

# Non-Invasive Prenatal Testing in Wales: Impact on Women's Choices

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Annual CARIS Meeting



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# Non-Invasive Prenatal Test

## Background and Context

- Estimated 60% of women choose Down's syndrome screening
- Decision to accept screening offer influenced number of factors including:
  - No change to commitment to pregnancy
  - Risk of pregnancy loss to confirm chromosomal condition
- NIPT is a blood test that can identify DNA from the placenta
- Estimated specificity of 99.9% for Down's syndrome, Edward's syndrome and Patau's syndrome
- Low chance NIPT = Highly unlikely to have the chromosomal condition

# Non-Invasive Prenatal Test

## Antenatal Screening Wales

- UK National Screening Committee recommended NIPT as second screening test
- To be offered following an initial higher chance primary screening result
- Wales was the first UK nation to implement in 2018
- Multi-agency project board – NIPT Evaluation Board
  - Including patient voice through support organisations
- Extensive preparatory work across all Health Boards
  - Information for health professionals
  - Resources for patients and families

# Non-Invasive Prenatal Test

## Antenatal Screening Wales

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- Additional step within the screening pathway
- How would this impact upon:
  1. Women's choices during their pregnancy
  2. Number of invasive diagnostic tests undertaken
  3. Performance of NIPT within real-life screening pathway

Study Objective: *To evaluate the implementation of non-invasive prenatal testing (NIPT) on pregnant women's choices in a national NHS antenatal screening programme for Down's syndrome, Edwards' syndrome and Patau's syndrome*

- Study focused on higher chance cohort in comparison to wider evaluation from ASW

# Study methods

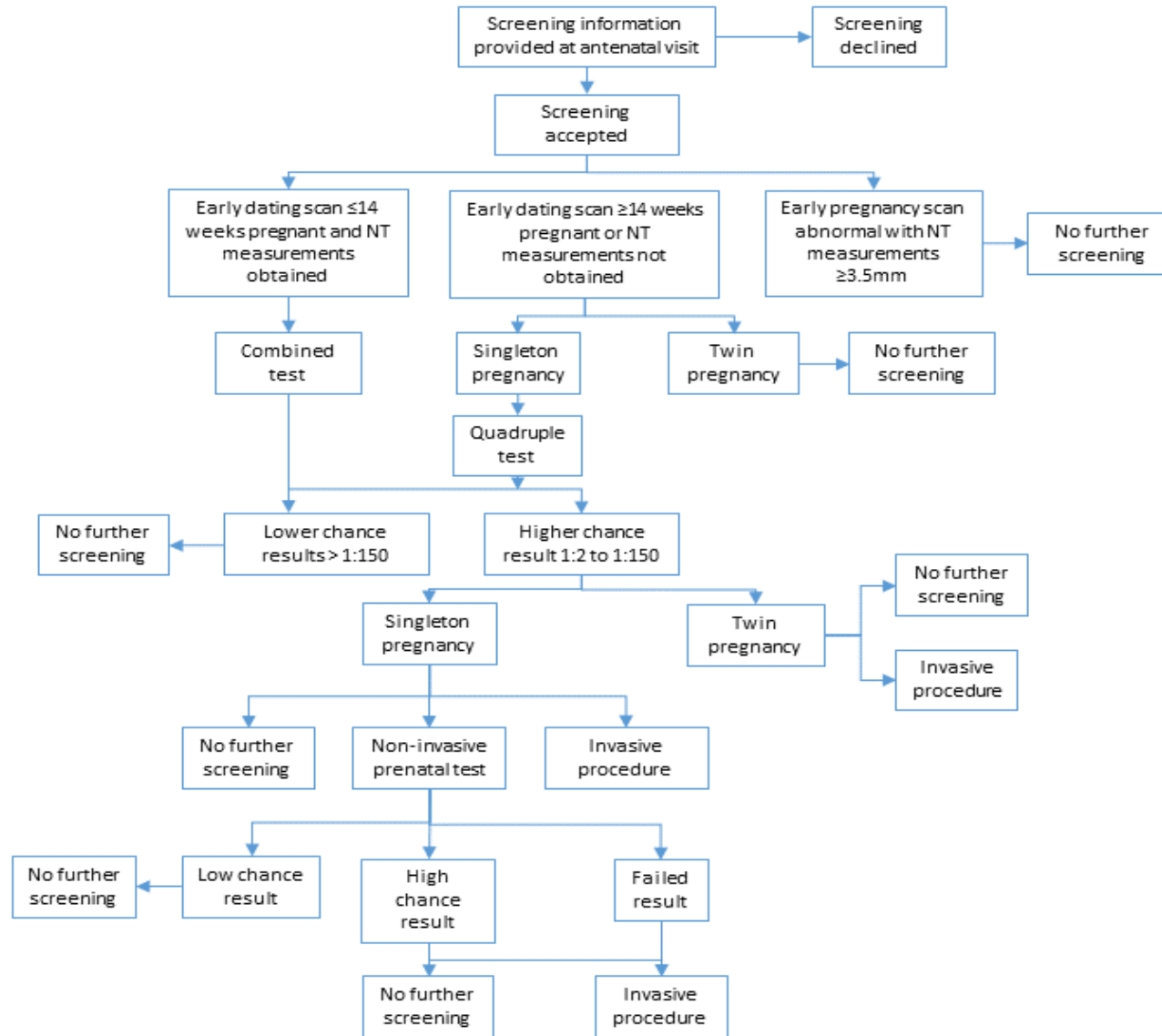
## Timeline and Data Collection



### Study cohort:

- Women with singleton pregnancy
  - Higher chance combined or quadruple screening result
  - All Wales
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- Study exclusion criteria
    - NIPT without following ASW pathway
    - Abnormal early pregnancy dating scan – entered diagnostic pathway

# Screening pathway decision tree



# NIPT evaluation

## Measures

- Women's choices through the screening pathway
  - Uptake of NIPT
  - Invasive Procedures
  - No further testing
- Performance of NIPT
  - Turnaround times
  - Failure rate
- Invasive procedures
  - Pre and post implementation
- Live birth rate for Down's syndrome
  - Pre and post implementation

# NIPT evaluation

## Data sources

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- CARIS
- 30 months of data from 30 April 2018 to 30 September 2020
- Variables included:
  - Initial screening result
  - NIPT result – Low chance; High chance or Failed
  - Invasive test: Amniocentesis or Chorionic Villus Sampling
  - Karyotyping
  - Pregnancy outcome
  - Down's syndrome diagnosis



# Results

## Women's choice following higher chance combined/quadruple test

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- 1273 higher chance screening results for singleton pregnancies
  - 1015 combined screening
  - 258 quadruple screening
- Higher chance results for singleton pregnancy:
  - 14% (174) women chose not to undertake any further testing
  - 84% (1073) chose NIPT
  - 2% (26) chose an invasive test
- 16 higher chance screening results for twin pregnancies:
  - NIPT not offered during study period
  - No invasive procedures

# Results

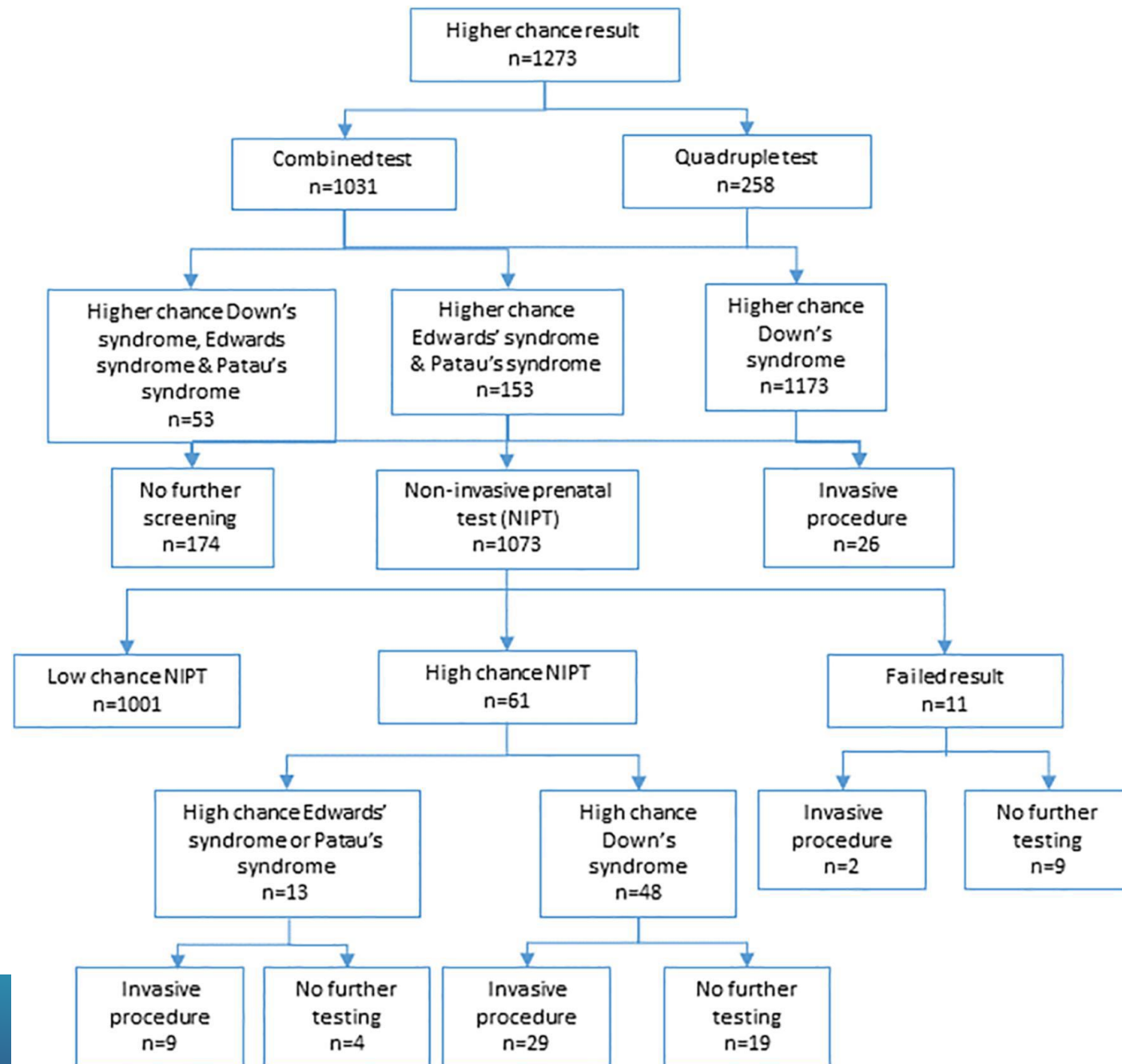
## NIPT outcomes

- NIPT result
  - 93% (1001) low chance result
  - 6% (61) high chance result
  - 1% (11) failed result
- High chance NIPT for Down's syndrome result (n=48)
- High chance NIPT for Edward's or Patau's syndrome (n=13)
- NIPT – no result (n=11)
  - 38% (2) chose an invasive test
  - 62% (9) chose no further testing

# Results

## Women's choice following NIPT

- High chance NIPT for Down's syndrome result (n=48):
  - 60% chose an invasive procedure
  - 40% chose no further testing
  - 63% (30) chose termination of pregnancy following invasive procedure/anomalies detected
  - 33% (16) continued to live birth with 12 confirmed as Down's syndrome post delivery
- High chance NIPT for Edward's or Patau's syndrome (n=13)
  - 70% chose invasive test
  - 30% no further testing



# Results

## Invasive procedures

- Nine-fold reduction in invasive procedures
- 26 per year over the evaluation period
- 229 per year prior to evaluation period
- Absolute annual reduction of 203 invasive procedures
- 97% amniocentesis & 3% CVS

# NIPT evaluation

## Live birth for Down's syndrome

- No difference in live birth rates during 2015 to 2020
- Included women who had not entered the screening pathway

Year	2015	2016	2017	2018	2019	2020
Live births with Down's syndrome	43	46	27	33	39	32
Total live births	33,119	33,004	32,236	31,329	29,728	28,781
Live birth rate per 10,000 with 95% confidence intervals	12.98 (9.10-16.86)	13.94 (9.91-17.97)	8.38 (5.22-11.54)	10.53 (6.94-14.13)	13.12 (9.00-17.24)	11.12 (7.27-14.97)

# NIPT evaluation

## Conclusions

- **NIPT** as a **contingent screening test** was **highly acceptable** to women with the majority (86%) opting for NIPT following an initial higher chance screening test.
- **Invasive procedures reduced** by nearly 9 fold following introduction of NIPT screening reducing the potential risk of associated pregnancy loss
- Women continued to **make varied choices** throughout the screening pathway and regarding continuing their pregnancy, with the **live birth rate for babies with Down's syndrome remaining unchanged**

# NIPT evaluation

## Reflections

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- Improved pathway for women
- Women made varied choices throughout the pathway
- High uptake of NIPT as contingent test
- Live birth rate findings consistent with international evidence
- Very small numbers for Edward's syndrome and Patau's syndrome
- Positive to have data in peer-reviewed publication



# NIPT evaluation

## Next Steps

- Implementation of the offer of NIPT in a twin pregnancy in June 2021
- Sharing learning and data – discussions with Scotland and England
- Exploring uptake of initial screening offer
- Qualitative insights into women's choices

## • Any Questions?