Public Health England

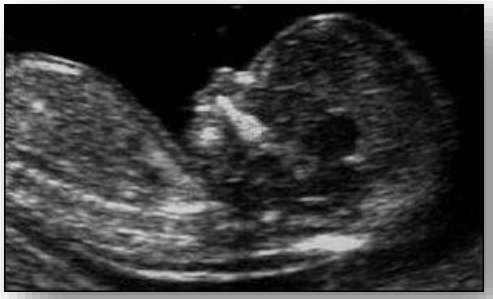
NHS

ACGS Summer Scientific Meeting 2017
Fetal Anomaly Screening Programme

Public Health England leads the NHS Screening Programmes

Fetal Anomaly Screening Programme (FASP)

The Fetal Anomaly Screening Programme (FASP) offers screening to all eligible pregnant women in England to assess the risk of the baby being born with Down's, and/or Edwards'/ Patau's syndromes or a number of fetal anomalies (structural abnormalities of the developing fetus).

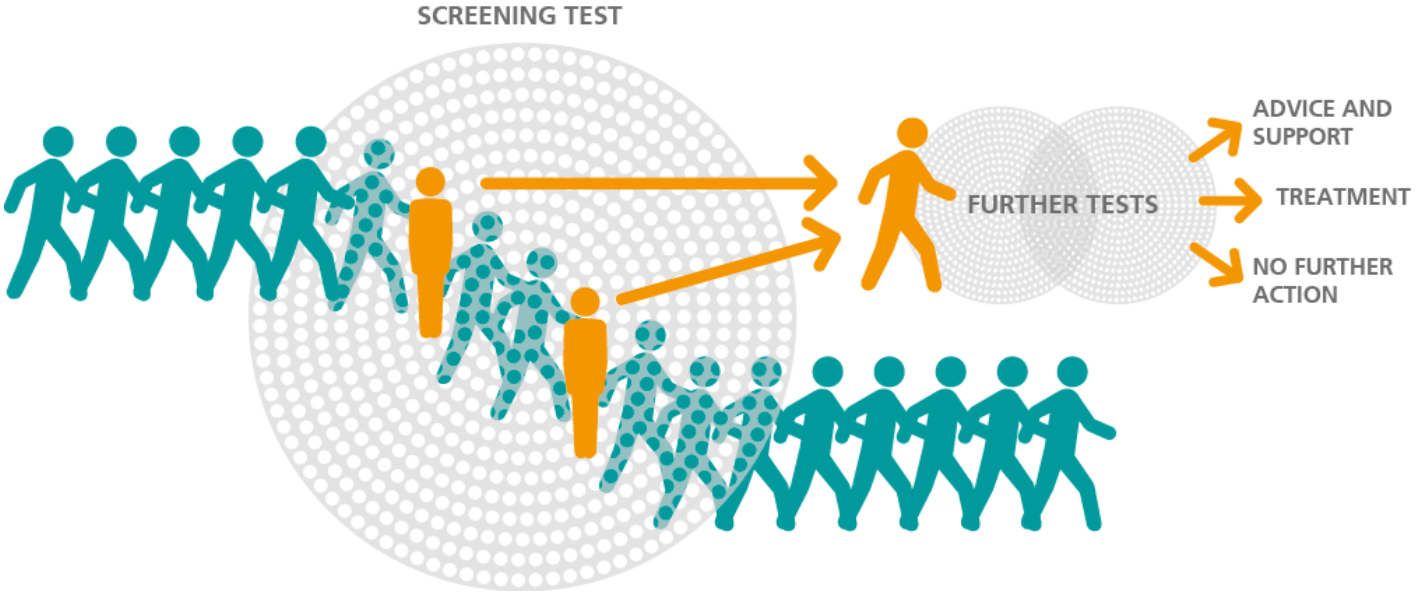


What is screening?

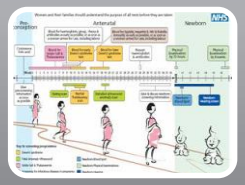
Screening can be thought of as putting people (who accept an offer to be screened) into a sieve to identify those who need further investigation

Those caught in the sieve are at increased risk of the condition being screened for and will be offered further tests

Those who pass through the sieve are discharged from the screen (this does not mean they have no risk but the same risk as the rest of the population)



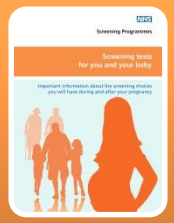
Screening is not just a test - its a pathway



A screening programme supports people throughout the process, from invitation to referral (of anyone who is found to have a particular condition) for treatment and advice

Generic screening pathway

1. Identify the people to invite
2. Discuss and offer screening
3. Carry out screening test
4. Provide and discuss results /options
5. Carry out diagnostic/ confirmatory test if required
6. Provide and discuss results /options
7. Offer advice/ treatment

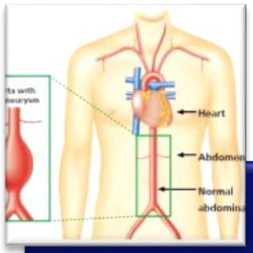


Screening can involve difficult choices, people offered screening should get the information they need in a format they can understand

Screening programmes

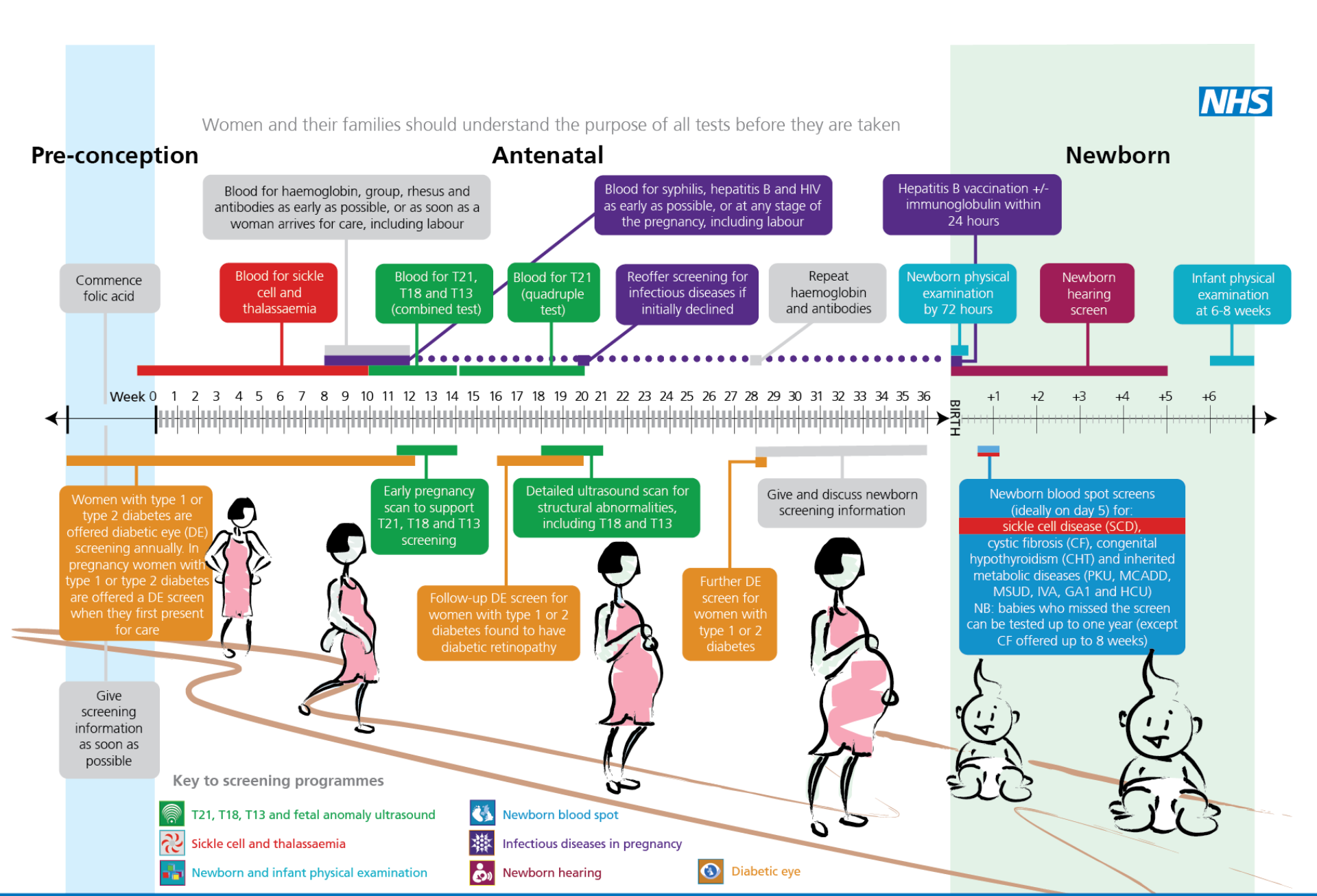
Antenatal/newborn

- Infectious diseases in pregnancy
- Sickle cell and thalassaemia
- Fetal anomaly
- Newborn and infant physical examination
- Newborn hearing
- Newborn blood spot



Young person /adult

- Diabetic eye
- Abdominal aortic aneurysm
- Bowel cancer
- Breast
- Cervical



Antenatal and Newborn Screening Timeline - optimum times for testing

Version 8.1, March 2016, Gateway ref: 2014696, Public Health England leads the NHS Screening Programmes

www.gov.uk/topic/population-screening-programmes

What Does FASP Screen for:

First Trimester Combined screening	18^{+0} to 20^{+6} week scan
Down's syndrome	Anencephaly
Edwards' syndrome	Open Spina Bifida
Patau's syndrome	Lethal Skeletal Dysplasia
	Cleft Lip
Second Trimester Quadruple Screening	Serious Cardiac Anomalies
Down's syndrome	Diaphragmatic Hernia
	Gastroschisis
	Exomphalos
	Bilateral Renal Agenesis
	Edwards' and Patau's syndromes

Fetal Anomaly: Down`s, Edwards` and Patau's Syndromes

Ultrasound & maternal blood tests are offered to assess the chance of a fetus having Down`s, Edwards` or Patau`s syndrome

In 2015/16, 13,920 women were told that there was a higher chance that their baby might be affected

- **All eligible pregnant women in England should be offered a screening test for Down`s, Edwards` and Patau`s syndromes that meets national standards**
- **First trimester screening is the test of choice for singleton and twin pregnancies**
- **The quadruple test should be offered to women who present later in pregnancy or if the nuchal translucency (NT) cannot be measured at first trimester screening**
- **The quadruple test screens for Down`s syndrome only**
- **All providers are expected to share data with: FASP, QA, SITs and NCARDRs**
- **Some women may choose not to be screened at all, it is important this choice is respected**

18^{+0} to 20^{+6} week fetal anomaly ultrasound scan

- **All eligible pregnant women in England should be offered an ultrasound scan between 18^{+0} to 20^{+6} weeks**
- **The purpose of the scan is to identify any problems in the structural development of the baby**
- **Screening for Edwards` and Patau`s syndromes is offered as part of the fetal anomaly scan for those women booking later in pregnancy or when measurement of the NT was not possible**
- **Women should be made aware of the limitations of fetal anomaly screening**
- **All providers are expected to share data with: FASP, QA, SITs and NCARDRs**
- **Some women may choose not to be screened at all, it is important this choice is respected.**

Core Remit of the UK NSC

Founded in 1996 the UK NSC is recognised in the NHS Constitution as the source of expert advice on screening

The UK NSC advises ministers and the NHS in all four UK countries on all aspects of screening policy

Reviews based on latest research evidence & international criteria & informed by multi-disciplinary groups including professionals and patients

Programmes directed through standards, protocols & monitoring and supported with high quality information to the public and professionals

Evidence reviews

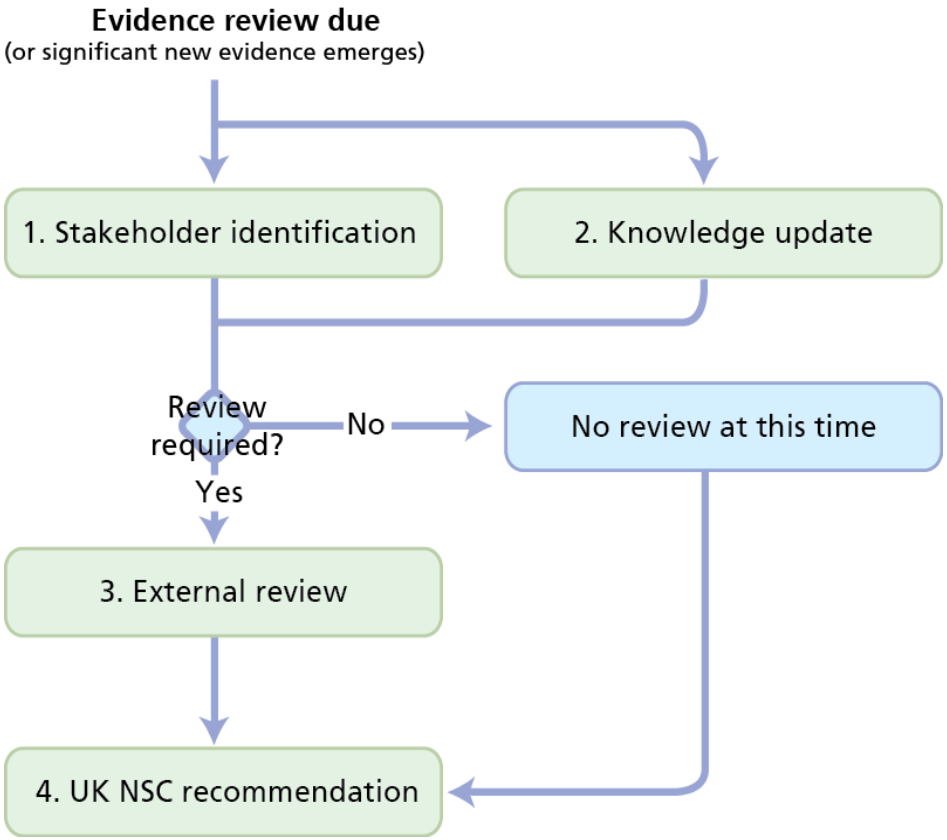
Over 100 topics are reviewed on a regular basis (approx 3 yearly)

Reviews are carried out against the UK NSC's internationally-recognised criteria; these cover the:

- **condition**
- **test**
- **treatment**
- **screening programme**

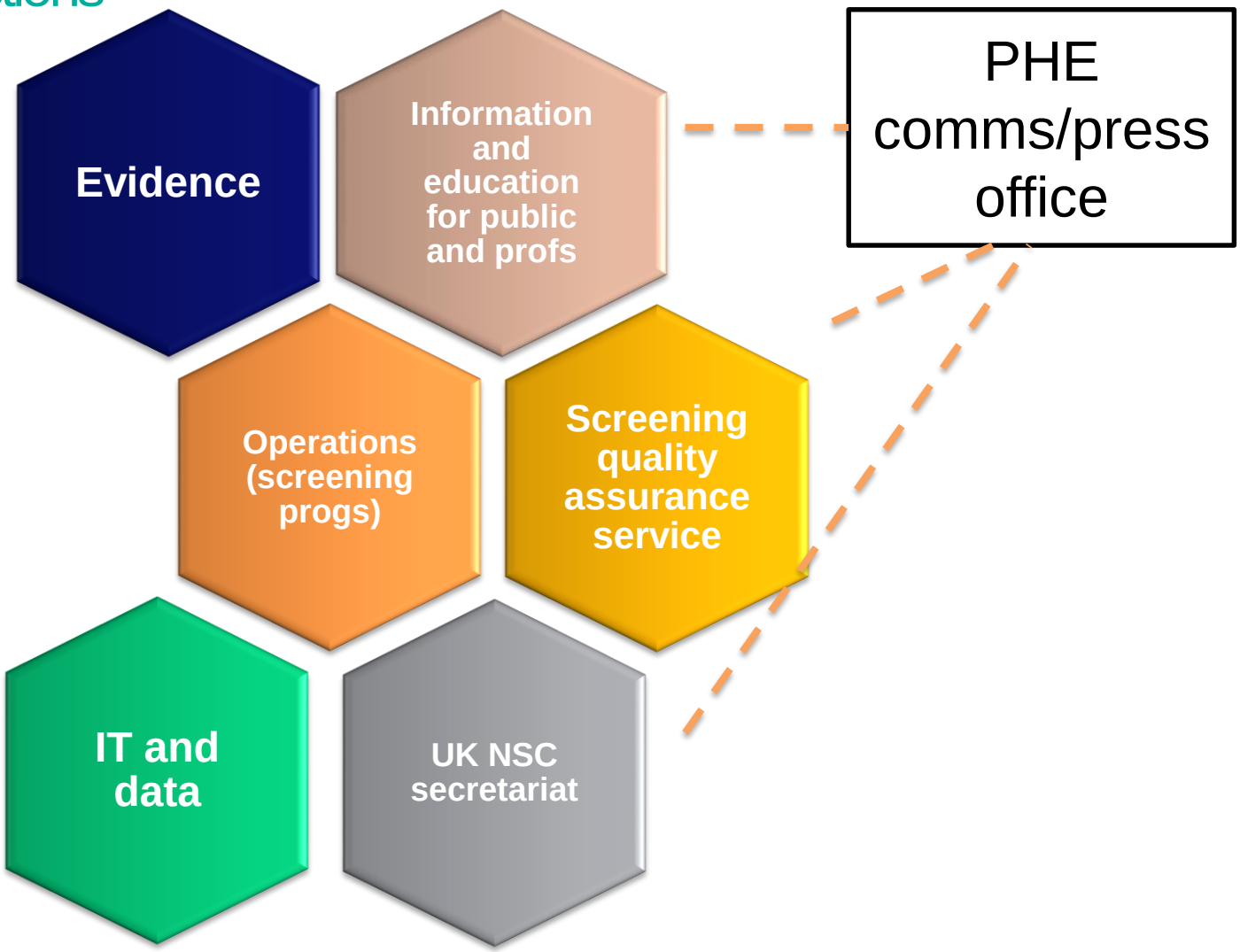
Draft reviews go out to public consultation for 3 months

See legacy.screening.nhs.uk/screening-recommendations.php



PHE (Health Improvement Directorate)

Screening functions



UK NSC non-invasive prenatal testing (NIPT) recommendation

The UK NSC recommended an evaluative implementation of NIPT to assess what impact it would have on the existing NHS Fetal Anomaly Screening Programme.

Pregnant women are already offered a screening test for Down's syndrome, Edwards' syndrome and Patau's syndrome from 10-14 weeks of pregnancy (the combined test, involving an ultrasound scan and blood test), or a screening test for Down's syndrome only (the quadruple test, involving a blood test alone) if booking between 14-20 weeks.

If the screening test shows that the chance of having a baby with Down's, Edwards' and Patau's syndromes is higher than 1 in 150, this is called a higher-risk result. Currently, women who have a higher risk result have the option of having an invasive diagnostic test (amniocentesis or CVS).

The proposed change is for Non-Invasive Prenatal Testing to be offered to women who are deemed at higher risk following the current primary screen. NIPT is not diagnostic and an invasive diagnostic test is still required to receive a definitive diagnosis.

Key findings supporting the UK NSC recommendation

- an invasive diagnostic test carries a small risk of miscarriage. The evidence suggests that NIPT will reduce the number of women being offered an invasive test
- however, while we know that the accuracy of NIPT is very good, we don't yet know how it will perform in an NHS screening programme pathway
- for women who choose to have NIPT, this will add in an extra step in the screening programme. The impact of this, and the choices women make at different points in the pathway, is something that we hope to gain a better understanding of through further research
- a recommendation has therefore been made to evaluate the introduction of non-invasive prenatal testing (NIPT) to Down's syndrome screening. This will include scientific, ethical and user input to better understand the impact on women, their partners and the screening programme around the offer of cfDNA or invasive testing following a screening test result where:
  - » the screening test risk score for trisomy 21 (T21) is greater than or equal to 1 in 150
  - » the combined test risk score for trisomy 18 (T18) and trisomy 13 (T13) is greater than or equal to 1 in 150

Planning the evaluative roll out

NIPT Project Board

Key Questions:

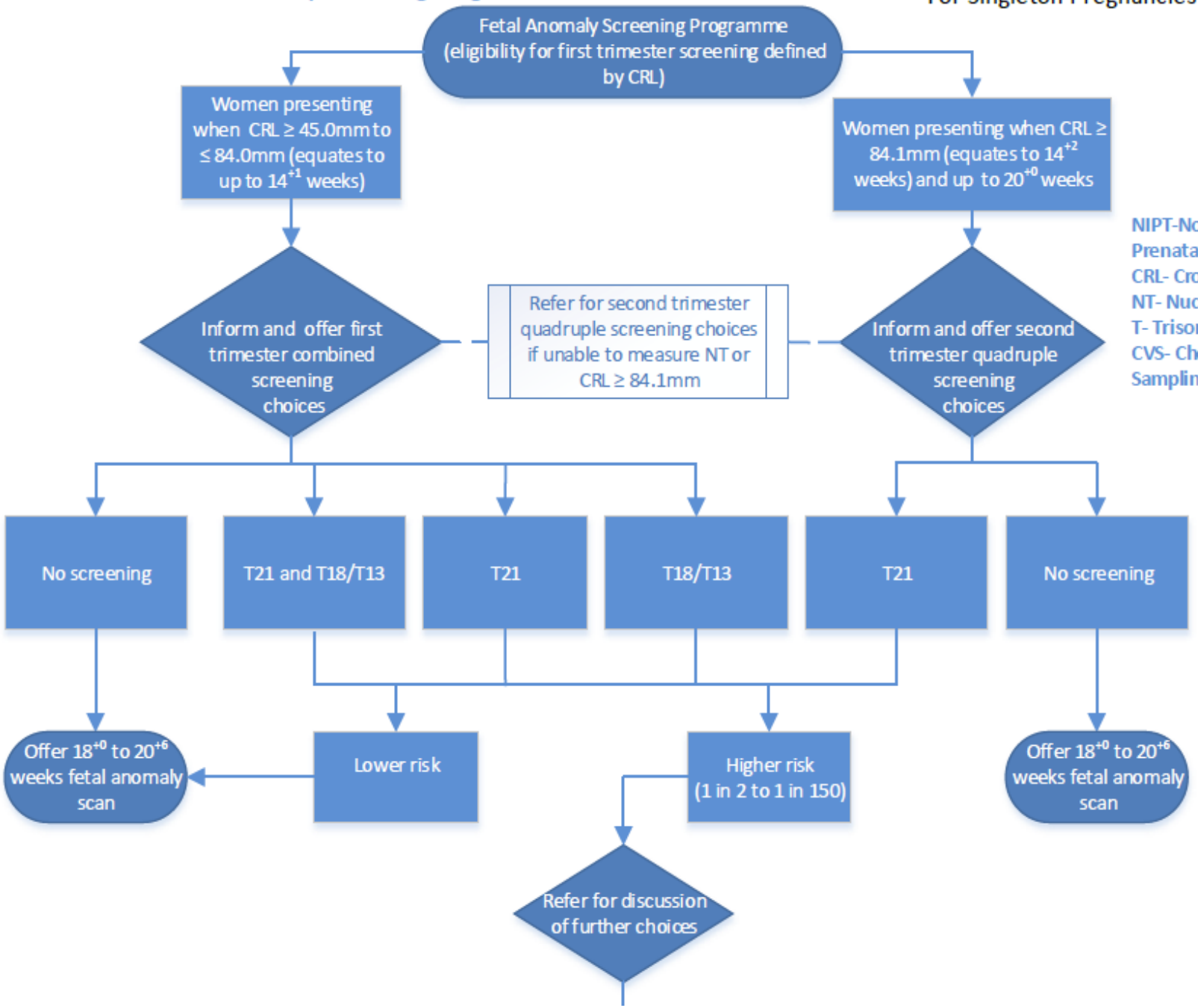
- what is the test performance for T18/T13?
- what is the test failure (“no result”) rate?
- what do women choose to do?
- what is the impact on the current screening pathway?

4 working groups:

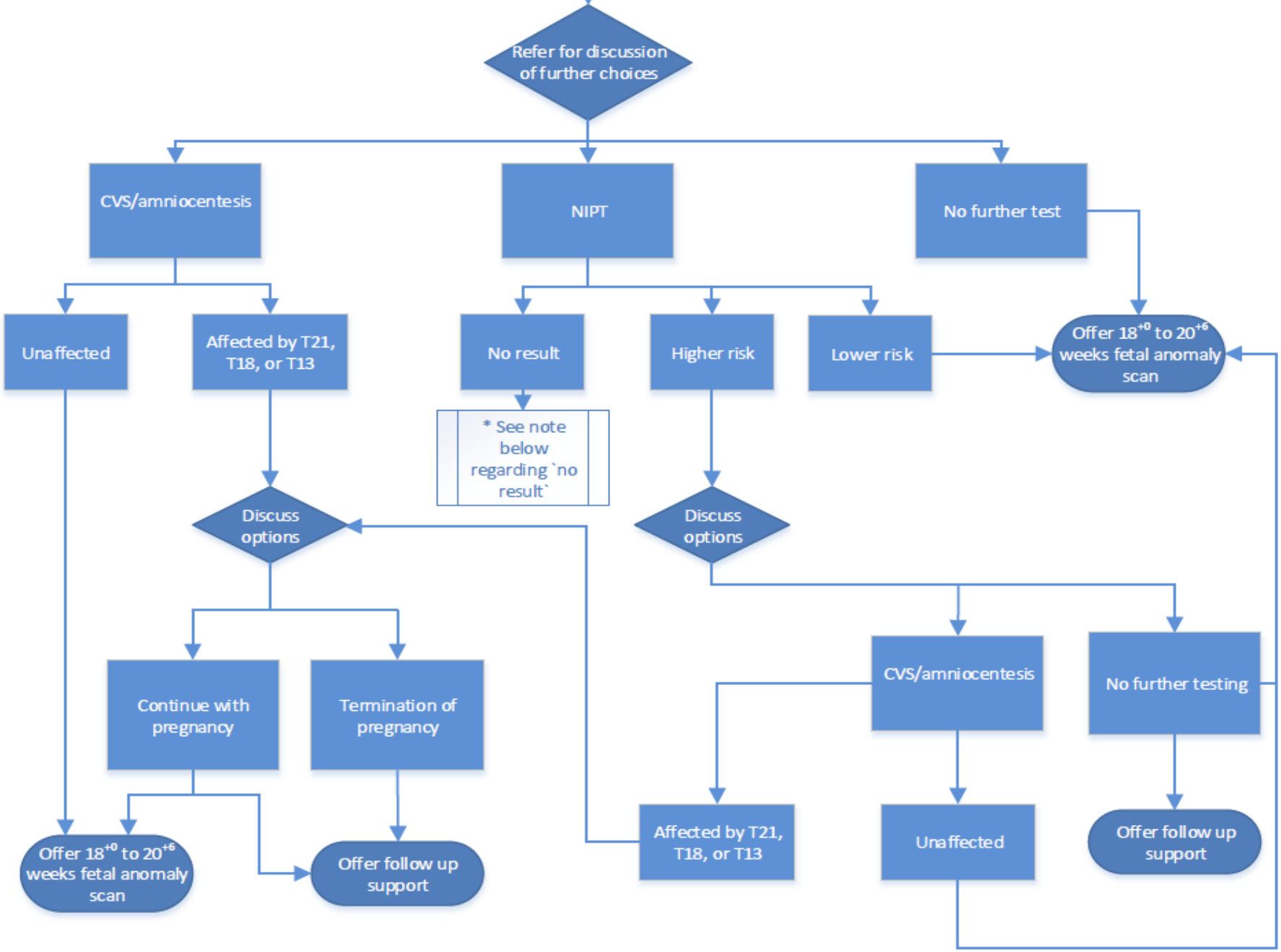
- Information. Education & Training
- Clinical & Laboratory
- Data – surveillance and QA
- Procurement of laboratory services

Additional option of Non Invasive Prenatal Testing as part of the NHS Fetal Anomaly Screening Programme

Down's (T21), Edwards' (T18) and Patau's (T13)
For Singleton Pregnancies Only



NIPT-Non Invasive Prenatal Testing
CRL- Crown Rump Length
NT- Nuchal Translucency
T- Trisomy
CVS- Chorionic Villus Sampling



* In cases where `*no result*` obtained from initial NIPT, offer of **one** further NIPT, CVS/amniocentesis or no further testing to be made. Please return to relevant points in the pathway. In cases where `*no result*` obtained a second time, follow `***Higher risk***` NIPT result.

NIPT

"Supporting personalised informed choice"

Information:

- revised STFYAYB – focus groups soon
- updated information explaining risk – work with Prof David Spiegelhalter , Prof Dave Wright & NHS Choices – animations
- revised resource cards for midwives
- parent information leaflets for women with a higher chance of Down's and Edward's/Patau's syndromes
- public facing film video for women about the screening pathway and the 3 conditions
- updated information on the 3 conditions: Down's; Edwards' and Patau's in all documents and website content

Working with key stakeholders – Down's syndrome association (DSA); Support for families with trisomy 18/13 (SOFT); Antenatal results & choices (ARC);Down's syndrome research foundation (DSRF)

NIPT (2)

Education & Training - 2 day training events across England

Day 1 - Information Day:

- practical aspects of NIPT
- screening pathway & eligibility
- results and further options

Day 2 – supporting personalised informed choice

- delivered in collaboration with DSA;SOFT; ARC
- what information to give
- how to support discussion
- women and parent stories

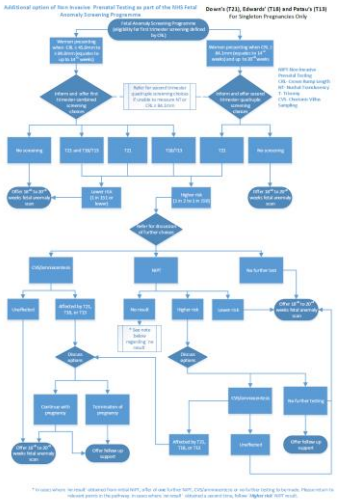
NIPT (3)

Data:

- real time evaluative data (surveillance)
- longer tem quality assurance (DQASS type data)
- standards
- KPIs

Clinical & Laboratory:

- pathway
- laboratory procurement criteria document
- update handbooks
- service specification



NIPT (4)

Laboratory Procurement:

Working with NHS England to procure within the current

Genomics laboratory services reconfiguration

2 strands of work

- procuring laboratory services for NIPT testing as part of the FASP NHS screening pathway
- maternity pathway payment team & specialised commissioning team (laboratory costs for PND) within NHS England to clarify funding streams

Procurement timetable yet to be finalised but working to a plan to introduce the offer of NIPT as an additional option on the NHS screening pathway in 2018.

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Questions?