The largest prospective NIPT study

Study Objective

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.
Aneuploidy detection

Demonstrated high performance in singleton pregnancies\(^1\)

- Included a large cohort of average-risk pregnancies\(^1\)
- Panorama™'s high sensitivity and specificity were maintained in real-world clinical practice

<table>
<thead>
<tr>
<th>Condition</th>
<th>Sensitivity</th>
<th>Specificity</th>
</tr>
</thead>
<tbody>
<tr>
<td>Trisomy 21</td>
<td>99.00%</td>
<td>99.96%</td>
</tr>
<tr>
<td>Trisomy 18</td>
<td>94.12%</td>
<td>99.98%</td>
</tr>
<tr>
<td>Trisomy 13</td>
<td>&gt;99.00%</td>
<td>99.98%</td>
</tr>
</tbody>
</table>

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\(^4, 25-29\)

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

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

Chromosome 22*

- Full A-D deletion (2.5+ Mb)
- Smaller, nested deletions (0.5+ Mb)

Focus of most NIP studies

Panorama™ is validated to also detect smaller deletions\(^5\)

**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™ was validated in SMART with high accuracy and high positive predictive value (PPV) for 22q11.2 deletions.\textsuperscript{1,4,5}

- >99.9% sensitivity for most common deletion size (2.5 Mb+)
- 83% sensitivity for full and nested deletions (0.5 Mb+)
- 53% PPV for full and nested deletions (0.5 Mb+)

Mb = Megabase pair

Importance of early, accurate 22q11.2 deletion screening\textsuperscript{11-14}

Perinatal interventions can improve quality of life of individuals affected by 22q11.2 deletion\textsuperscript{30-34}

- Hypocalcemia 50%
- Developmental delay & learning differences 70%-90%
- Immune deficiencies 77%
- Palatal abnormalities 69%
- Congenital heart defects 74%
- Schizophrenia in adulthood 25%

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

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

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Why choose Panorama™ NIPT?

**Advantages**

1. **Highest clinical PPVs, supported by Canadian data**
2. **Best-in-class sensitivity & specificity**
3. **Distinguishes between maternal and fetal DNA**
4. **Zygosity detection** (Identical/Fraternal twins)
5. **Highest individual fetal sex accuracy**

**Zygosity detection** (Identical/Fraternal twins)

**Triploidy detection** (Extra copy of each chromosome)

**Vanishing twin detection**

**Fetal fraction of each twin**

**Support by Canadian genetic counsellors at every step**

**Every result matters**

**Twins differentiation**

**Services included with Panorama™ NIPT:**

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

- **Convenient Sample Collection and Testing Within Canada**
  - Panorama™ NIPT samples can be collected at any LifeLabs patient service centre or national affiliates and testing is performed within Canada.

- **Genetic Counselling Support**
  - 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.

LifeLabsGenetics.com/Panorama

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