Northern Health | Western Health | the women's the royal women's hospital victoria australia | Mercy Health Care first | phn NORTH WESTERN MELBOURNE An Australian Government Initiative

Shared Maternity Care Collaborative Workshop 2023: Session 2

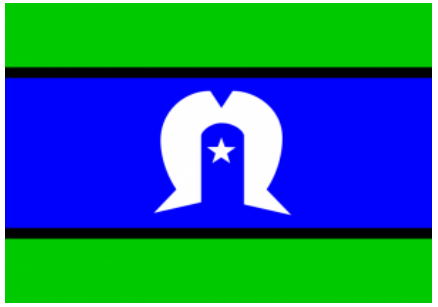
Monday 4 December 2023

The content in this session is valid at date of presentation

Acknowledgement of Country

North Western Melbourne Primary Health Network, the Royal Women's Hospital, Mercy Health, Northern Health and Western Health would like to acknowledge the Traditional Custodians of the land on which our work takes place, The Wurundjeri Woi Wurrung People, The Boon Wurrung People and The Wathaurong People.

We pay respects to Elders past, present and emerging as well as pay respects to any Aboriginal and Torres Strait Islander people in the session with us today.



Housekeeping – Zoom Webinar

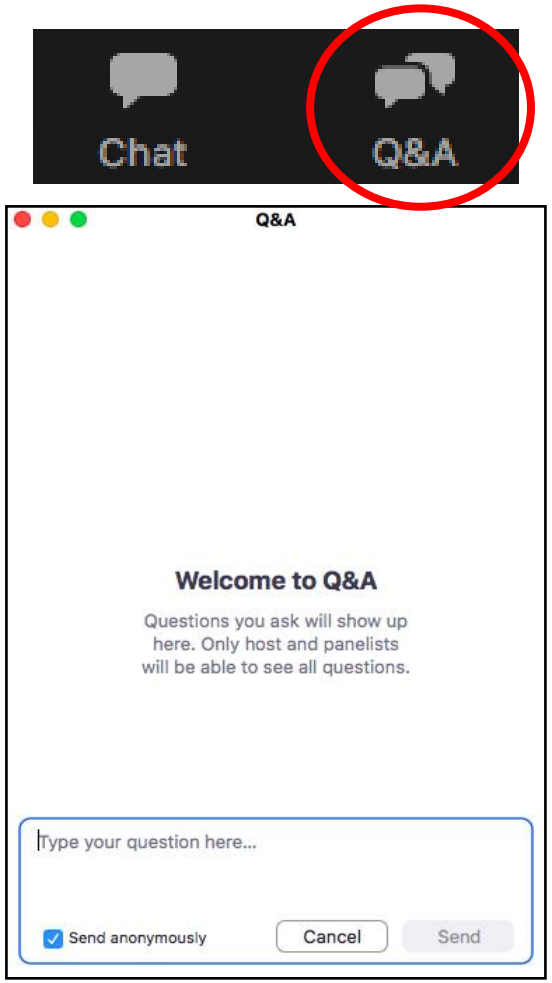
All attendees are muted

Please ask questions via the Q&A box only

Q&A will be at the end of the presentation

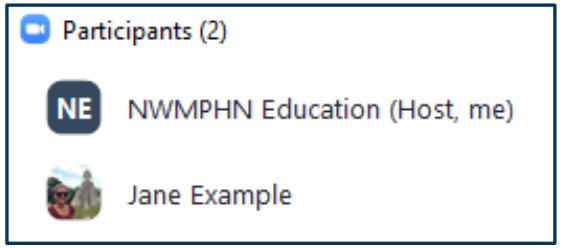
This session is being recorded, you will receive a link to this recording and copy of slides in post session correspondence.

Questions will be asked anonymously to protect your privacy



Housekeeping – Zoom Webinar

Please ensure you have joined the session using the same name as your event registration (or phone number, if you have dialled in)



NWMPHN uses Zoom's participant list to mark attendance and certificates and CPD will not be issued if we cannot confirm your attendance.

If you are not sure if your name matches, please send a Chat message to 'NWMPHN Education' to identify yourself.

Shared Maternity Care Collaborative

Northern Health

Primary Care Liaison officer Kirra McGaw – nh-primarycareliaison@nh.org.au
GP Liaison officer Dr Richard Sia – nh-primarycareliaison@nh.org.au

Mercy Health

Primary Care Liaison manager Caitlin Shaw – primarycare@mercy.com.au
Primary Care Liaison officer Sharon Tijssen – primarycare@mercy.com.au

The Royal Women's Hospital

Head of GP Liaison unit A/Prof Ines Rio – gp.liaison@thewomens.org.au
Primary Care Liaison officer Emily Lawson – gp.liaison@thewomens.org.au

Western Health

GP Advisor Jo Silva – gp@wh.org.au

Hospital Updates

Mercy Health

Mercy Health is now accepting HealthLink eReferrals.

Benefits of eReferral via HealthLink:

- eReferrals via HealthLink are already embedded into most GP practice software, making the transition simple and cost-free for most general practices.
- eReferral will auto-populate important patient data such as demographics, medical history and medications from the GP practice management software and guide them through the referral process, ensuring compliance to the Victorian state-wide referral criteria for each specialty.
- Referrers are sent a notification when their referral is received by Mercy Health.

For more information visit our Refer a patient website

Hospital Updates

Northern Health

Northern Health is now sending digital discharge summaries via HealthLink. Please ensure all your details are up to date with the National Health Services Directory (NHSD).

Northern Health's Medical Community Virtual Consult (MCVC) service provides Victorian GPs and Nurse Practitioners access to hospital-based specialist expertise to discuss complex patient management in the community. Specialties available include Paediatrics, Endocrinology and Rheumatology. More information is available at https://mcvc.nh.org.au/

Hospital Updates

The Royal Women's Hospital

The new Public Fertility Care Service is led by the Women's and Monash Health, with support from a range of partner health services across the state. The service provides access to comprehensive fertility treatment including:

- Genetic counselling
- Fertility preservation
- Fertility assessment and treatment

For more information on eligibility criteria and referral information please go to the Women's website:

https://www.thewomens.org.au/patients-visitors/clinics-and-services/fertility-genetics/public-fertility-services

Moderator

A/Prof Ines Rio – The Royal Women's Hospital

A/Prof Ines has extensive experience in many facets of health care. Ines is a Chairperson for the North Western Melbourne PHN, Director of Sexual Health Victoria, Head of the General Practice Liaison Unit and GP Obstetrician at The Royal Women's Hospital, General Practitioner North Richmond Community Health, member of the TGA advisory committee on vaccines, and newly appointment as Chief Medical Officer at Monash University and as member of the National Women's Health Advisory Council.

Ines is committed to quality, effective, efficient, equitable and integrated health care services and the central importance and role of general practice and primary care in this provision.

Speakers

A/Prof Lisa Hui FRANZCOG CMFM – Mercy Health

Prenatal screening and NIPT - what is the current standard of care?

Lisa is a maternal fetal medicine specialist with special interests in prenatal screening and diagnosis, particularly the use of cell-free DNA for the detection of fetal chromosome conditions. She is Director of Genetics at the Mercy Hospital for Women, which looks after patients throughout the northeast of Melbourne and regional Victoria.

She holds an MRFF investigator fellowship in the Genomics Health Futures Priority scheme, is a team leader in the Reproductive Epidemiology group at the Murdoch Children's Research Institute. She is an active member of the International Society for Prenatal Diagnosis and an associate editor of its official scientific journal *Prenatal Diagnosis*. She also sits on the editorial board of *Ultrasound in Obstetrics and Gynaecology* and is a regular invited speaker at international and national conferences.

Speakers

Dr Vicki Carson – The Royal Women's Hospital
Modes of delivery – is there such a thing as a 'normal' birth?

Vicki Carson is a general obstetrician with an interest in high-risk obstetrics. She works both privately at Frances Perry House and is leader of the Yellow Maternity Clinic at RWH.

Vicki has a strong interest in Indigenous Women's health working with the Reconciliation Action Plan at RWH and the Baggarook caseload team. She has a strong interest in improving outcomes for women with previous birth trauma. Recently Vicki was tasked with setting up the Covid ward at RWH and enjoys the team environment at RWH that comes with such initiatives. Vicki is the current medical lead for the introduction of the homebirth programme. Vicki sits on the federal RANZCOG council.

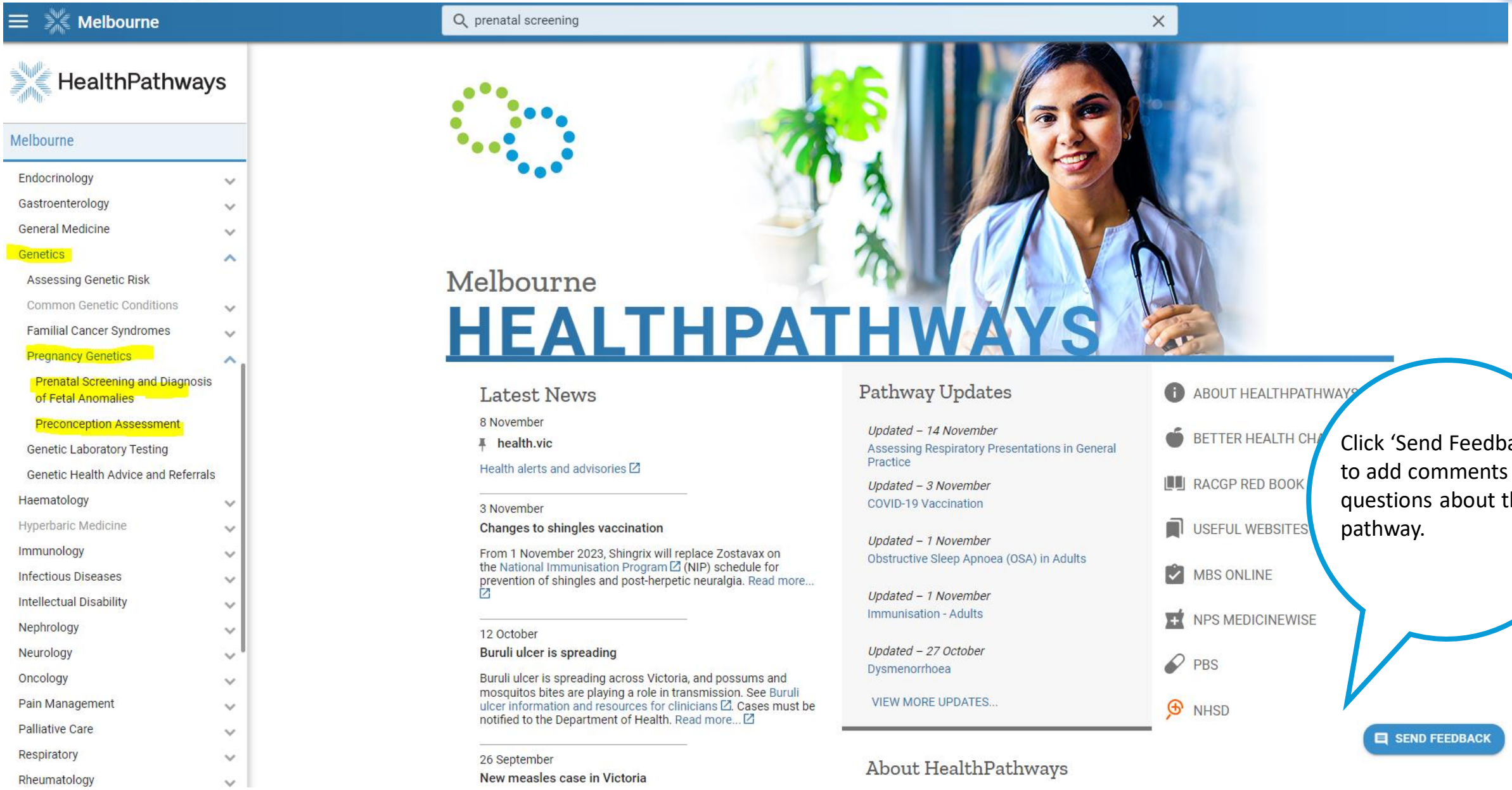
1

HealthPathways

Pathways are written by GP clinical editors with support from local GPs, hospital-based specialists and other subject matter experts

- clear and concise, evidence-based medical advice
- Reduce variation in care
- how to refer to the most appropriate hospital, community health service or allied health provider.
- what services are available to my patients

HealthPathways – Shared Maternity Care



Navigating HealthPathways – Shared Maternity Care

Shared Maternity Care Pathways Resources and Referral pages

Obstetrics

Preconception Assessment
Antenatal Care
Antenatal Care - First Consult
Antenatal - Second and Third Trimester Care
Early Pregnancy Bleeding
Pregnancy Bleeding
Recurrent Pregnancy Loss

Referrals and Resources

Acute Obstetric Referral or Admission (Same-day)
Non-acute Obstetric Referral (> 24 hours)
Early Pregnancy Assessment Service (EPAS) Pregnancy Booking
Fertility Specialised Referral
Acute Gynaecology Referral or Admission (Same-day)
Non-acute Gynaecology Referral (> 24 hours)
Pregnancy Booking
Radiology Services and Advice
Pregnancy Genetics
Prenatal Screening and Diagnosis of Fetal Anomalies
Genetic Laboratory Testing
Genetic Health Advice and Referrals

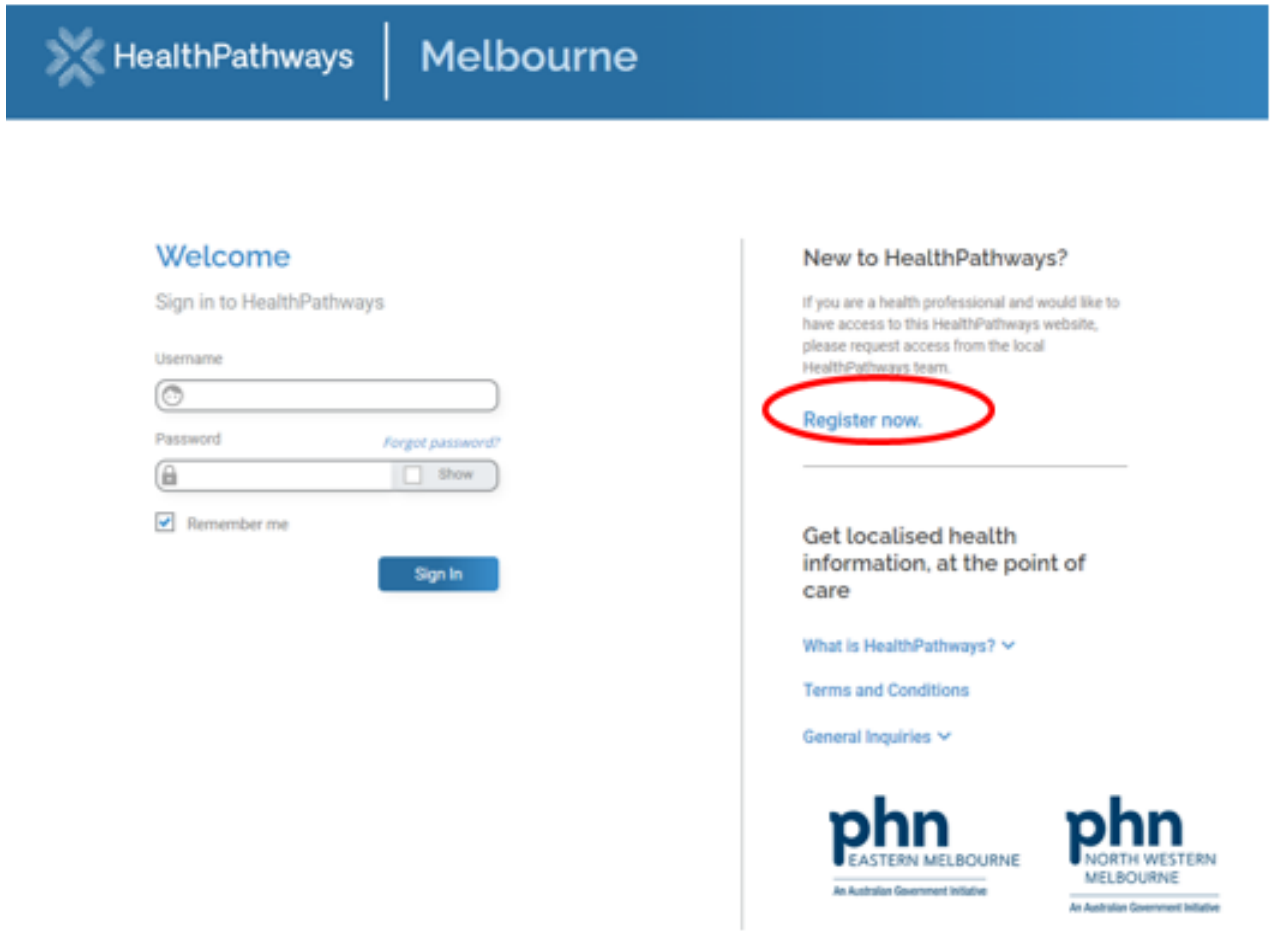
Pregnancy Medical Conditions

Anaemia in Pregnancy
Asthma in Pregnancy
Pregnancy Bleeding
Hypertension in Pregnancy and Postpartum
Nausea and Vomiting in Pregnancy
Obesity in Pregnancy and Pre-pregnancy
Skin Conditions (Rash and Itch) in Pregnancy
Thyroid Disease in Pregnancy
UTI and Asymptomatic Bacteriuria in Pregnancy
Varicella and Pregnancy
Diabetes in Pregnancy
  Hyperglycaemia in Pregnancy
  Pre-pregnancy Planning for Type 1 and Type 2 Diabetes
  Type 1 and Type 2 Diabetes and Pregnancy

Related and relevant LGBTIQA+ pages

LGBTIQA+ Fertility, Parenting, and Children
LGBTIQA+ Friendly Clinics
LGBTIQA+ Resources
Transgender Health and Gender Diversity Referral

Accessing HealthPathways: Go to melbourne.healthpathways.org.au



Register via QR code

info@healthpathwaysmelbourne.org.au

2

Prenatal screening and NIPT - what is the current standard of care?

A/Prof Lisa Hui
University of Melbourne
Mercy Hospital for Women
Murdoch Children's Research Institute
Northern Health

The Royal Australian and New Zealand College of Obstetricians and Gynaecologists

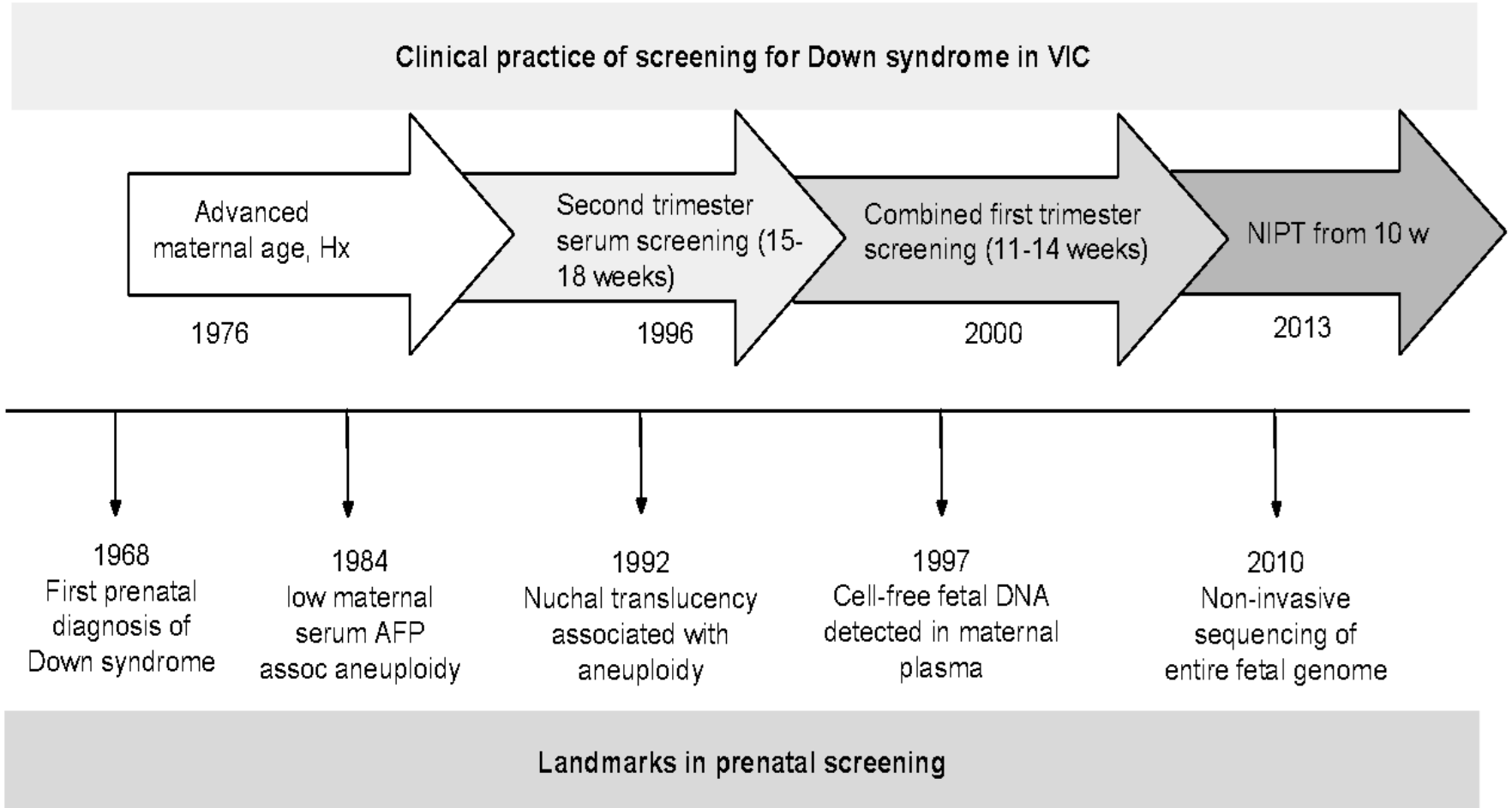
Excellence in Women's Health

HUMAN GENETICS SOCIETY OF AUSTRALASIA

Prenatal screening and diagnostic testing for fetal chromosomal and genetic conditions

- ALL pregnant women should be offered some form of screening **for trisomy 21** (RANZCOG, ACOG, ISPD)
- Accurate dates are required for correct performance
- Women with increased risk result offered genetic counselling and diagnostic testing with amniocentesis or chorionic villus sampling (CVS)

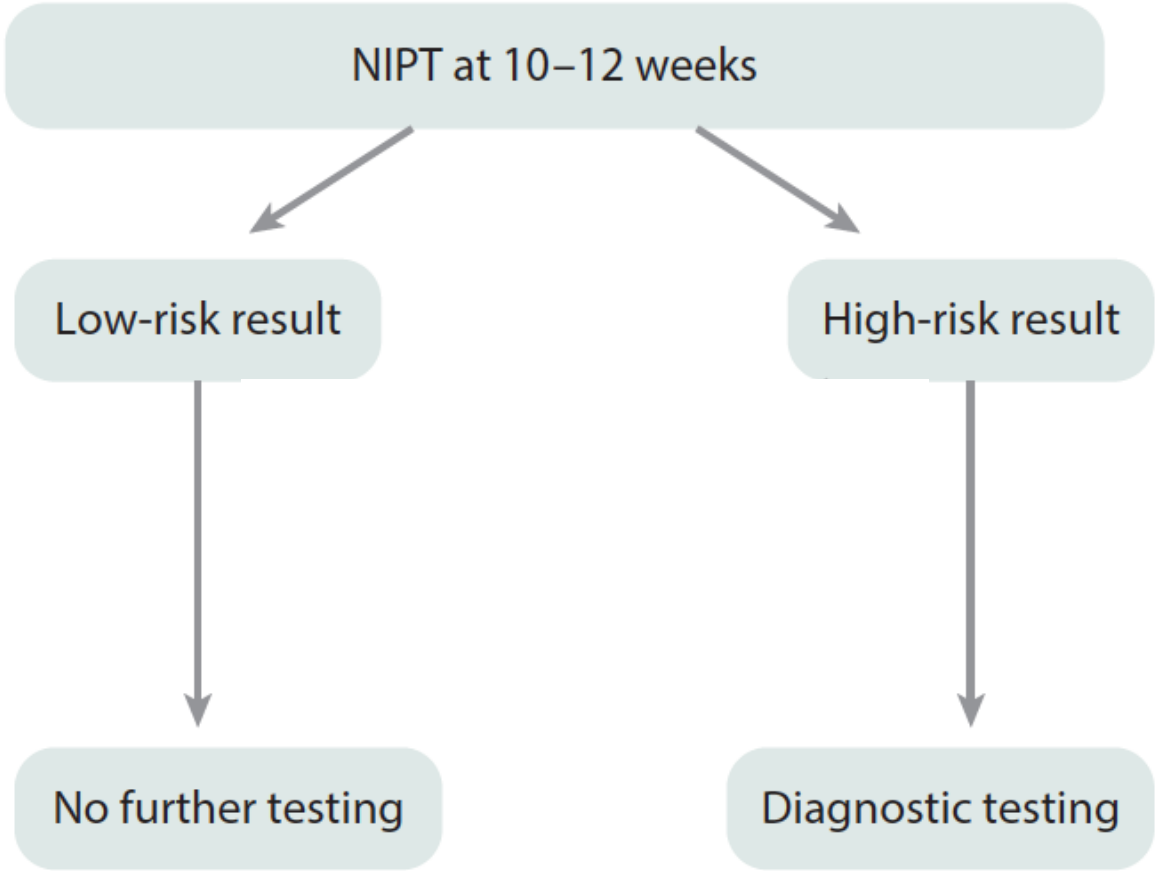
Timeline of prenatal screening for chromosome abnormalities



RANZCOG statement 2015

Recommendation 4	Grade and supporting references
Acceptable first-line screening tests for fetal chromosome abnormalities in the first trimester include either: a) combined first trimester screening with nuchal translucency and serum pregnancy-associated plasma protein A (PAPP-A) and beta human chorionic gonadotropin (βHCG) measurements OR b) cell-free DNA (cfDNA)-based screening. The choice of first line screening test will depend on local resources, patient demographics, and individual patient characteristics.	Consensus-based recommendation

NIPT at 12 weeks as first-line screen without first trimester scan



Provides higher detection rate of T21/18/13 than CFTS

No NT ultrasound provided

Received: 9 March 2022 | Revised: 21 June 2022 | Accepted: 10 August 2022

DOI: 10.1002/pd.6224

ORIGINAL ARTICLE

PRENATAL DIAGNOSIS WILEY

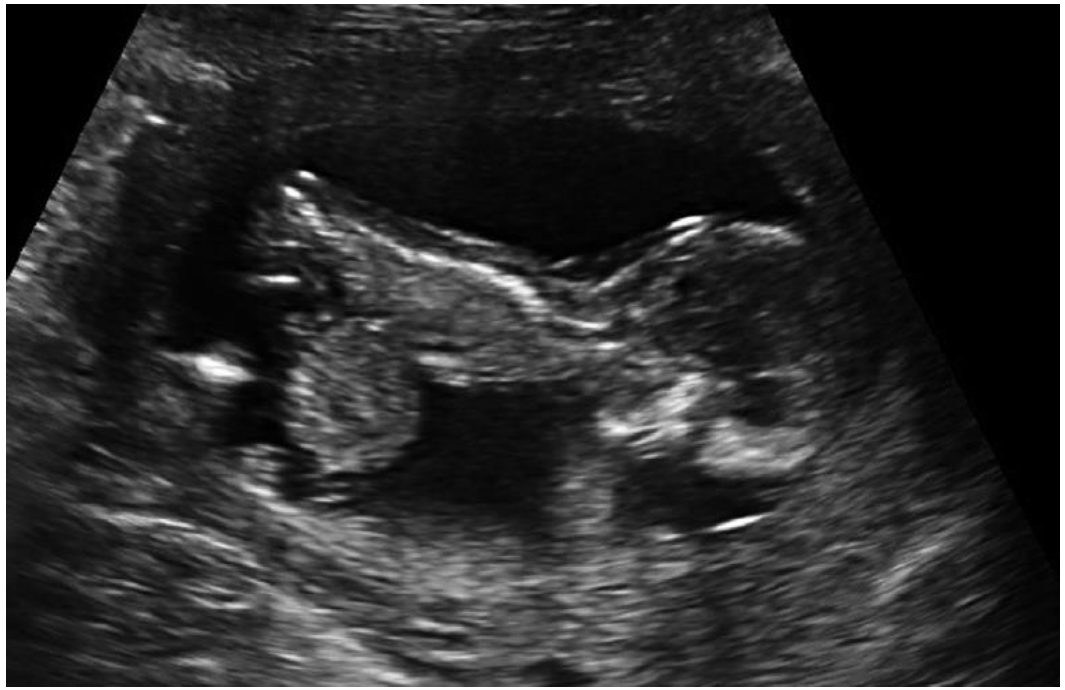
Timing of diagnosis of fetal structural abnormalities after the introduction of universal cell-free DNA in the absence of first-trimester anatomical screening

Francesca Bardi[1] | Anne Marie Beekhuis[1] | Marian K. Bakker[1] | Ayten Elvan-Taşpınar[1] | Caterina Maddalena Bilardo[2]

- In the Netherlands, NIPT has replaced the CFTS
- Audit in single tertiary centre showed that 56% of anomalies that should be detected in T1 are not diagnosed until T2
  - e.g anencephaly, gastroschisis, limb reduction defects, multiple anomalies
- Authors concluded that T1 anatomy US should be reintroduced
- A structured ultrasound protocol improves performance for T1 anatomy US

Karim JN. Ultrasound Obstet Gynecol. 2017 Oct;50(4):429-441.

First trimester ultrasound for anatomy



Edwards SFNM https://doi.org/10.1016/j.siny.2017.11.005

Table 1
Anatomical assessment checklist at 11–13^{+6}-week ultrasound scan.

Organ/ anatomical area	Present and/or normal
Head	Present
	Cranial bones
	Midline falx
	Choroid-plexus-filled ventricles
Neck	Normal appearance
	Nuchal translucency thickness (if accepted after informed consent and trained/certified operator available)[a]
Face	Eyes with lens[a]
	Nasal bone[a]
	Normal profile/mandible[a]
	Intact lips[a]
Spine	Vertebrae (longitudinal and axial)[a]
	Intact overlying skin[a]
Chest	Symmetrical lung fields
	No effusions or masses
Heart	Regular cardiac activity
	Four symmetrical chambers[a]
Abdomen	Stomach present in left upper quadrant
	Bladder[a]
	Kidneys[a]
Abdominal wall	Normal cord insertion
	No umbilical defects
Extremities	Four limbs each with three segments
	Hands and feet with normal orientation[a]
Placenta	Size and texture
Cord	Three-vessel cord[a]

Reproduced with permission from Salomon et al. [11].
[a] Optional structures.

First trimester anatomy assessment

Table 2
Detection rates of fetal malformations in the first trimester.

Detection rate	Fetal malformation
100%	Acrania, anencephaly, ectopia cordis, encephalocele
50–99%	Cystic hygroma, double-outlet right ventricular flow, Fallot, gastroschisis, omphalocele, holoprosencephaly, hypoplastic left heart syndrome, limb reduction, megacystis, polydactyly, septal defects, transposition of great vessels, valvular disease
1–49%	Spina bifida, hydrocephalus, skeletal dysplasia, facial cleft, Dandy–Walker, aortic coarctation, arthrogryposis
0%	Corpus callosum agenesia, bladder exstrophy, congenital cyst adenomatoid malformation, cerebellar hypoplasia, duodenal atresia, hydronephrosis, renal agenesia, duplex kidneys, bowel obstruction, extralobar sequestration

Reproduced with permission from Rossi and Prefumo [8].

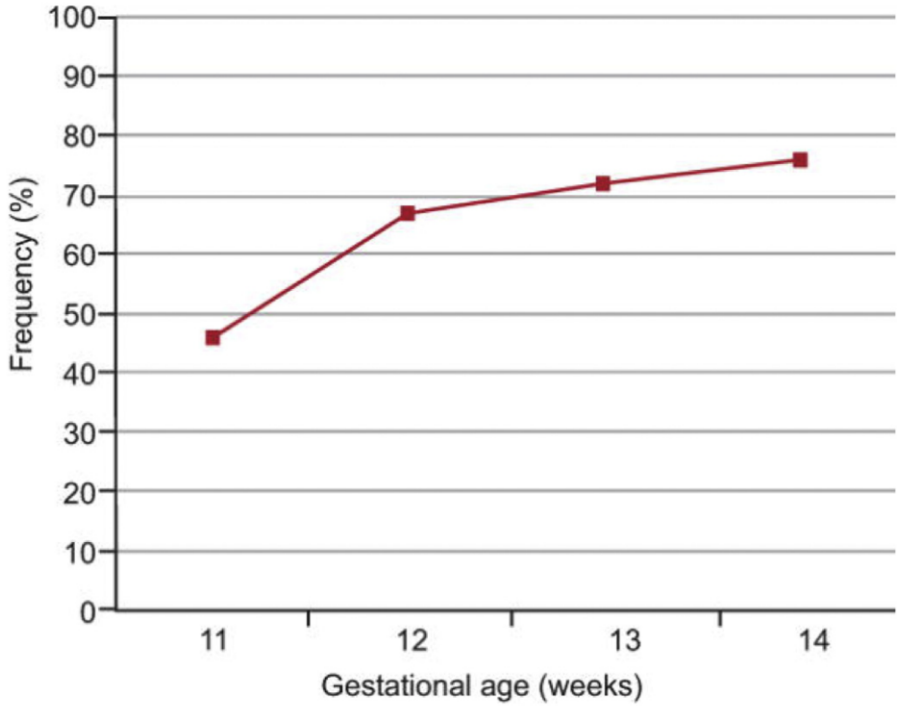
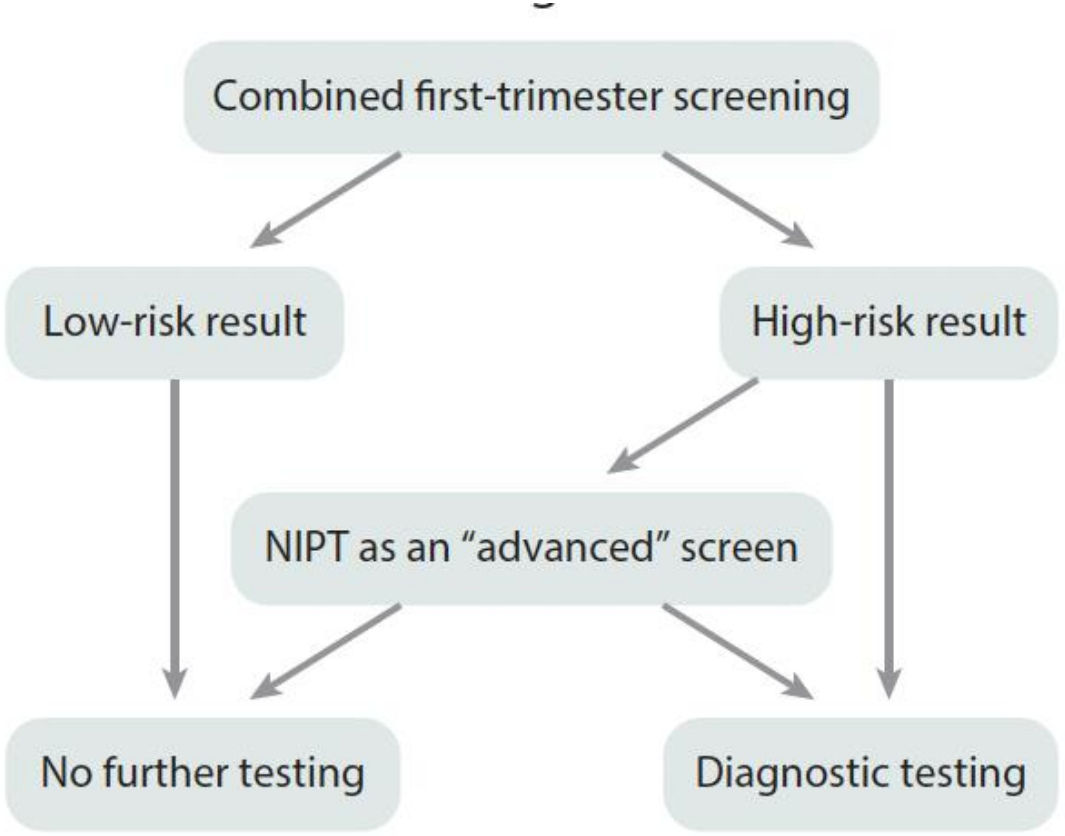


Fig. 2. Detection rates of fetal structural anomalies from 11 to 14 weeks of gestation. Reproduced with permission from Rossi and Prefumo [8].

Edwards SFNM https://doi.org/10.1016/j.siny.2017.11.005

NIPT a second-line “advanced” screen

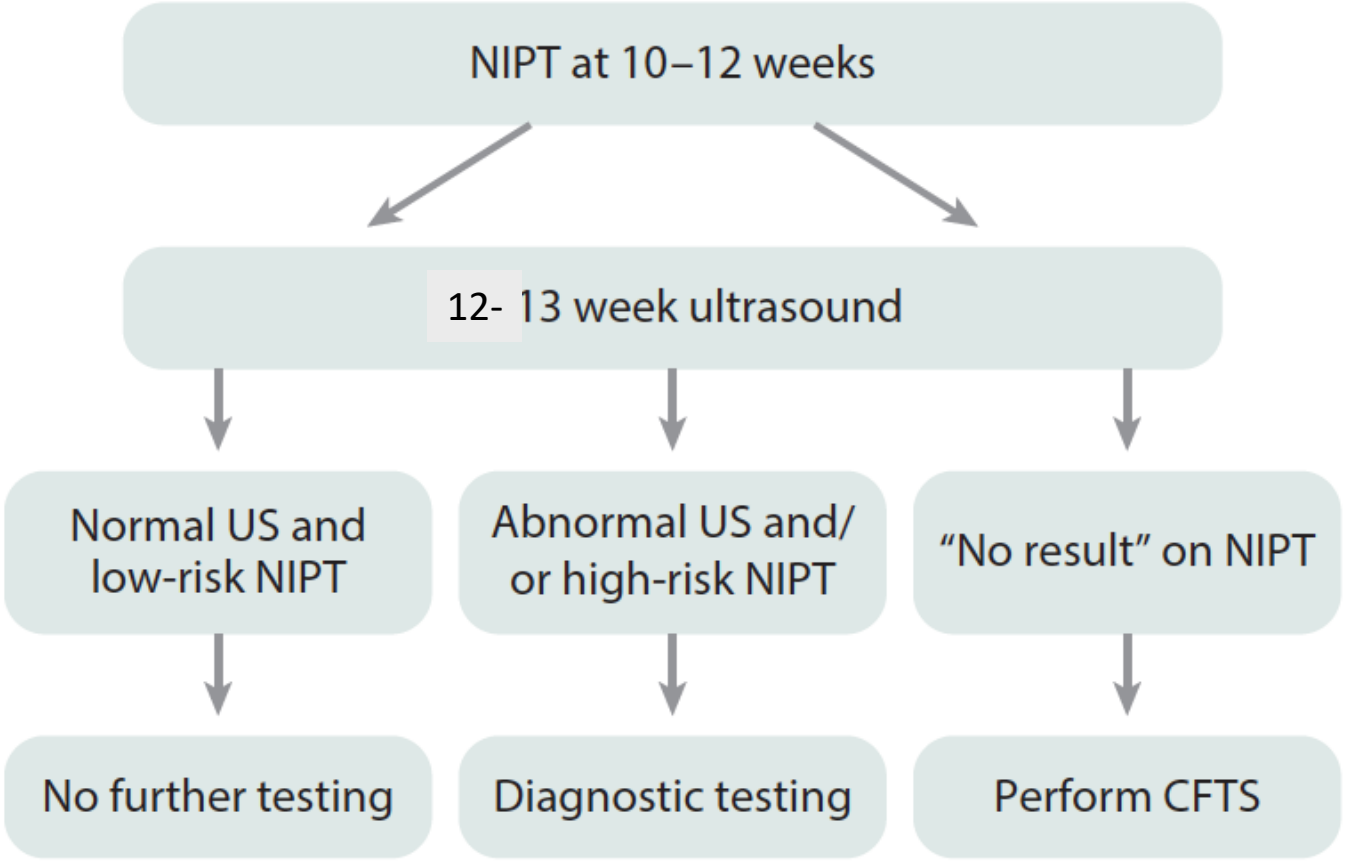
- After first trimester combined test



More affordable as fewer women require NIPT (<5%)
Still get first trimester ultrasound

Detection rate will not be improved because only those identified as increased chance by FTC will be offered NIPT

As a first-line screen with a 12-13w scan



Provide advantages of both NIPT and US

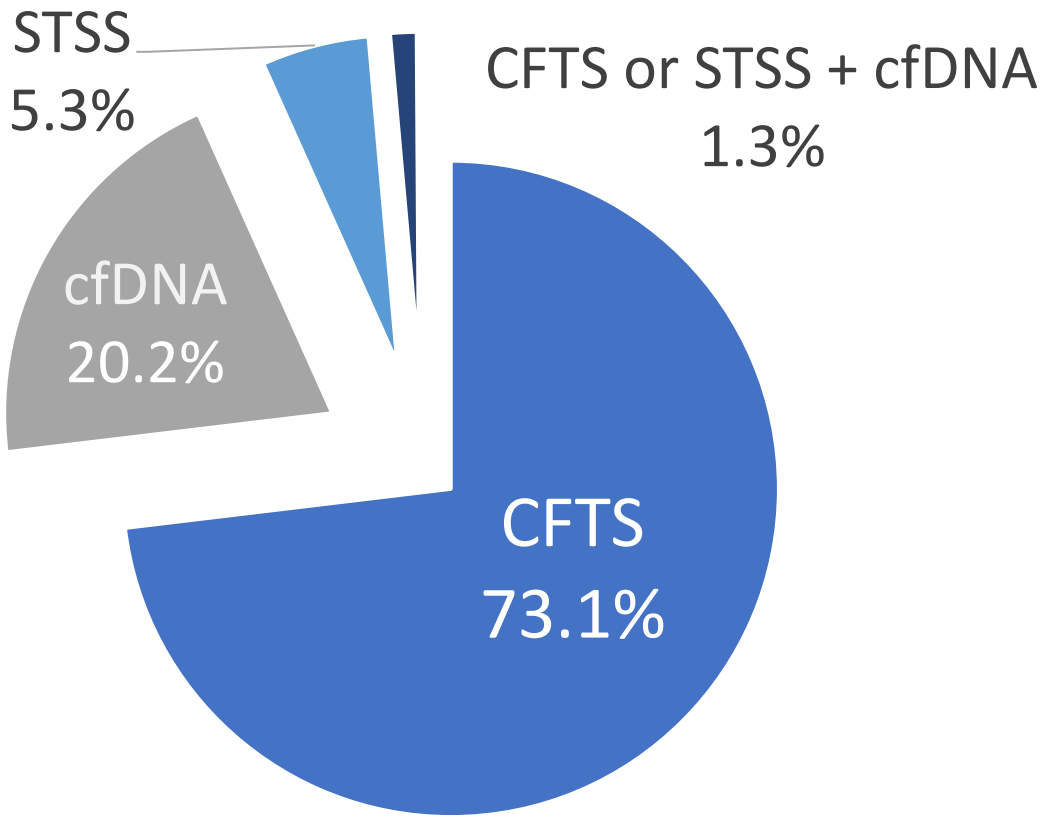
Costly!

murdoch children's research institute

State-wide utilization and performance of traditional and cell-free DNA-based prenatal testing pathways: the Victorian Perinatal Record Linkage (PeRL) study

A. LINDQUIST[1,2,3#], L. HUI[1,2,3,4#], A. POULTON[1], E. KLUCKOW[1], B. HUTCHINSON[2], M. D. PERTILE[5,6], L. BONACQUISTO[5], L. GUGASYAN[7], A. KULKARNI[7], J. HARRAWAY[8], A. HOWDEN[9], R. MCCOY[10], F. DA SILVA COSTA[11,12], M. MENEZES[6,13], R. PALMA-DIAS[3,14,15], D. NISBET[14,15,16], N. MARTIN[17], M. BETHUNE[18,19], Z. POULAKIS[6,20,21] and J. HALLIDAY[1,6]

- 79,140 births during 2015
- 83.4% (n=66,166) had screening



The Victorian Perinatal Record Linkage Study
Ultrasound Obstet Gynecol 2020; **56**: 215–224

State-wide utilization and performance of traditional and cell-free DNA-based prenatal testing pathways: the Victorian Perinatal Record Linkage (PeRL) study

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Prenatal screening	T21/13/18 Sensitivity %	Specificity for 21/13/18	Screen positive rate %
FTC N = 45,275	**89.6** (103/115)	97.25	**2.94**
NIPT N = 12,486	**100** (73/73)	99.93*	1.21* **2.42#**

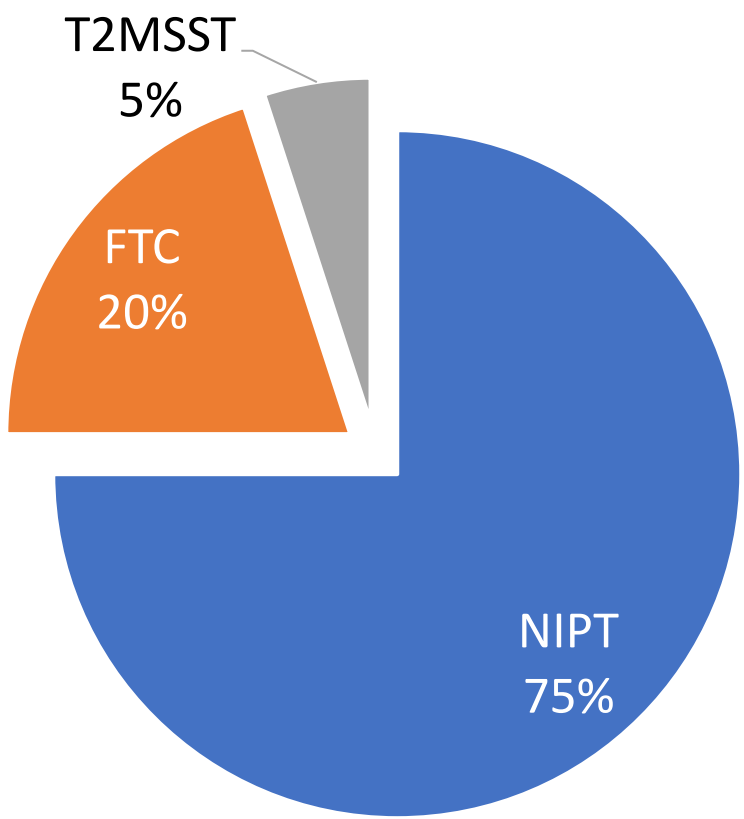
*Only high-risk results for T21/13/18 included

includes high risk results for all reported chromosomes and “no call” results

Ultrasound Obstet Gynecol 2020; **56**: 215–224

2021-2022 screening in Melbourne

- Prenatal screening data collected from 11 public hospital maternity databases (BOS)
- Data available on = 29,495 births ≥ 20w
  - 83% had screening
  - 15% declined
  - 2% not offered
- NIPT associated with:
  - Younger
  - Nulliparous
  - higher SES postcode
  - born in Australia, North East Asia, Europe



Healthcare professional survey

Australian Government
Australian Research Council

- Online survey of Australian maternity clinicians Sept-Oct 2022
- RANZCOG, HGSA, Rural Doctors Association mailing lists, social media,
- N = 540 respondents
  - Obstetricians 49%
  - GPs 35%
  - Genetics professional 9%
  - Midwives 7%
- Type of practice: public only 30%, private only 35%, both 35%
- All states /territories – QLD n = 60 (16% total)
- >90% involved in pretest counselling, consent, and returning results

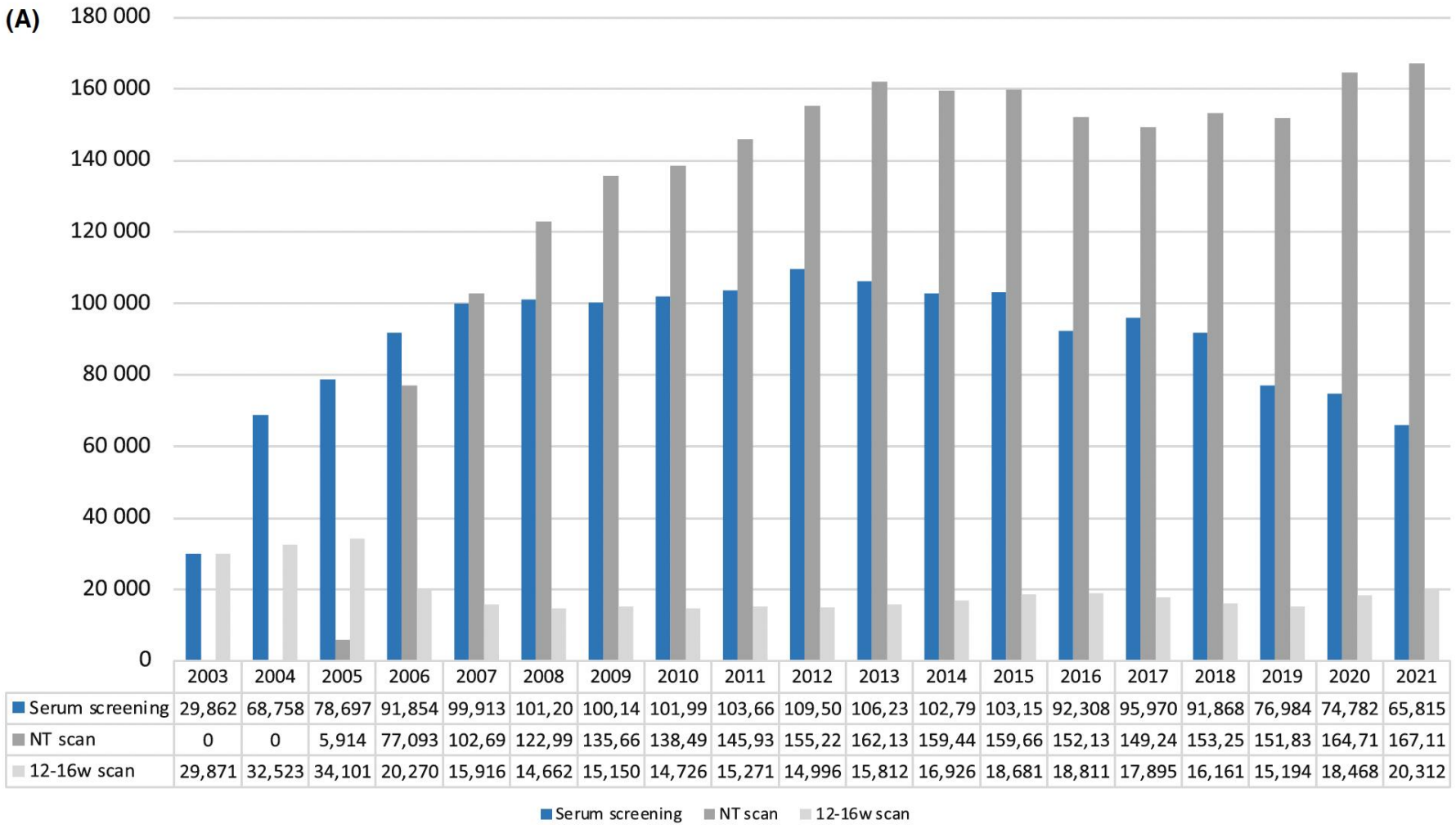
How do you offer NIPT?

	N (%)
Offer a choice between NIPT and combined first trimester screening (CFTS) for patients in the first trimester	279 (60.3)
First-line screening test for all patients	88 (19.0)
First-line screening test only for patients of advanced age or with other risk factors for aneuploidy	12 (2.6)
Second-tier screening test after combined first-trimester screening (CFTS)	49 (10.6)
Other	35 (7.6)

What first trimester ultrasounds do you offer to patients having NIPT?

	N (%)
6-8 weeks (dating)	257 (54.1)
10 weeks (pre-NIPT)	137 (28.8)
11-13 weeks (early fetal structural survey or concurrently with NIPT)	409 (86.1)
Other	24 (5.1)

National serum screening and NT scans (MBS claims)



	2003	2004	2005	2006	2007	2008	2009	2010	2011	2012	2013	2014	2015	2016	2017	2018	2019	2020	2021
Serum screening	29,862	68,758	78,697	91,854	99,913	101,20	100,14	101,99	103,66	109,50	106,23	102,79	103,15	92,308	95,970	91,868	76,984	74,782	65,815
NT scan	0	0	5,914	77,093	102,69	122,99	135,66	138,49	145,93	155,22	162,13	159,44	159,66	152,13	149,24	153,25	151,83	164,71	167,11
12-16w scan	29,871	32,523	34,101	20,270	15,916	14,662	15,150	14,726	15,271	14,996	15,812	16,926	18,681	18,811	17,895	16,161	15,194	18,468	20,312

Stable no. 11-13w scans

40% reduction in serum screening

Do you offer a choice of conditions?

Choice of conditions to be screened with NIPT offered to patients	n (%)
Yes	245 (53.0)
No	94 (20.3)
Sometimes	79 (17.1)
N/A to my role	44 (9.5)
Other screening options discussed besides T21/13/18 (N=324)	
Sex chromosome aneuploidies	278 (85.8)
Genome-wide NIPT	93 (28.7)
Microdeletions	102 (31.5)
Single gene disorders	51 (15.7)
Other	18 (5.6)

How well do you know the market?

Massively parallel sequencing (MPS)

Chromosome-targeted sequencing

SNP-targeted sequencing

nest

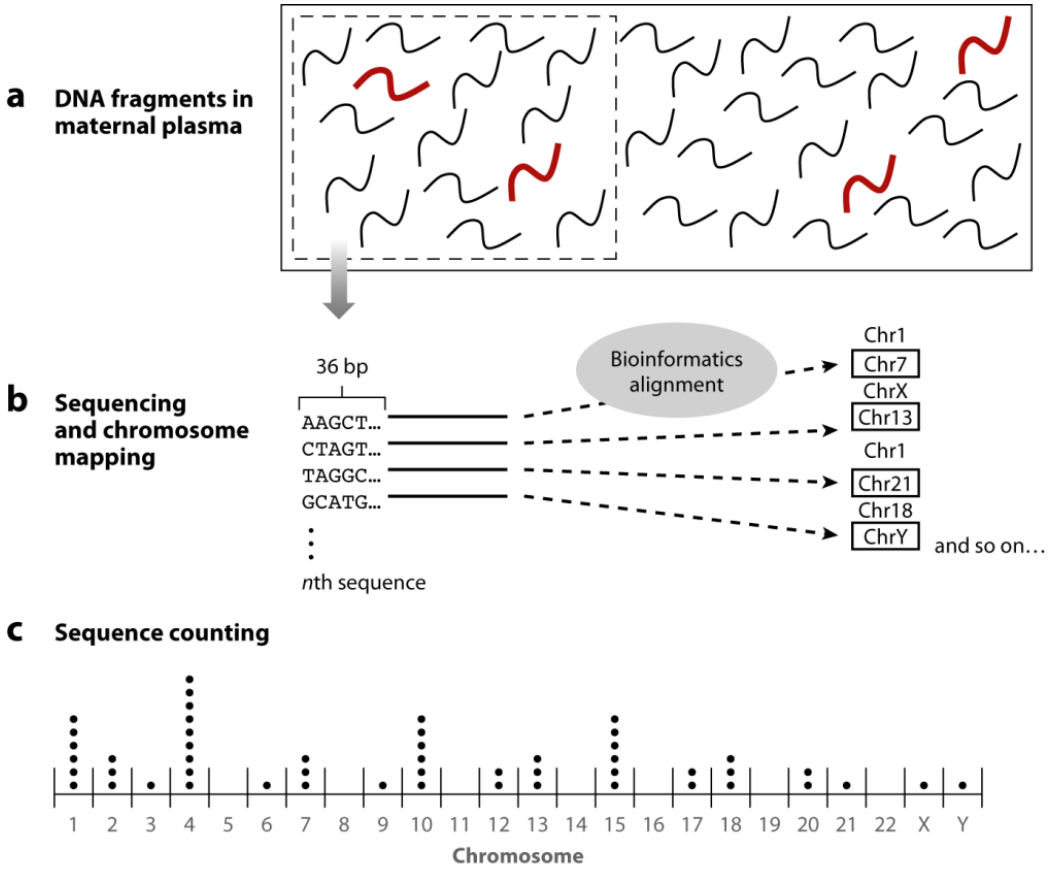
Harmony PRENATAL TEST

panorama®
natera prenatal screen

kickstart for life

1. Whole genome (random) sequencing

- Sequences all fragments of plasma DNA nonselectively (random massively parallel sequencing)
- No distinction between maternal and fetal DNA
- Able to perform genome-wide assessment (gwNIPT)
- Resolution/ analysis can be adjusted at bioinformatics stage
- Products
  - NEST – 21/13/18,X,Y
  - Percept – genome wide
  - MateriT21 Plus (Sequenom)
  - verifi (Illumina)
  - NIFTY (BGI)
  - Generation



mercy perinatal
kickstart for life

2. Chromosome-targeted NIPT

- Harmony (Roche)
- 384 polymorphic loci unique to the target chromosomes (21,13,18,X,Y) are amplified in a PCR reaction

→template enriched for selected chromosomes

→ microarray based quantification of cfDNA (formerly NGS)

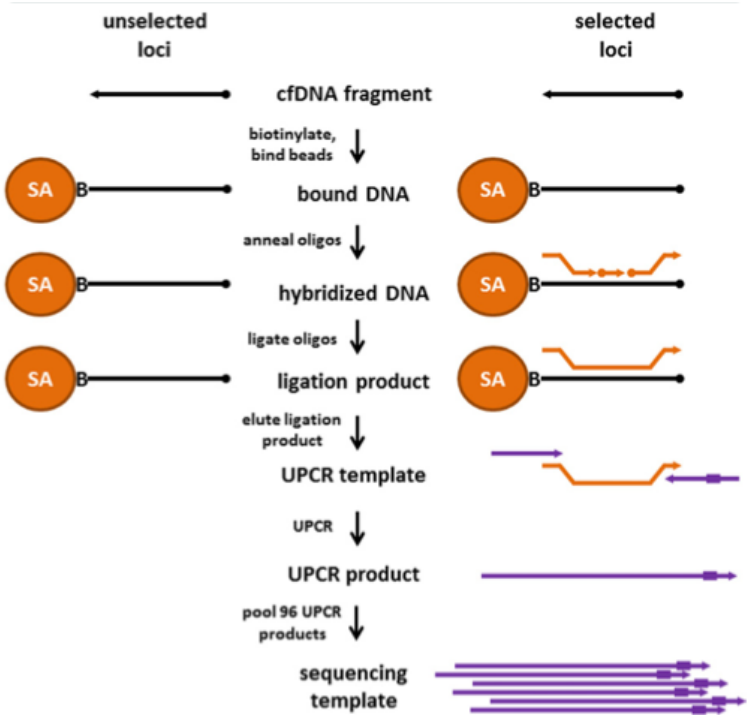
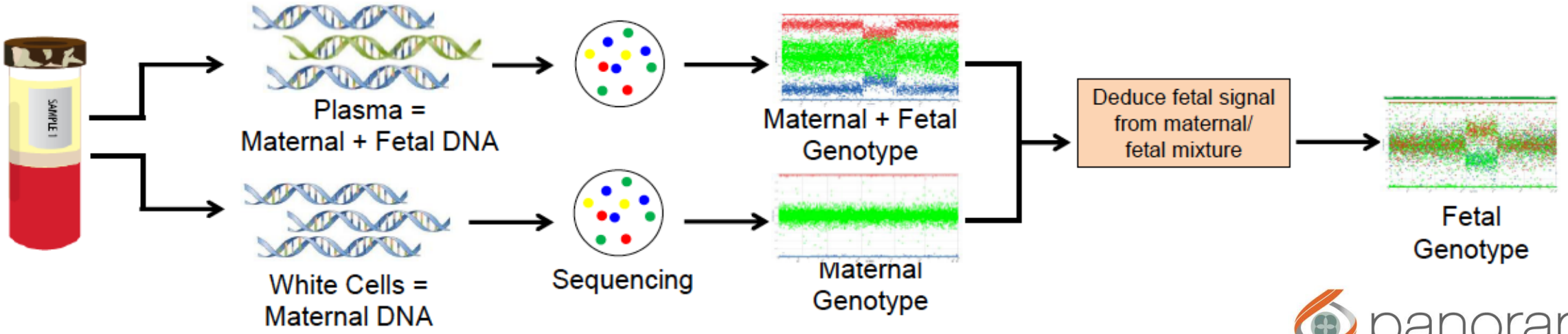


Fig 1. DANSR assay. Sparks et al AJOG 2012

Harmony™ PRENATAL TEST

3. Single nucleotide polymorphism-based

- DNA extracted from plasma and buffy coat
- Targeted amplification and analysis of ~20,000 SNPs on chr 21, 18, 13, X and Y
- Analyses allele distributions at each SNP locus
- Deduces fetal genotype by subtracting maternal signal



panorama®
natera prenatal screen

Figure from Levy et al 2013 Presented at SMFM 2013

Match the sequencing methods with test features

1. Massively parallel sequencing (MPS)
2. Chromosome-targeted sequencing
3. SNP-targeted sequencing

A. Can distinguish cell-free maternal DNA from fetal DNA 3
B. Can detect triploidy 3
C. Can detect subchromosomal imbalances 1
D. Can be used for twins 1,2,3
E. Can be used for triplets ?
F. Can detect maternal cancer 1
G. Can distinguish MZ from DZ twins 3

The Royal Australian and New Zealand College of Obstetricians and Gynaecologists

Excellence in Women's Health

Prenatal screening and diagnostic testing for fetal chromosomal and genetic conditions

Recommendation 5	Grade and supporting references
Pre-test counselling for cfDNA-based screening should include informed decision making regarding testing for fetal sex and sex chromosome aneuploidy. The potential for other unanticipated findings of relevance to maternal health (including maternal genomic imbalances), should be included in pre-test counselling.	Consensus-based recommendation

The Royal Australian and New Zealand College of Obstetricians and Gynaecologists

Excellence in Women's Health

Prenatal screening and diagnostic testing for fetal chromosomal and genetic conditions

Recommendation 9	Grade and supporting references
Routine population-based screening for genome-wide chromosome abnormalities and microdeletion syndromes are not recommended due to the absence of well-performed clinical validation studies.	Consensus-based recommendation

DOI: 10.1002/pd.6357

COMMENTARY

PRENATAL DIAGNOSIS WILEY

Position statement from the International Society for Prenatal Diagnosis on the use of non-invasive prenatal testing for the detection of fetal chromosomal conditions in singleton pregnancies

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- ISPD Board consensus opinion based on current knowledge and clinical practice
- Statement relevant to high income settings where prenatal screening for aneuploidy is considered an established part of antenatal care.

Outline of the 2023 ISPD statement

- Introduction, scope and purpose, biology and technological background
- Performance characteristics: T21, T18, T13, PPV
- Fetal fraction, test failures, maternal neoplasia
- Implementation models of NIPT
- Expanded NIPT
- Sex chromosome aneuploidy and fetal sex determination
- Rare autosomal trisomies
- Subchromosomal imbalances
- Microdeletions and microduplications
- Role of ultrasound in complementing NIPT
- Pre and post test counselling challenges
- Ethical issues
- Future of cfDNA

Implementation of NIPT for T21, T18, T13

- NIPT is the most accurate screening test for the common autosomal aneuploidies in unselected singleton populations, and those at known increased probability

TABLE 2 The performance of non-invasive prenatal testing (NIPT) in a general unselected population for trisomy 21, 18 and 13 (adapted from Rose et al[6]).

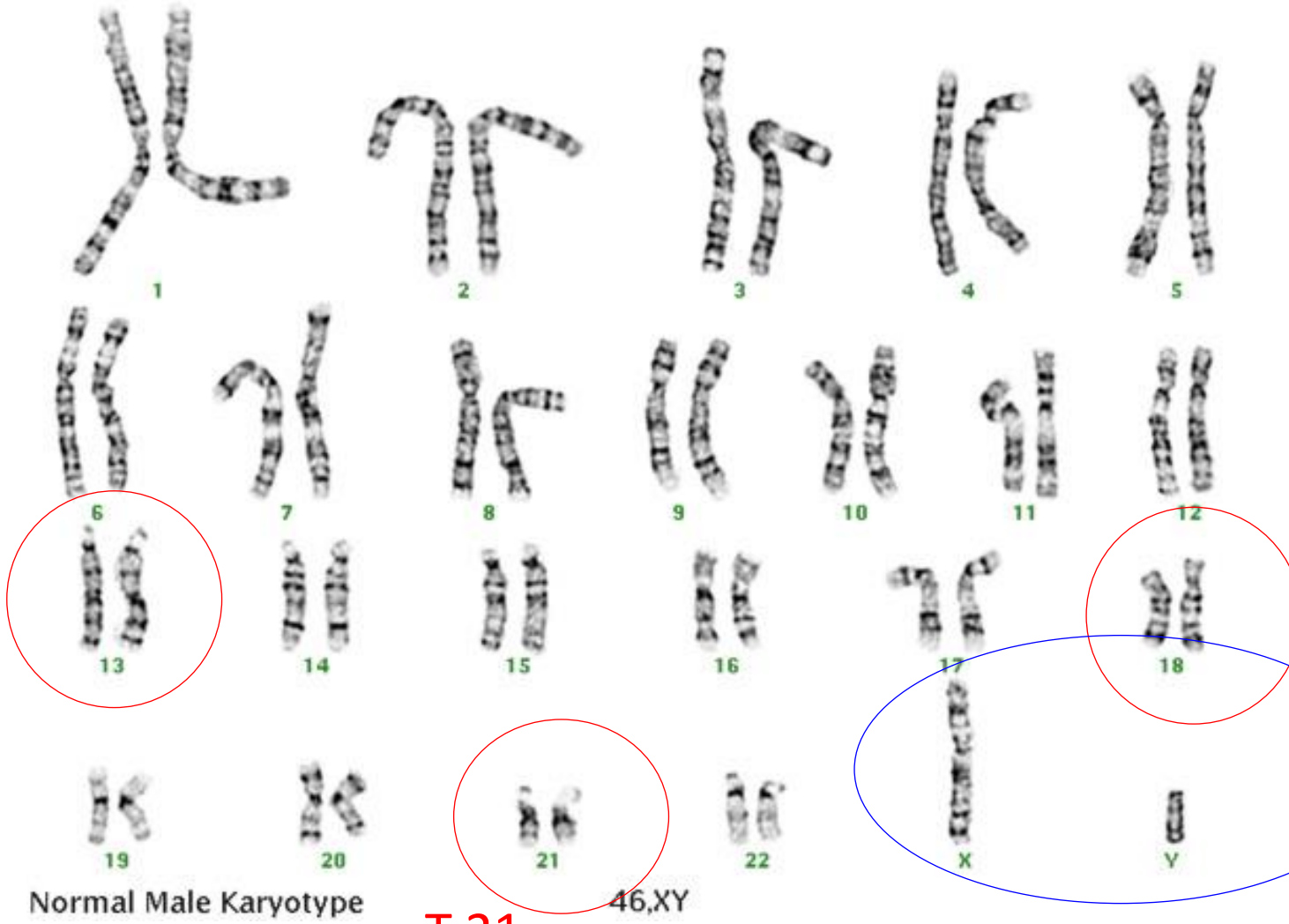
Condition	Sensitivity (%)	95% CI	Specificity (%)	95% CI	PPV (%)	95% CI
Trisomy 21	98.80	97.81–99.34	99.96	99.92–99.98	91.78	88.43–94.23
Trisomy 18	98.83	95.45–99.71	99.93	99.83–99.97	65.77	45.29–81.68
Trisomy 13	100	0–100	99.96	99.92–99.98	37.23	26.08–49.93

Rose et al. Systematic evidence-based review: the application of noninvasive prenatal screening using cell-freeDNA in general-risk pregnancies. Genet Med.2022;24(9):1992

NIPT for common autosomal trisomies T21, T18, T13

- NIPT for the common autosomal aneuploidies performs sufficiently well to be offered in primary or contingent screening models
- The ISPD board acknowledges that context-specific considerations in health policy influence decisions and implementation models

Expanded NIPT



T13
5%

T 21
53%

T18
13%

SCA
12%

Other abnormalities
17%

EUROCAT Wellesley 2012

NIPT for sex chromosome aneuploidies

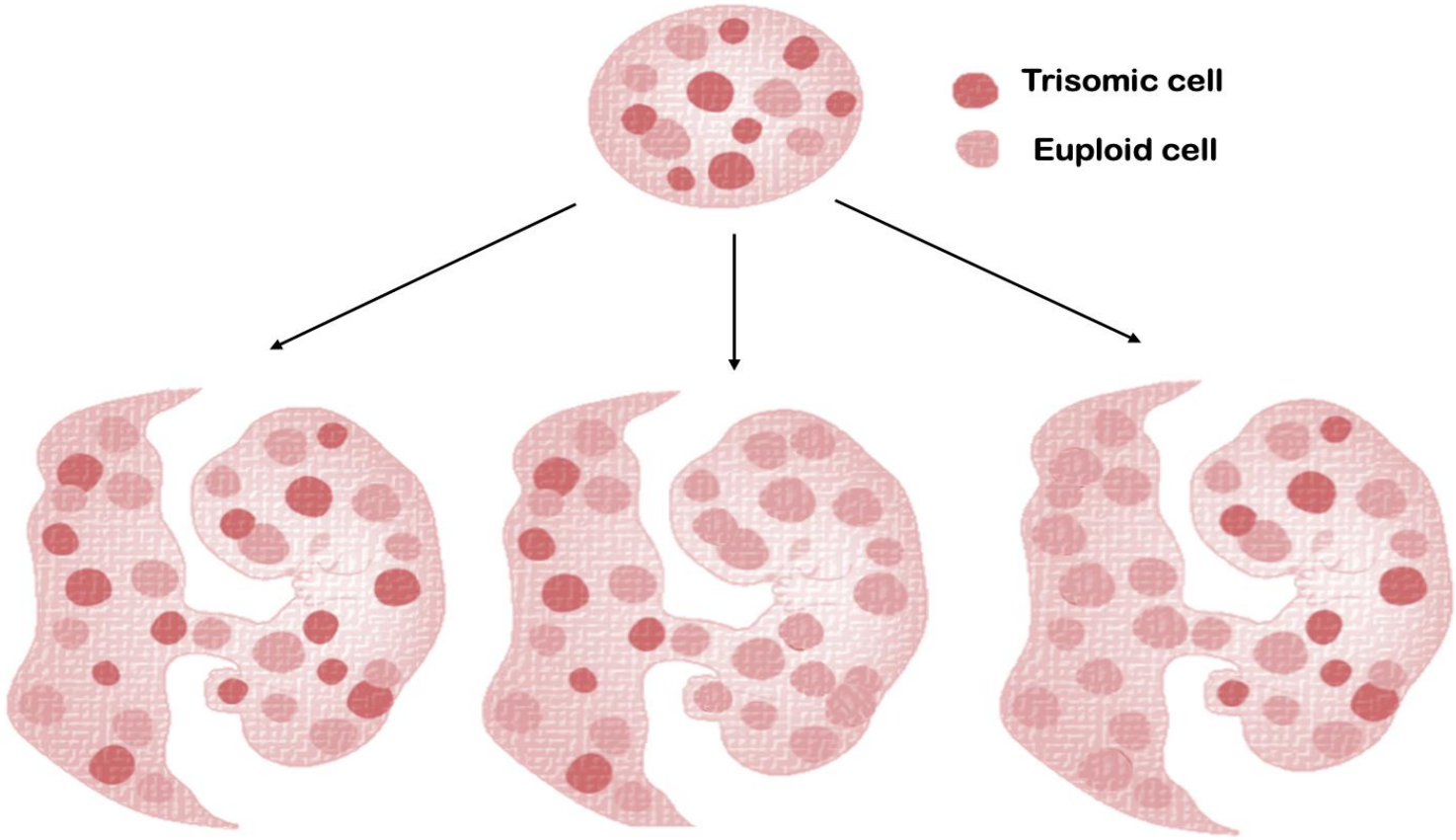
- American College of Medical Genetics and Genomics 2023 guideline: recommended SCA screening to be offered to all women
- NIPT for SCA is sufficiently accurate to be offered alongside autosomal aneuploidy screening with specific pretest counseling and consent.
- Other societal, economic, cultural and ethical factors may need to be considered in health policy decisions regarding population-based screening for SCAs
- Further studies to evaluate the downstream impacts of offering NIPT for sex chromosome conditions should be considered where such screening is offered.

NIPT for rare autosomal trisomies (RATs)

- Full trisomies of autosomes other than 21, 13, 18 are rare in live fetuses
- 97% of all RATs detected on CVS are confined to the placenta
- NIPT has a lower PPV for RATs than other trisomies ~ 11%
- TRIDENT study reported results of genome-wide NIPT in Netherlands → Dutch Health council advised RATS no longer routinely reported
- Risk of UPD if false positive NIPT result is for an imprinted chromosome

Dutch Health Council advice to the Minister of Health, Welfare and Sport. The Non-invasive Prenatal Test (NIPT) as Population Screening. No. 2023/03, the Hague, 2023. Accessed on 3 March 2023 at.https://www.gezondheidsraad.nl/?utm_source=gr.nl&utmmedium=redirect&utm_campaign=redirect

Fetoplacental mosaicism and NIPT



Fetoplacental mosaicism, Potentially detectable by NIPT

Confined placental mosaicism, False pos NIPT

Isolated fetal mosaicism, Not detectable by NIPT

Adverse obstetric outcomes after positive NIPT for RAT

- CPM also confers increased risk of adverse outcome but cost effectiveness of gw-NIPT for obstetric indications is not established

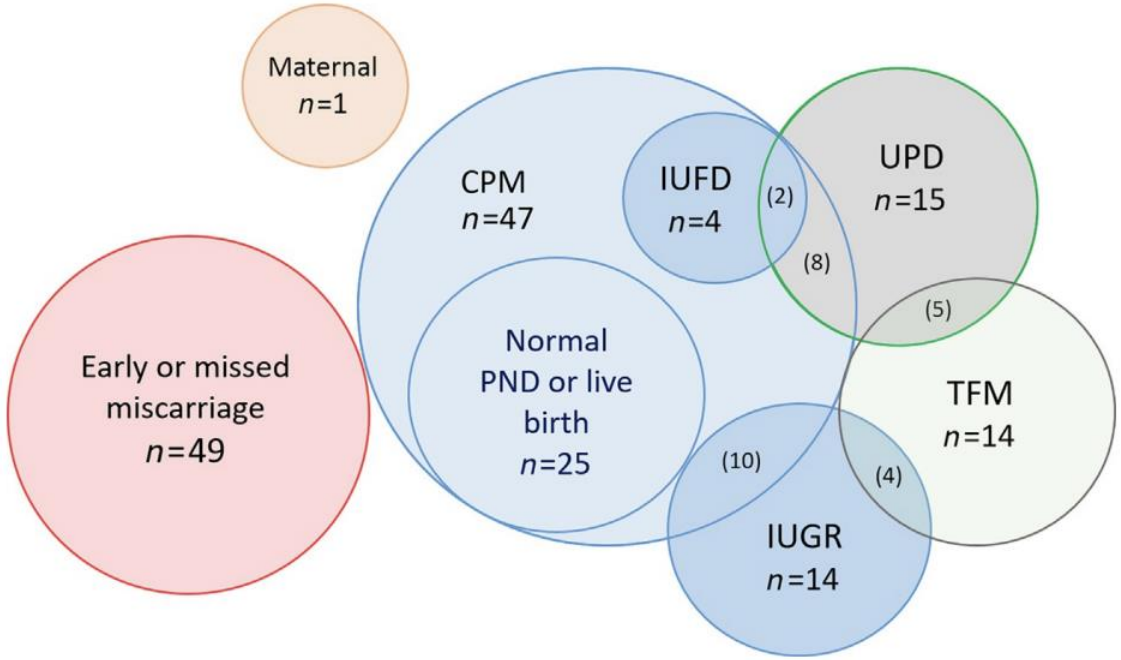


FIG. 1

Outcomes of 120 rare autosomal trisomies identified using cfDNA-based screening. Genome-wide screening was undertaken at the Victorian Clinical Genetics Services, Melbourne, Australia. A subset of the cohort has been reported previously [26].

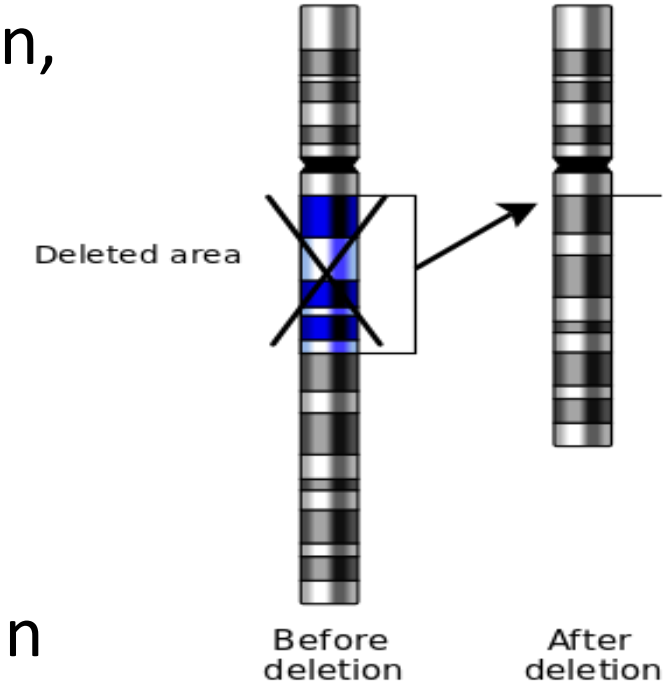
Noninvasive Prenatal Testing (NIPT). https://doi.org/10.1016/B978-0-12-814189-2.00007-4

NIPT for rare autosomal trisomies

- There is insufficient data to assess the performance and clinical utility of routine NIPT for RATs. NIPT for RATs is therefore not recommended for the routine care of unselected populations.
- Where screening for RATs is performed, management after a high chance result requires expert post-test counselling and specialist management.

Expanded NIPT for subchromosomal imbalances >7Mb

- Genome-wide NIPT can also detect subchromosomal imbalances (segmental imbalances, CNVs)
- Detection depends on size of imbalance, fetal fraction, sequencing depth, and for SNP-based approaches, included targets regions
- Belgium and the Netherlands experience
  - PPVs of 47% and 32%
  - Similar to the PPVs of NIPT for trisomy 13
- No sensitivity data because of incomplete follow up in screen-negative cases

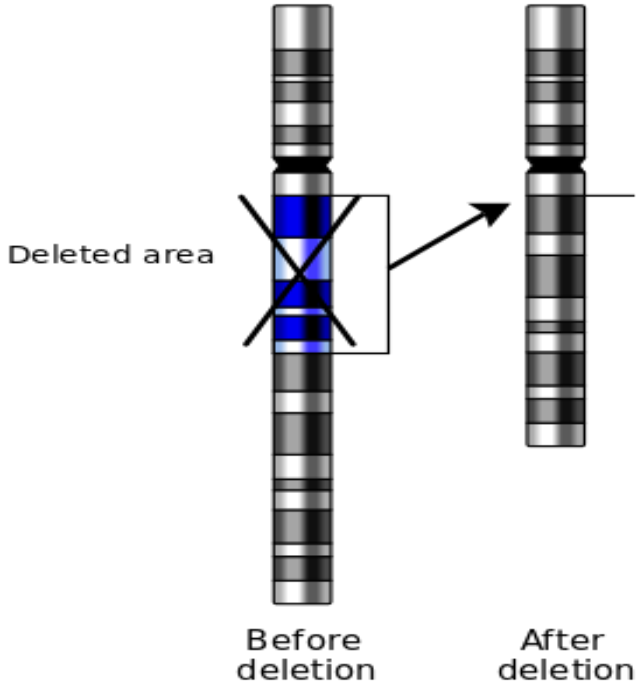


Expanded NIPT for subchromosomal imbalances >7Mb

- There is insufficient data to assess the performance and clinical utility of routine NIPT for subchromosomal imbalances.
- Large scale population-based evaluations of routine screening for subchromosomal imbalances are being undertaken in several countries and data continue to emerge.
- Until such time as the outcome data are clear and shown to be reproducible in other settings, NIPT for subchromosomal imbalances is not recommended for the routine care of unselected populations.
- Where screening for subchromosomal imbalances is performed, management after a high chance result requires expert post-test counselling and specialist management.

Expanded NIPT for microduplication/microdeletion syndromes

- Microdeletions/duplications:
  - < 5 Mb size
  - Different technological approach to gw-NIPT for larger imbalances
- Most common is 22q11.2 deletion syndrome (DiGeorge syndrome)
- Wolf-Hirschhorn syndrome (4p16.3 DS), Cri-du-chat syndrome (5p15.33 DS), Prader-Willi/Angelman (15qdel), 1p36 deletion



Wapner NEJM 2012

Expanded NIPT for microduplication/microdeletion syndromes

- Familiari et al : systematic review of NIPT for MMS included 7 studies
  - None performed genetic confirmation in cases that did not undergo prenatal diagnostic testing or those with a negative screen result – therefore specificity and sensitivity undetermined
- 1 prospective study of 22q11 DS (Dar et al) performed genetic testing in all analysed pregnancies
  - Sensitivity 75%, screen positive rate 0.2%, PPV 24%
  - 12 affected cases; high frequency of 22q in this cohort (1 in 1524)
  - Updated algorithm improved the Sn to 83% (10/12), PPV to 53%

Dar P, et al. Cell-free DNA screening for prenatal detection of 22q11.2 deletion syndrome. Am J Obstet Gynecol 2022

Expanded NIPT for 22q11 deletion syndromes

- 22q11.2 deletion syndrome is the most common microdeletion syndrome. Only one study has evaluated cfDNA-based screening for 22q11.2 deletion syndrome in a clinical cohort with genetic confirmation of all participants.
- There is insufficient data to assess the performance and clinical utility of routine NIPT for MMS. NIPT for MMS is therefore not recommended for the routine care of unselected populations.

Challenges with synthesizing literature on expanded NIPT

- Difference in technological platforms
- Published studies have variable population characteristics that influence the performance of NIPT, such as gestational age at testing, referral indications, frequency of fetal structural abnormalities, other risk factors
- Rarity of some conditions impeded clinical validation studies
- Variable clinical phenotype makes ascertainment of false negative NIPT results challenging

The Royal Australian and New Zealand College of Obstetricians and Gynaecologists

Excellence in Women's Health

Prenatal screening and diagnostic testing for fetal chromosomal and genetic conditions

Recommendation 9	Grade and supporting references
Routine population-based screening for genome-wide chromosome abnormalities and microdeletion syndromes are not recommended due to the absence of well-performed clinical validation studies.	Consensus-based recommendation

Ethical issues

- The ethical implementation of NIPT requires attention to provision of quality pre and post-test counseling, equity of NIPT access, and access to appropriate downstream clinical services
- All stakeholders including health care consumers should be involved in determining local implementation models and future directions for NIPT

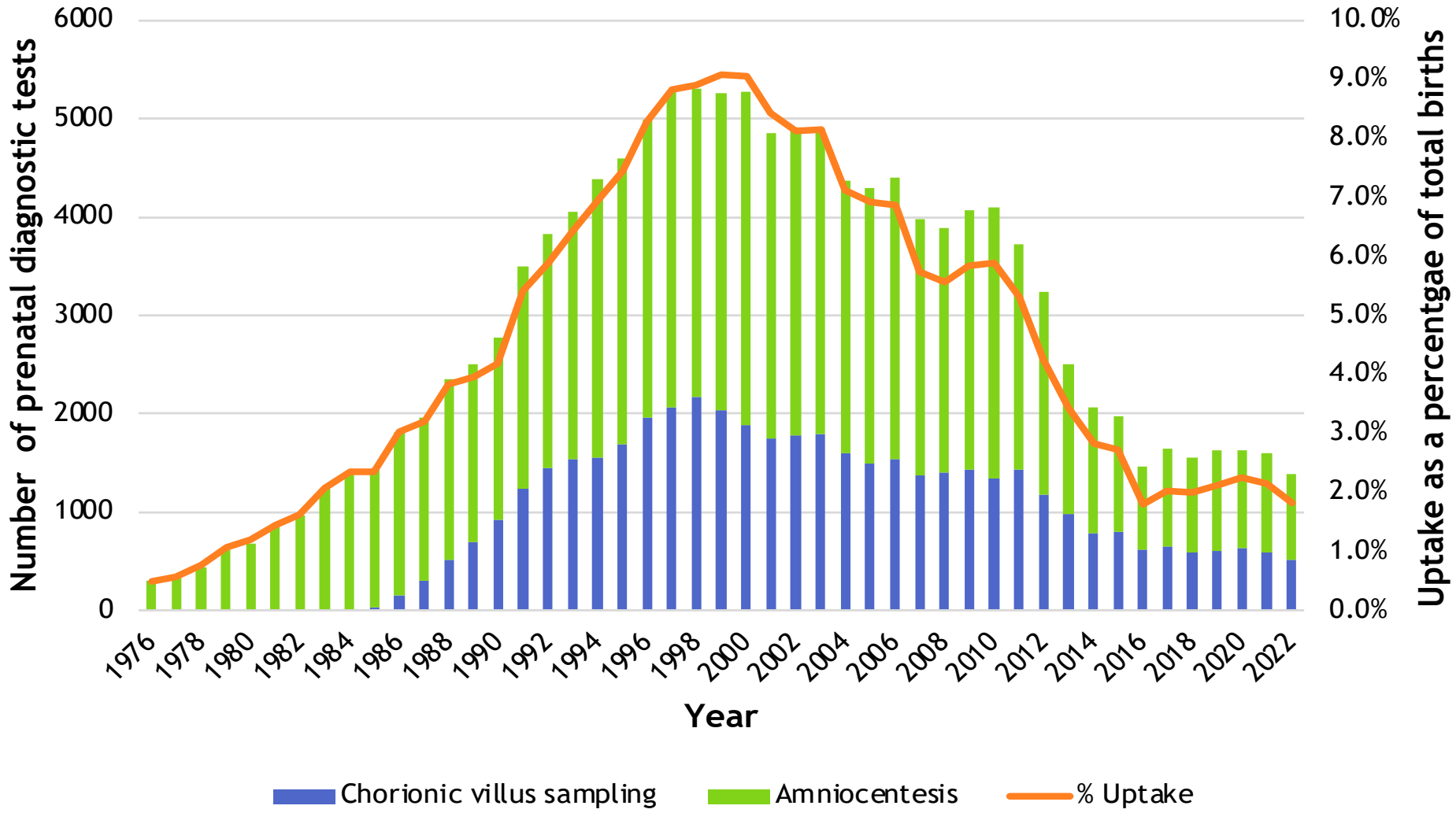
Summary

- NIPT is now the most commonly used first line screening test
- First trimester ultrasound has additional value for early detection of anomalies
- Increasing complexity with genome-wide NIPT and microdeletion screening
- Screening for T21/13/18 should be offered
  - Sex chromosome screening optional with specific consent
  - Expanded NIPT – only if sufficiently informed and resources to manage consequences

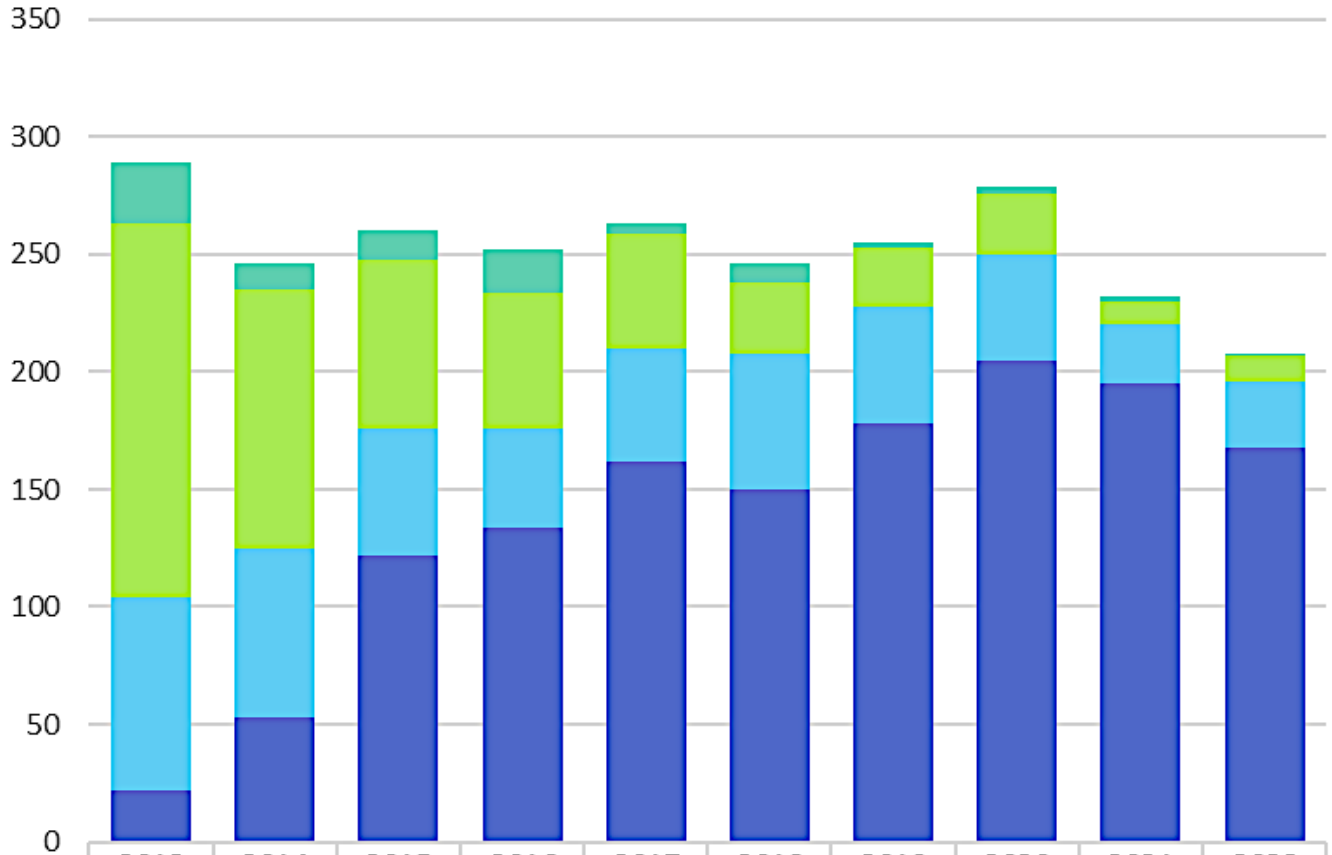
"He was doing well at first, but then he started drifting in and out of health coverage."

Where are we now with prenatal diagnosis?

VIC prenatal diagnosis 1976-2022



Indications for testing in confirmed cases of trisomy 21/18/13 2013-2022



	2013	2014	2015	2016	2017	2018	2019	2020	2021	2022
All other indications	26	11	12	18	4	8	2	3	2	1
Positive first trimester combined screening	159	110	72	58	49	30	25	26	10	11
Ultrasound abnormality	82	72	54	42	48	58	50	45	25	28
Positive NIPT	22	53	122	134	162	150	178	205	195	168

80% of common trisomies (90% of T21) ascertained via NIPT (VIC)

Prenatal screening for chromosome conditions – it's YourChoice

Screening tests provide more information about the health of your baby.

Learn about testing for chromosome conditions in pregnancy and what choices you may wish to discuss with your maternity care provider.

Start YourChoice

Developed by murdoch children's research institute, JAMES COOK UNIVERSITY AUSTRALIA

Why use this decision aid?

https://yourchoice.mcri.edu.au/dashboard

Screening tests give a chance result: understanding chance

A prenatal screening test gives an estimate of the chance that your pregnancy has a condition. It does not give a definite (yes or no) answer. If you receive a high chance result, follow up counselling and a diagnostic test will be offered. A diagnostic test (CVS or amniocentesis) will give a definite (yes or no) answer as to whether your baby has a condition or not.

- Why aren't screening results perfect? +
- What will a screening test tell me? +
- What does low chance mean? +
- What does high chance mean? +
- What does 1 in 100 mean? +
- When is the test result called 'high chance'? +

Making decisions with your screening results

Screening test results that show an increased chance of a chromosome condition can only be confirmed with diagnostic testing. Diagnostic testing is a procedure performed by a specialist doctor.

- What happens if I receive a high chance screening result? +
- What does diagnostic testing involve? +
- What happens if a condition is confirmed in my unborn baby? +
- What does a termination of pregnancy involve? +

Have you thought about what you might do with a high chance screening result?

If I receive a high chance screening result, I would have a CVS or amniocentesis to confirm the result

- Strongly Agree
- Agree

How does YourChoice work?

1. Learn and answer questions
2. Find the right test for you
3. Save the result and discuss with your doctor or midwife

Recommended time
15 - 20 mins

Steps 6 of 7

So far you are

Leaning away from screening | Unsure | Leaning towards screening

Unsure about prenatal screening

How much information do you want?

Before having a screening test, it's worth thinking about the type and amount of information they can provide. Some chromosome conditions, such as Down syndrome, are well understood, with good information on the expected health outcomes of affected children. Some conditions that can be detected by screening are rare and may not be well understood. Different tests give different information.

What information can I get from combined first trimester screening (CFTS)? +

What information can I get from non-invasive prenatal testing (NIPT 'core')? +

Can I find out if I am having a girl or boy from NIPT? +

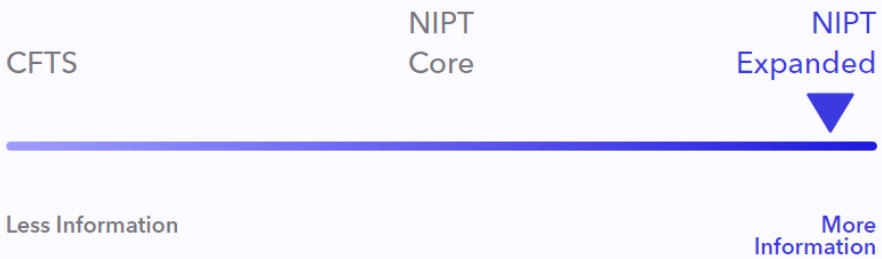
I would like to be told in pregnancy if my baby might have a rare condition, even if it has an uncertain health outcome.

- Strongly Agree (●)
- Agree ( )
- Neither Agree Nor Disagree ( )
- Disagree ( )
- Strongly Disagree ( )

Well done for completing YourChoice!
The following results are based on how you answered each step.

Which test are you leaning towards:

Scale for prenatal screening tests



What does this mean?

From your answers, it looks like you would like as much information as possible about the health of your baby, even if the results may be uncertain and the costs might be higher. You may wish to discuss NIPT options with your maternity care provider.

Remember that this is not medical advice and should always be discussed with your doctor or midwife. Make sure you let your doctor or midwife know about the type of information you want when you are asking about which screening test is available to you.

Overview

A genetics course designed specifically for maternity health professionals by clinicians with expertise in prenatal screening and diagnosis.

mercy perinatal kickstart for life
murdoch children's research institute
Monash IVF Group

Developed as a joint initiative between the Mobile Learning Unit, Melbourne Medical School, Mercy Perinatal, Mercy Hospital for Women, Monash IVF Group and the Murdoch Children's Research Institute Reproductive Epidemiology Group.

Genetic technology advances rapidly, and it is challenging for

Course Information

Fees

For Doctors (specialists/GPs): AUD $385 (incl. GST)

For Midwives/ Sonographers/ Trainees/ Students: AUD $250 (incl. GST)

Location

Online

Entry requirements

Participants of this course should be either enrolled in, or have graduated from a midwifery, nursing, genetic counselling, genetics, or medical degree.

https://study.unimelb.edu.au/find/short-courses/genetics-in-pregnancy/

RACGP CPD Approved Activity — Educational Activities 12 hours

RANZCOG CPD APPROVED ACTIVITY 2023

Knowledge and Skills

1. Understand the basis of genetic inheritance, mechanisms of genetic variation, and the difference between “genetics” and “genomics”
2. Apply the clinical and ethical principles underpinning prenatal screening to clinical practice
3. Identify couples that require a genetic counselling referral
4. Concisely articulate options for prenatal screening
5. Provide general information to patients about prenatal diagnostic procedures
6. Understand the indications and methods used for pre-implantation genetic testing
7. Describe the current options for genetic carrier screening
8. Recognise and reflect on the ethical considerations in prenatal genomic testing
9. Use disability-positive language when discussing genetic variation and congenital health conditions

Assessment

A Certificate is issued upon satisfactory completion of the course assessment requirements: 6 tutorial unit self-assessments and 10 case studies

Delivery Mode

Course completion requires approximately 12 hours of eLearning. This course is delivered online and students can study in their own time and location. Course materials can be accessed using a web browser

Thank you

Overview

A genetics course designed specifically for maternity health professionals by clinicians with expertise in prenatal screening and diagnosis.

Developed as a joint initiative between the Mobile Learning Unit, Melbourne Medical School, Mercy Perinatal, Mercy Hospital for Women, Monash IVF Group and the Murdoch Children's Research Institute Reproductive Epidemiology Group.

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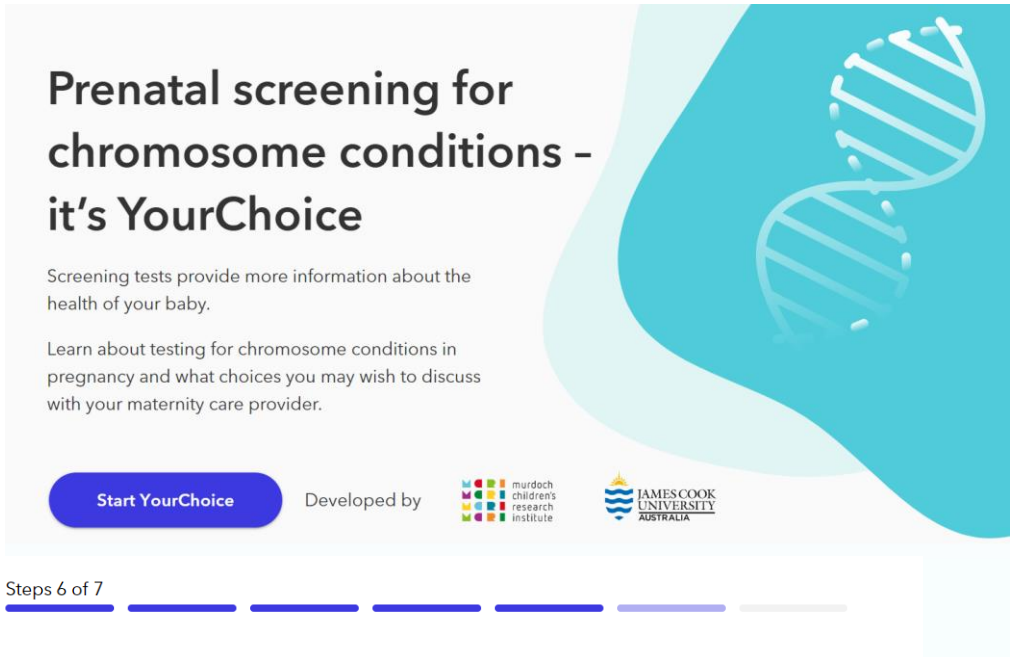
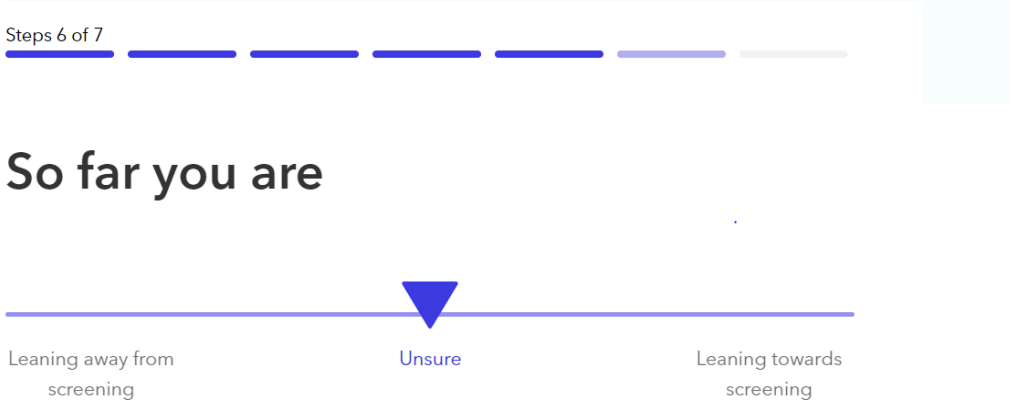
RACGP CPD Approved Activity — Educational Activities 12 hours

RANZCOG CPD APPROVED ACTIVITY 2023





3

Modes of Delivery
Is there such a thing as a normal birth?

Dr Vicki Carson
The Royal Women's Hospital
Frances Perry House

DR VICKI CARSON

- Trained RWH, Mercy, Warrnambool
- 3 years as a consultant in Warrnambool
- Group private practice in East Melbourne
- Now private via FPH
- RWH consultant since return from Warrnambool
- Head of Unit Yellow ANC
- Medical lead Baggarook
- Medical lead homebirth project
- DDU – ultrasound RWH 1 day/week

OUTLINE

- Definitions
- Incidence
  - Differences in parity
- Reasoning behind use of modes
- Risks and benefits
- Controversies
- Useful stats to remember for counselling
- Summary

DEFINITIONS

- NVB
  - Official WHO definition
    - Spontaneous
    - Low risk at start of labour and throughout
    - 37-42 weeks
    - Cephalic
    - No augmentation/instrumentation
    - Mother and baby 'well' after birth
  - Typically at RWH is used to denote a vaginal birth
    - May or may not be 'normal' or uncomplicated
  - Many centres do not require the labour to be SPONTANEOUS for the birth to be considered 'normal'

FOR TODAY – NVB =

- Cephalic
- No instrumentation
- Induced or spontaneous
- Augmented

DEFINITIONS

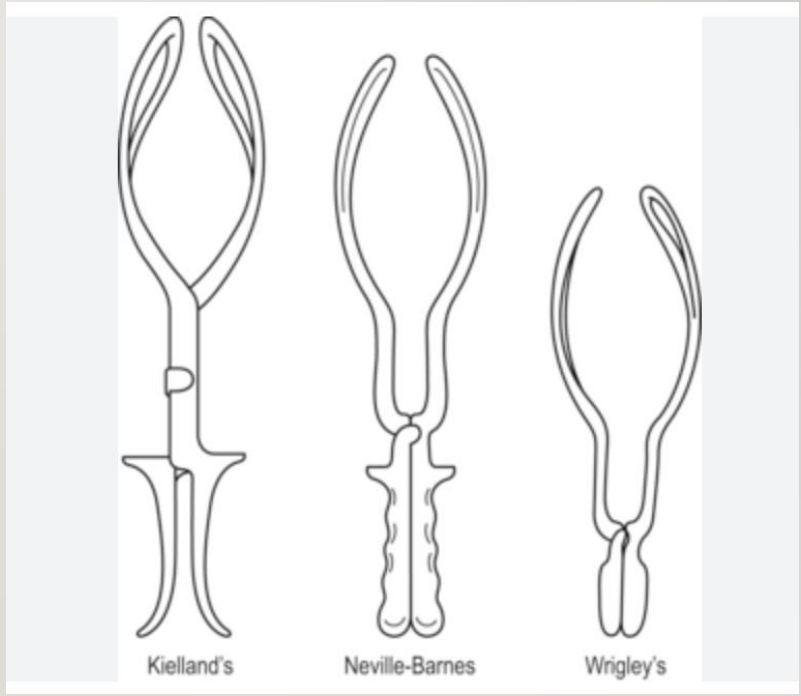
- Instrumental delivery
  - Forceps
    - Neville Barnes
    - Keilland's
  - Vaccuum/ventouse
- Water birth
- Homebirth
- Freebirth
- Caesarean section
  - Emergency vs planned
  - Classical vs LUS
  - Maternally assisted

NEVILLE BARNES FORCEPS

- Requires epidural or pudendal
- OA (or DOP) position
- Episiotomy recommended
- Axis traction handle optional
- Indications
  - Failure to progress at fully dilated
  - Fetal compromise
  - Maternal exhaustion

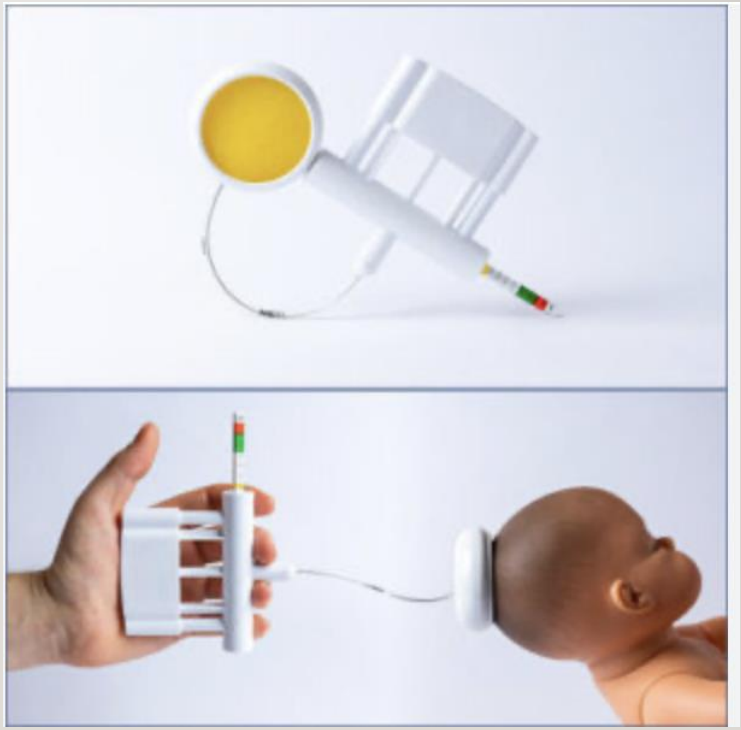
KEILLAND'S FORCEPS

- Requires epidural or pudendal
- Rotational – from OT/OP
- Episiotomy recommended
- Controversial role in obstetrics
- Requires experienced operator
- Indications
  - As for NBF at fully dilated AND malpositioned



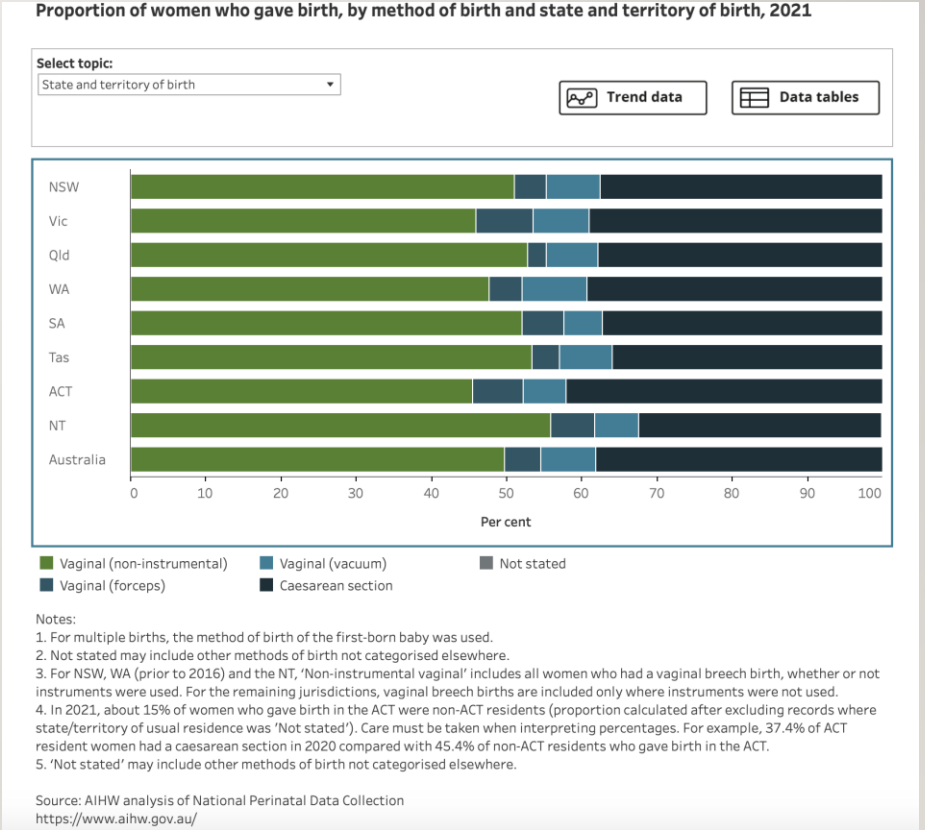
VENTOUSE (VACUUM)

- Can be done with or without epidural – consider pudendal or local infiltration
- Any position as long as position known and cup placed correctly
- Episiotomy as needed – can allow perineum to stretch as per NVB
- Will cause chignon (cephalohaematoma)
- Difficult deliveries can be associated with subgaleal haemorrhage
- Can not be done <34w
- Indications (all at fully dilated)
  - Fetal compromise
  - Maternal exhaustion
  - Delay in second stage



INCIDENCE

- 50% of women had a non-instrumental vaginal birth
- 7.2% of women had a vaginal birth assisted by vacuum
- 4.9% of women had a vaginal birth assisted by forceps
- 38% of women had a caesarean section birth



LOCAL INCIDENCES AT RWH

- NVB – unassisted
  - PG, spontaneous labour 57%
  - PG, induced/no labour 24%
  - MG, spontaneous labour 92%
  - MG, induced/no labour 70%
  - Previous CS 9.4%
  - Breech 11%

LOCAL INCIDENCES AT RWH

- Instrumental delivery
  - PG, spontaneous labour 30%
  - PG, induced/no labour 26%
  - MG, spontaneous labour 6%
  - MG, induced/no labour 7.5%
  - Previous CS 4.6%
  - Breech 0%

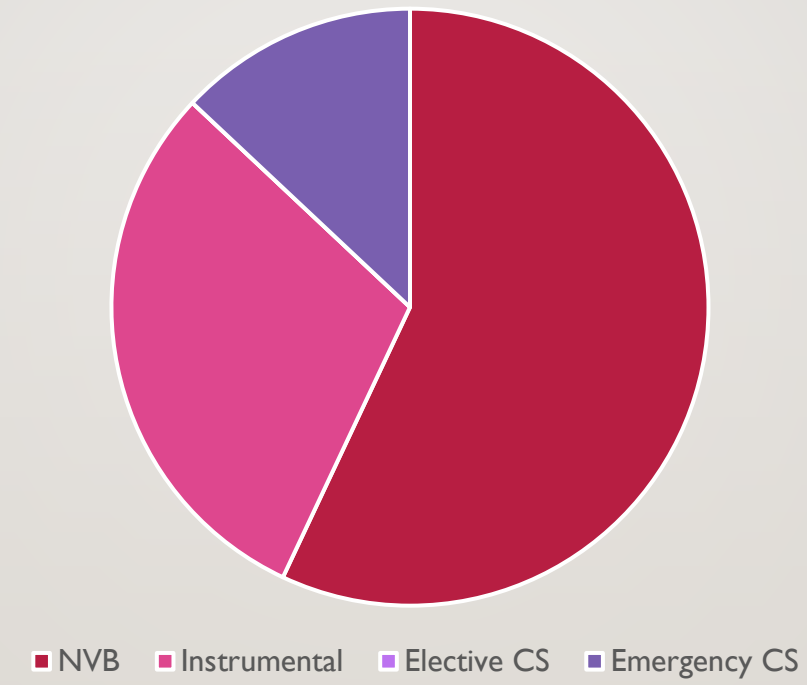
LOCAL INCIDENCES AT RWH

- Elective CS
  - PG, spontaneous labour 0%
  - PG, induced/no labour 12%
  - MG, spontaneous labour 0%
  - MG, induced/no labour 11%
  - Previous CS 63%
  - Breech 49%

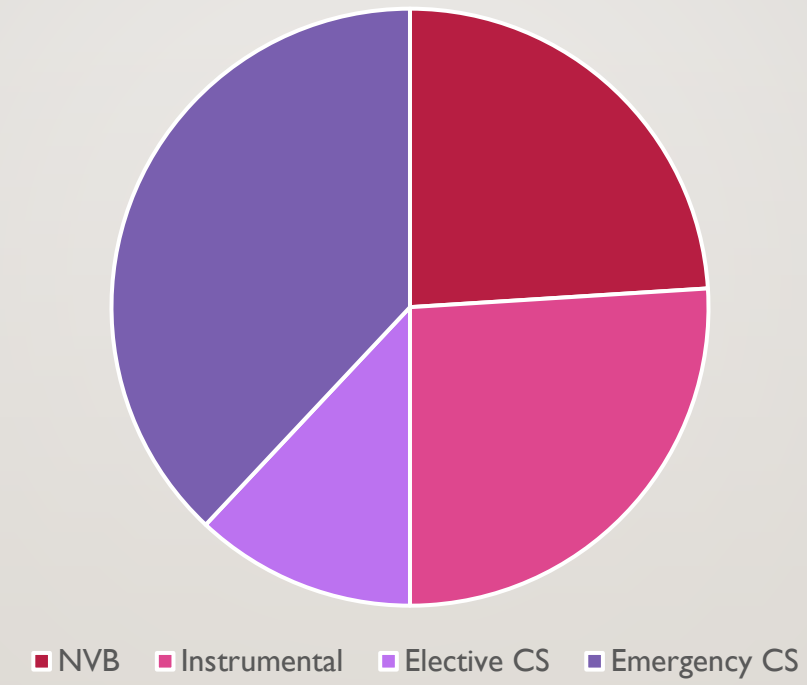
LOCAL INCIDENCES AT RWH

- Emergency CS
  - PG, spontaneous labour 13%
  - PG, induced/no labour 38%
  - MG, spontaneous labour 2%
  - MG, induced/no labour 12%
  - Previous CS 23%
  - Breech 39%

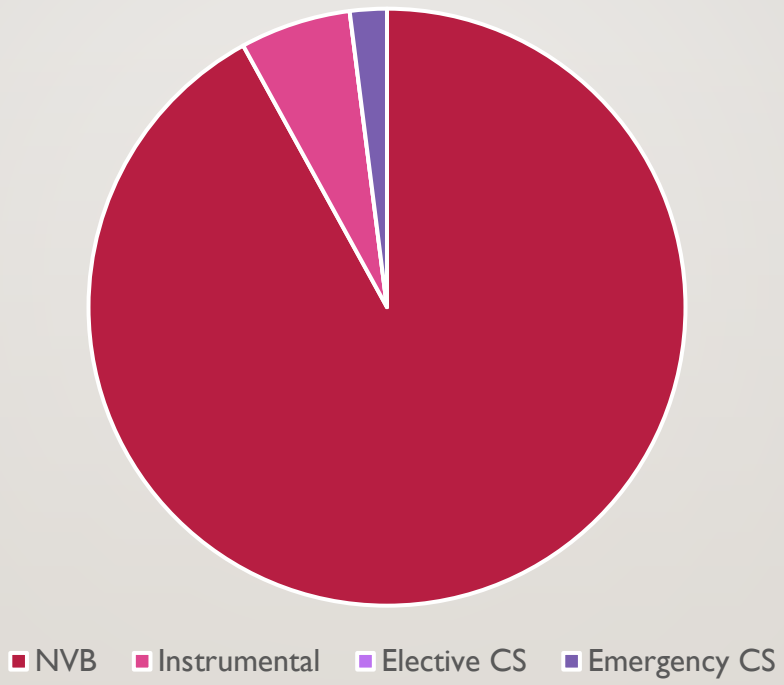
PG IN SPONTANEOUS LABOUR



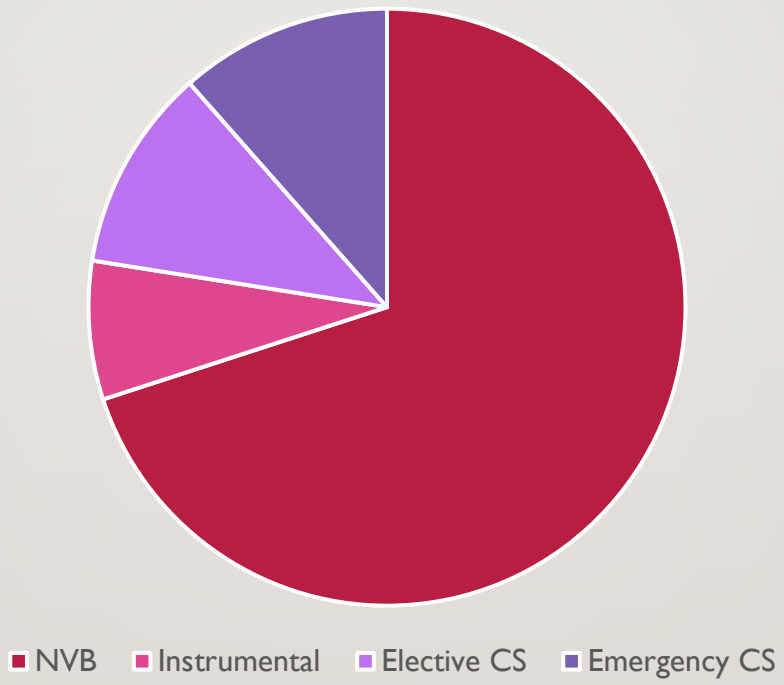
PG – INDUCED OR NO LABOUR



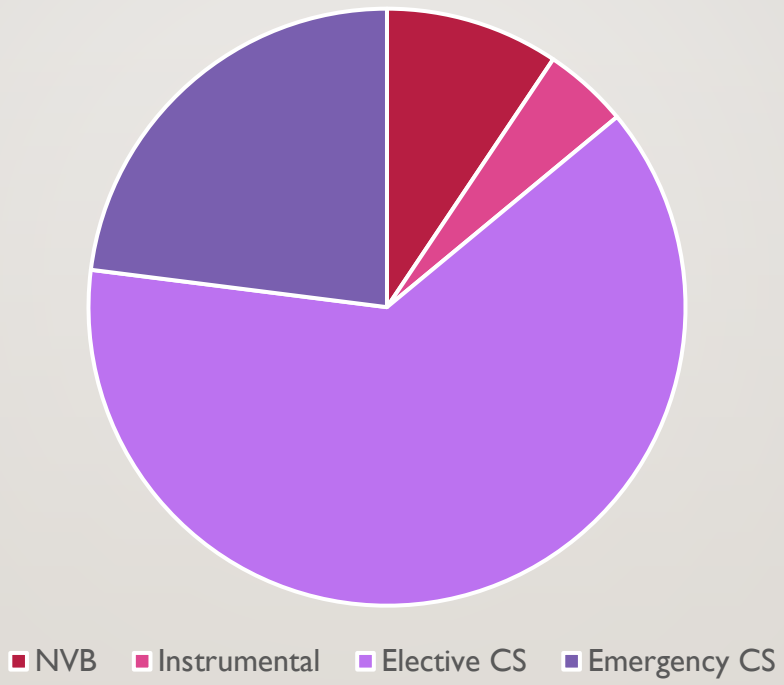
MG IN SPONTANEOUS LABOUR



MG – INDUCED OR NO LABOUR



PREVIOUS CS



65% of women attempting VBAC at RWH have a vaginal birth

REASONS FOR …….

- NVB
  - ‘natural’
  - Quicker recovery
  - Go home sooner
  - Less analgesia required after birth
  - Less risk for future pregnancies
  - ‘Buy yourself a vaginal birth in the future’

REASONS FOR …….

- Homebirth
  - Relaxed, comfortable, private atmosphere
  - Safe (data)
  - ‘Normalises’ birth
  - Can have family by side (kids etc)
  - Save hospital beds
  - Less CS, instrumental, tears, PPH
- Freebirth
  - No medical intervention

REASONS FOR …….

- Elective CS
  - Breech
  - Placenta praevia/vasa praevia
  - Previous CS
  - Multiple pregnancy
  - Suspected macrosomia
  - Obstructing fibroid
  - Maternal choice

REASONS FOR …….

- Emergency CS
  - Non reassuring CTG/abnormal fetal scalp lactate
  - FTP/obstructed labour
  - Abruption
  - Cord prolapse
  - Failed instrumental delivery
- Maternally assisted CS
  - Maternal involvement in birth

RISKS AND BENEFITS

RISKS	BENEFITS
Bleeding/transfusion	Chosen time and date (planned) - controlled
DVT/PE	Less risk HIE to baby
Scar	?Less pelvic floor damage
Infection – UTI/chest/wound	Maternal autonomy
Damage to other structures	Quick – over in an hour
Anaesthetic risks	
Slower recovery	
Risk to future pregnancies – scar rupture, scar ectopic, placenta accreta	

CONTROVERSIES

- Should we consent for VB and what would that look like?
- Should women be 'allowed' to have a CS upon request?
  - Should this be different in private vs public?
- Should we support homebirth?
- What about breech vaginal birth?
- What about pelvic floor protection?

News_

Birth-related PTSD is 'strangely overlooked' in Australia

‘MeToo’ for mothers: Australian inquiry hears troubling accounts of birth trauma

recently developed tool to identify at-risk mothers

By Hilary Whiteman, CNN

7 minute read · Updated 9:27 PM EDT, Fri September 8, 2023

traumati

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A third of new mothers are traumatised by childbirth, but there’s one easy way to help

Rhiannon Lucy Cosslett

Advertisement

Women speak out about birth trauma and poor pelvic health care in public system

By the Specialist Reporting Team's Mary Lloyd and Rhiannon Hobbins

Posted Thu 3 Aug 2023 at 6:04am

Aust N Z J Obstet Gynaecol. 2018 Dec;58(6):701-703. doi: 10.1111/ajo.12885.

We need to treat pregnant women as adults: Women should be consented for an attempt at normal vaginal birth as for operative delivery, with risks and potential complications explained

Hans P Dietz [1], Sascha Callaghan [2]

Affiliations + expand

PMID: 30536511 DOI: 10.1111/ajo.12885

HOW DO WE COMPARE THINGS?

CONSENT FOR VAGINAL BIRTH?

- No – it is a normal 'default' process
- Yes – things can and do go wrong, we consent for less (iron infusions, Anti D)
- Should probably include:
  - Risk of perineal tear (90% PG - RCOG) – 5% 3rd or 4th degree tear
  - Risk of life long prolapse
  - Risk of emergency CS (20-25% PG)
  - Risk of instrumental delivery (15%)
  - Risk of haemorrhage (30%) and possible blood transfusion

WHAT WOULD YOU INCLUDE?

- Should probably include:
  - Need for regular vaginal examinations
  - Possible need for CTG monitoring (belts around belly, no water)
  - Risk of perineal tear (90% PG - RCOG) – 5% 3rd or 4th degree tear
  - Risk of life long prolapse
  - Risk of emergency CS (20-25% PG)
  - Risk of instrumental delivery (15%)
  - Risk of haemorrhage (30%) and possible blood transfusion
  - Risk of fetal hypoxic brain injury (3/1000)

HOW MUCH DETAIL/WHAT DATA DO YOU USE?

- Faecal incontinence
- Urinary incontinence
- Scarring
- Sexual dysfunction
- Retained placenta
- Hysterectomy?
- Shoulder dystocia, meconium aspiration

ARTICLE OF FURTHER INTEREST

- https://jme.bmj.com/content/early/2023/06/11/jme-2022-108283

- Julian Savelescu and team discuss risks of modes of delivery and the ethical framework around maternal decision making in the BMJ

Disclosure and consent: ensuring the ethical provision of information regarding childbirth

Kelly Irvine [1], Rebecca CH Brown [2], Julian Savulescu [3, 4, 5]

Correspondence to Professor Julian Savulescu, Faculty of Philosophy, University of Oxford, Oxford, OX2 6GG, UK;
julian.savulescu@philosophy.ox.ac.uk

PELVIC FLOOR PROTECTION

- CS does not completely remove risk of prolapse/incontinence long term but significantly reduces it
  - OR 9.3 after a single vaginal birth
- Does anything help?
  - Perineal massage
  - Hot perineal compresses
  - Epi-No

USEFUL STATS TO USE IN COUNSELLING

- PG
  - Spontaneous labour
    - 6/10 NVB, 3/10 instrumental, 1/10 CS
  - Induced or no labour
    - ¼ NVB, ¼ instrumental, ½ CS (most of those are emergency – 80%)
  - Overall
    - 1/3 NVB, 1/3 instrumental, 1/3 CS
  - 3rd degree tear rate
    - Unassisted VB 4.5%
    - Instrumental 5.8%
  - VBAC success rate
    - 65%

SUMMARY

- Honest discussion around mode of birth should occur at multiple points of the antenatal journey
- Assess each women's fears and preferences early so detailed discussions can occur
- Encourage child birth education for all women and their partners
  - RWH runs classes for CALD women
  - FPH runs classes in Mandarin

Session Conclusion

You will receive a post session email within a week which will include slides and resources discussed during this session.

Attendance certificate will be received within 4-6 weeks.

RACGP CPD hours will be uploaded within 30 days.

To attend further education sessions, visit,

https://nwmphn.org.au/resources-events/events/

This session was recorded, and you will be able to view the recording at this link within the next week.

https://nwmphn.org.au/resources-events/resources/

We value your feedback, let us know your thoughts.

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