPT) in a prospective study for trisomies 21, 18 and 13; monosomy X; 22q11.2 deletion syndrome; and microdeletion panel (1p36, Cri-du-chat, Prader-Willi and Angelman syndromes) in a large cohort of pregnant people clinically receiving NIPT.



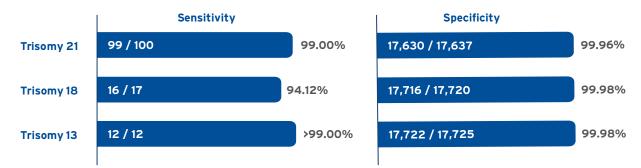
Certified Lab



## **Aneuploidy detection**

#### Demonstrated high performance in singleton pregnancies<sup>1</sup>

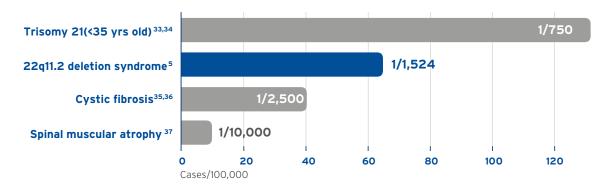
- Included a large cohort of average-risk pregnancies<sup>1</sup>
- Panorama™'s high sensitivity and specificity were maintained in real-world clinical practice



#### 22Q11.2 deletion detection

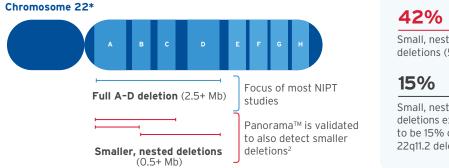
#### SMART study showed that 22q11.2 deletion syndrome had a higher than expected prevalence.

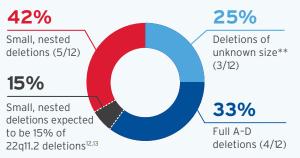
This is comparable to other conditions broadly recommended for routine screening, by professional medical guidelines, such as cystic fibrosis<sup>4, 25-29</sup>



### SMART data suggests that the incidence of small 22q11.2 deletions is much higher than expected.

Panorama<sup>™</sup> was able to detect all cases of the most common (2.5-3Mb)<sup>1</sup> 22q11.2 deletion, and 82% of all 22q11.2 deletions (0.5Mb+)



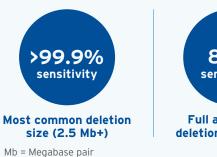


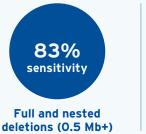
\*Not to scale

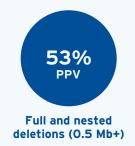
\*\*FISH or BACS-on-beads used for confirmatory testing; deletion spans A-B region at a minimum

#### Panorama™ delivered higher sensitivity for 22q11.2 deletion syndrome, enabling accurate and early detection

Panorama<sup>™</sup> was validated in SMART with high accuracy and high positive predictive value (PPV) for 22q11.2 deletions.<sup>1,4,5</sup>

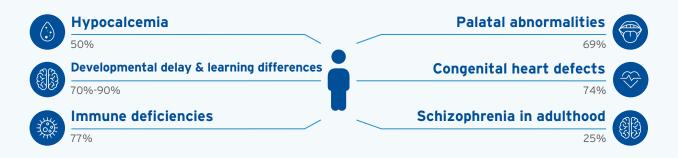






# Importance of early, accurate 22q11.2 deletion screening<sup>11-14</sup>

Perinatal interventions can improve quality of life of individuals affected by 22q11.2 deletion<sup>30-34</sup>



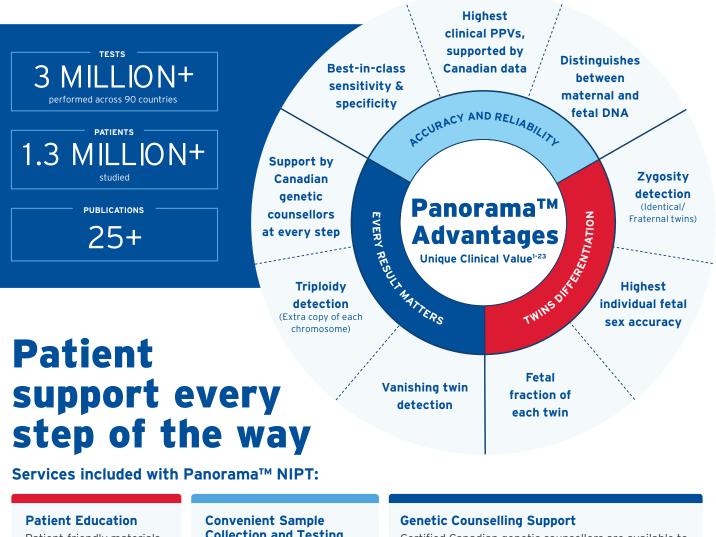
#### Early intervention can reduce the severity of these conditions associated with 22q11.2 deletion syndrome:<sup>11-14</sup>

- Delivery at tertiary center
- Calcium-level monitoring at birth
- Delayed live-vaccine administration
- Palatal evaluation for potential feeding and breathing issues



LifeLabsGenetics.com/Panorama

#### Why choose Panorama<sup>™</sup> NIPT?



Patient-friendly materials available on our website and patient service centres in English and French.

#### **Collection and Testing** Within Canada

Panorama<sup>™</sup> NIPT samples can be collected at any LifeLabs patient service centre or national affiliates and testing is performed within Canada.

Certified Canadian genetic counsellors are available to healthcare providers and patients to answer any questions about the test or results at Ask.Genetics@LifeLabs.com or 1-844-363-4357. In addition, our genetic counsellors proactively contact healthcare providers about abnormal results to provide clear next steps for their patients. Services are available from 8am-7pm EST in both English and French.

1.Dar et al. Am J Obstet Gynecol. Published online January 24, 2022. doi:10.1016/j.ajog.2022.01.019 ] 2. Pergament et al. Obstet Gynecol. 2014 Aug; 124(2 Pt 1):210-8. ] 3. Ryan et al. Fetal Diagn Ther. 2016; 40(3):219-23. ] 4. Dar et al. Am J Obstet Gynecol. Published online January 13, 2022. doi:10.1016/j.ajog.2022.01.002 ] 5. Norton et al. Perinatal and genetic outcomes associated with no call cfDNA results in 18, 497 pregnancies, SFMS's 40th Annual Pregnancy Meeting 2021. ] 6. Norwitz et al. J Clin Med. 2019 Jun; 8:937. ] 7. Hedriana et al. Prenat Diagn. 2020 Jun; 40(2):179-84. ] 8. Nicolaides et al. Fetal Diagn Ther. 2014; 35(3):212-7. ] 9. DiNonno et al. J Clin Med. 2019 Aug; 8(9);1311. ] 10. Internal data, Natera. ] 11. Stokowski et al. Prenat Diagn. 2020 Jun; 40(2):179-84. ] 12. Jones et al. Ultrasound Obstet Gynecol. 2018 Feb; 51(2):275-6. ] 13. Hooks et al. Prenat Diagn. 2014 May; 34(5):496-9. ] 14. Schmid et al. Fetal Diagn Ther. 2018; 44(4):299-304. ] 15. Palomaki et al. Genet Med. 2011 Nor; 13(11):913-20. ] 16. Palomaki et al. Genet Med. 2017 Nor; 19(5):890-901. ] 21. Bianchi et al. Direr, 120(1):57(7):1042-9. ] 20. Bianchi et al. Obstet Gynecol. 2012 May; 19(5):890-901. ] 21. Bianchi et al. NEMS 37th Annual Pregnancy Meeting 2017. ] 19. Sehnert et al. Clin Chem. 2011 Jun; 57(7):1042-9. ] 20. Bianchi et al. Obstet Gynecol. 2012 May; 19(5):890-901. ] 21. Bianchi et al. NET MIT AND AUGUNT Pregnancy Meeting 2017. ] 19. Sehnert et al. Ne Engl J Med. 2014 Feb; 370(9):799-808. ] 22. Verinata Health. Analytical validation of the Verifi prenatal test, 2012. ] 23. The commercial protocol is not validated. Illumina marketing materials cite Strinvasan et al. Am J Hum Genet. 2013 Feb; 92(2):167-76, which does not match the number of reads used in commercial trisomies across pregnancy. UpToDate. Accessed January 20, 2022. https://doi.org/10.1016/j.ajog.2022.01.036. ] 25. Maternal age-related risk for common fetal trisomies across pregnancy. UpToDate. Accessed January 20, 2022. https://www.uptodate.com/conte

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