



# R445 non-invasive prenatal testing (NIPT) referral pathway audit

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## Introduction

- NIPT for Down's, Edward's and Patau's syndrome is now available as part of the Fetal Anomaly Screening Pathway (FASP) for women who have had a previous pregnancy with reported full trisomy of chromosomes 13, 18 or 21 (R445).
- We performed an audit of the R445 NIPT service at the Birmingham Women's laboratory, TDL Genetics & Health Service Laboratories and St George's Hospital to determine how many patients were referred in the first 6 months of the service going live (April 2024-October 2024) and what the outcomes of this testing were.

## Methods

The questions we wanted to answer from the R445 NIPT audit included:

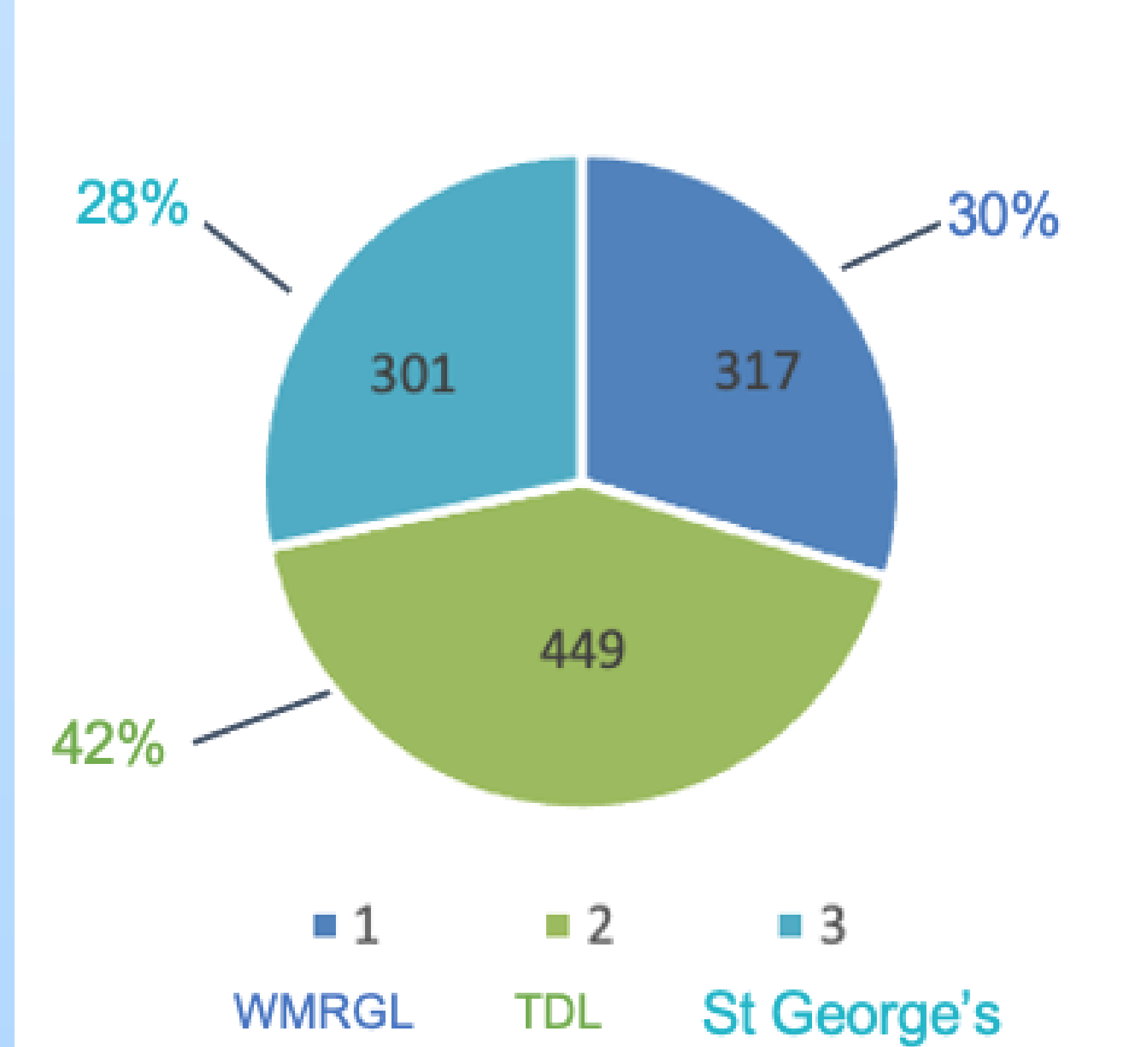
- How many women have accessed this service since it went live in April 2024. How many women do we estimate will use this service in 12 months?
- How many of these women had a high chance NIPT result and for which trisomy?
- What trisomy was their previously affected pregnancy?
- Did these women go on to have invasive testing and what was the outcome of this?
- What was the average maternal age and gestation at sampling?

## Results

Results were obtained using Illumina VeriSeq NIPT solution 2 or IONA@Nx test prenatal test.

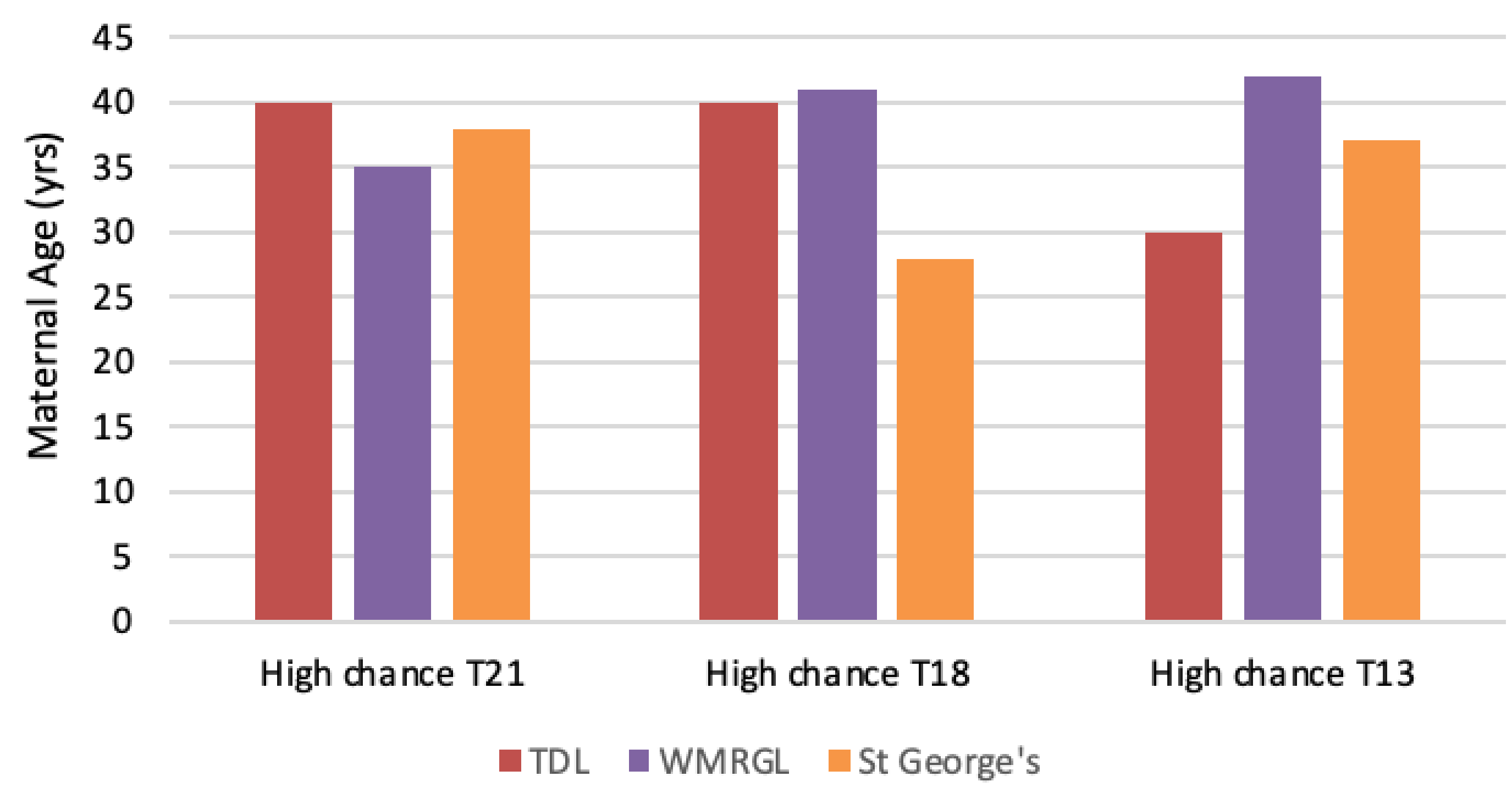
- Between April and October 2024, a total of 1,067 referrals for **R445 NIPT** were received nationally, with similar referral patterns seen at all three laboratories (*Figure 1*). We estimate receiving approx. 2,200 samples by April 2025.
- Among women referred through the R445 pathway, **1.78%** received a high-chance NIPT result, with 42% of these cases involving the same trisomy as in their previous pregnancy (*Table 1*).
- In cases where we had access to the invasive test results (CVS or Amnio), the NIPT high-risk findings were confirmed in **92%** (*Table 1*).
- The average maternal age of the women referred for testing was **33 years**. Those with a high chance NIPT result had an average age of **37 years** (*Figure 2a*).
- The average gestational age at referral was **11-12 weeks** (*Figure 2b*).

Number of R445 NIPT referrals



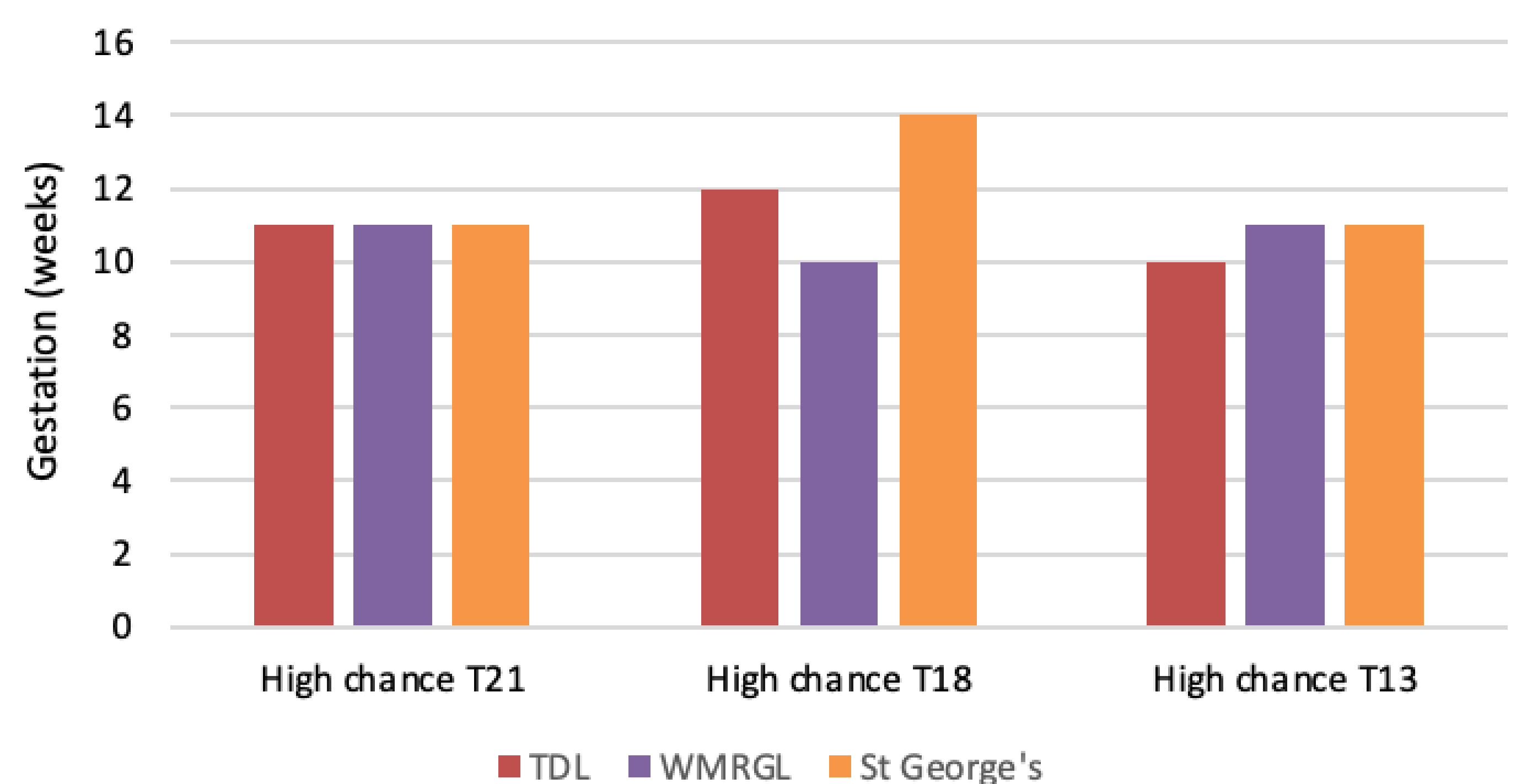
**Fig. 1.** The number of referrals received for R445 NIPT between April and October 2024, categorized by the referring laboratory.

High Chance NIPT result Vs Maternal Age



**Fig 2a.** The high-chance NIPT result alongside the mother's maternal age as classified by the referring laboratory.

High Chance NIPT result Vs Gestation



**Fig 2b.** The high-chance NIPT result alongside the gestational age as classified by the referring laboratory.

Referral reason	NIPT result		
	High Chance T21	High Chance T18	High Chance T13
Prev T21	8	3	3
Prev T18	5	0	0
Prev T13	0	0	0
<b>Total</b>	<b>13 (1.22%)</b>	<b>3 (0.28%)</b>	<b>3 (0.28%)</b>
<b>Follow up testing performed</b>	<b>10</b>	<b>1</b>	<b>2</b>
<b>Trisomy confirmed</b>	<b>10 (100%)</b>	<b>1 (100%)</b>	<b>1 (50%)</b>

**Table 1:** Testing outcomes. 19 cases (1.78% of total referrals) referred due to a previous affected pregnancy were given a high chance NIPT result.