

SATURDAY 1 MARCH 2025Clinical Skills Development Service | RBWH







WELCOME

Dr Meg Cairns
GPLO | Metro North Health & Brisbane North PHN





Metro North Hospital and Health Service and Brisbane North PHN respectfully acknowledge the Traditional Owners of the land on which our services and events are located. We pay our respects to all Elders past, present and future and acknowledge Aboriginal and Torres Strait Islander people across the State.

Acknowledgements

- Metro North Health
- Brisbane North PHN
- Caboolture Hospital, Redcliffe Hospital, Royal Brisbane & Women's Hospital
- Metro North Health Women, Children and Families Clinical Stream
- Metro North Health Outpatient Strategies
- Mater Mothers Hospital GP Alignment Program

Maternity Workshop



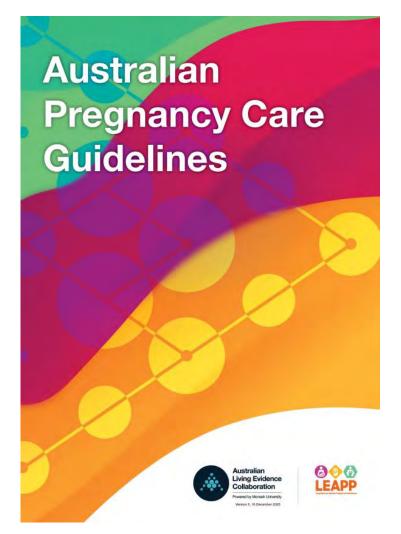
Kindly sponsored by:



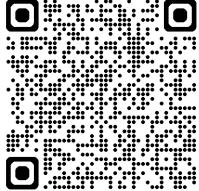


Session 1 Welcome and Orientatation 8:00am Referral pathways Diabetes in pregnancy Case studies: First trimester Epilepsy in pregnancy 10:40am Morning Tea **Session 2** 11:00am **Genetic Screening Multidisciplinary Panel** Case studies: Complex presentations 1:10pm Lunch **Session 3** 1:50pm Interactive skills stations Breast feeding and newborn concerns Healthy eating and exercise in pregnancy, weight gain charts Abdominal palpation, SFH measurement & fetal growth charts Perinatal Mental Health Q+A **Paediatrics** Case studies: Postnatal 4:00pm Evaluation & workshop close

National Guidelines



Guideline Australian Pregnancy Care Guidelines

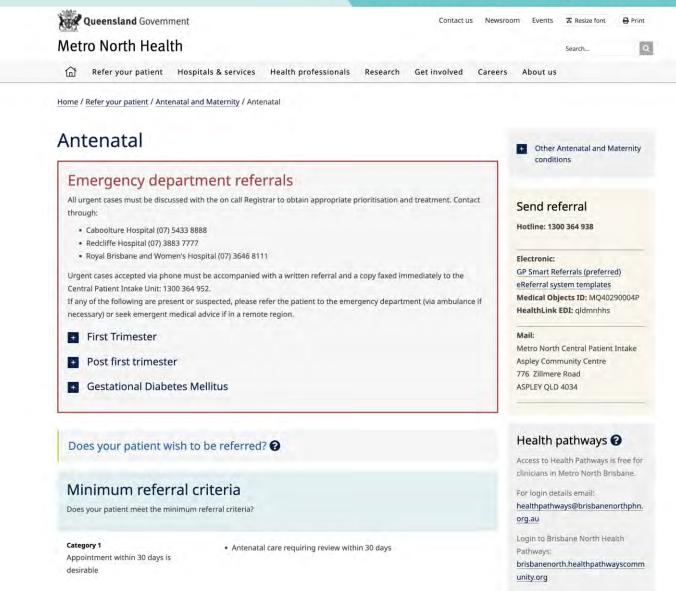


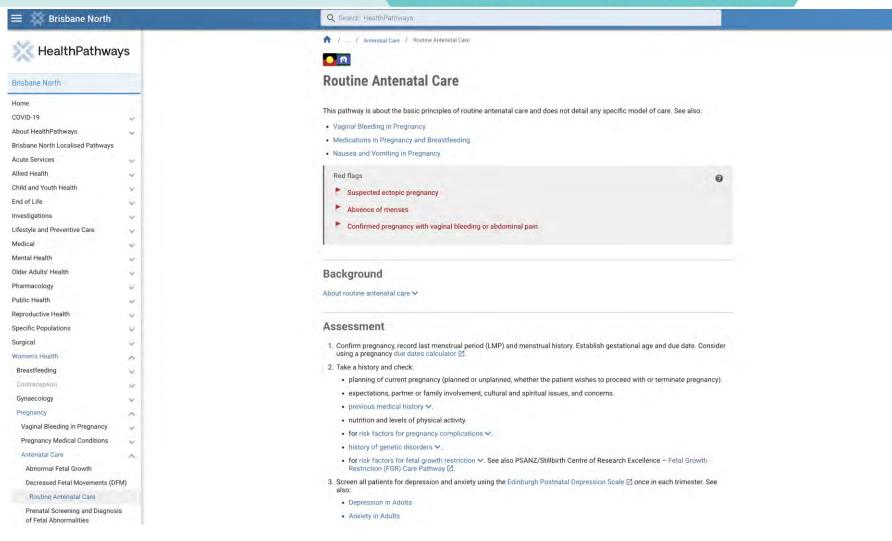
Queensland Clinical Guidelines



Maternity and
Neonatal Clinical
Guidelines |
Queensland Clinical
Guidelines |
Queensland Health |
Queensland Health







Brisbane North HealthPathways <u>Home - Community HealthPathways Brisbane North</u>

Username: Brisbane

Password: North

Useful resources

RACGP clinical guidelines

RACGP - Clinical guidelines

RANZCOG statements & guidelines

Statements and guidelines directory - RANZCOG

Australian Journal of General Practice

RACGP - Home

RACGP gplearning and check

RACGP - Online learning

RACGP Antenatal and Postnatal Specific Interests Group Maternity Moments Webinar Series

RACGP - Specific Interests Groups Resources

Therapeutic guidelines

Therapeutic Guidelines | Therapeutic Guidelines

Choosing Wisely Australia

Recommendations - Choosing wisely

Royal College of Obstetricians and Gynaecologists

Guidance | RCOG

Royal Women's Hospital Victoria

For GPs | The Royal Women's Hospital

Society of Obstetric Medicine of Australia and New Zealand

Guidelines - Society of Obstetric Medicine Australian and NZ

Australasian Diabetes in Pregnancy Society

ADIPS

Australasian Society for Infectious Diseases

ASID | Australasian Society for Infectious Diseases

Stillbirth Centre for Research Excellence

Stillbirth CRE Home | The Centre of Research Excellence in Stillbirth

Safer Baby Bundle

Home | Stillbirth CRE eLearning

Australian Preterm Birth Alliance

The Australian Preterm Birth Prevention Alliance - APBPA

Genetic Health Queensland

Genetic Health Queensland Resources for Health Professionals

RACGP Guidelines Genomics in General Practice

RACGP Guidelines | Genomics in General Practice

NSW Health Centre for Genetics Education

NSW Health | Centre for Genetics Education

Australian Preterm Birth Alliance

The Australian Preterm Birth Prevention Alliance - APBPA

COPE Centre of Perinatal Excellence

COPE | Perinatal Mental Health Resources for Health Professionals

SMS4Dads sms4dads

PANDA Perinatal Anxiety & Depression Australia

Home | Perinatal Anxiety & Depression Australia

Peach Tree Perinatal Wellness

Home | Peach Tree Perinatal Wellness

GP Psychiatry Support Line

GP Psychiatry Support Line

Gidget Foundation Australia

Home | Gidget Foundation Australia



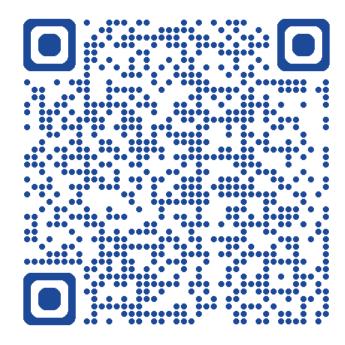
REFERRAL PATHWAYS

Dr Meg Cairns GPLO | Metro North Health & Brisbane North PHN

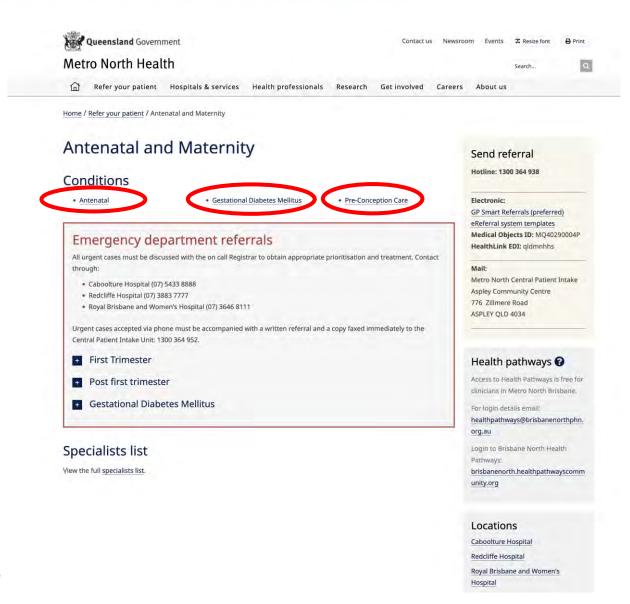




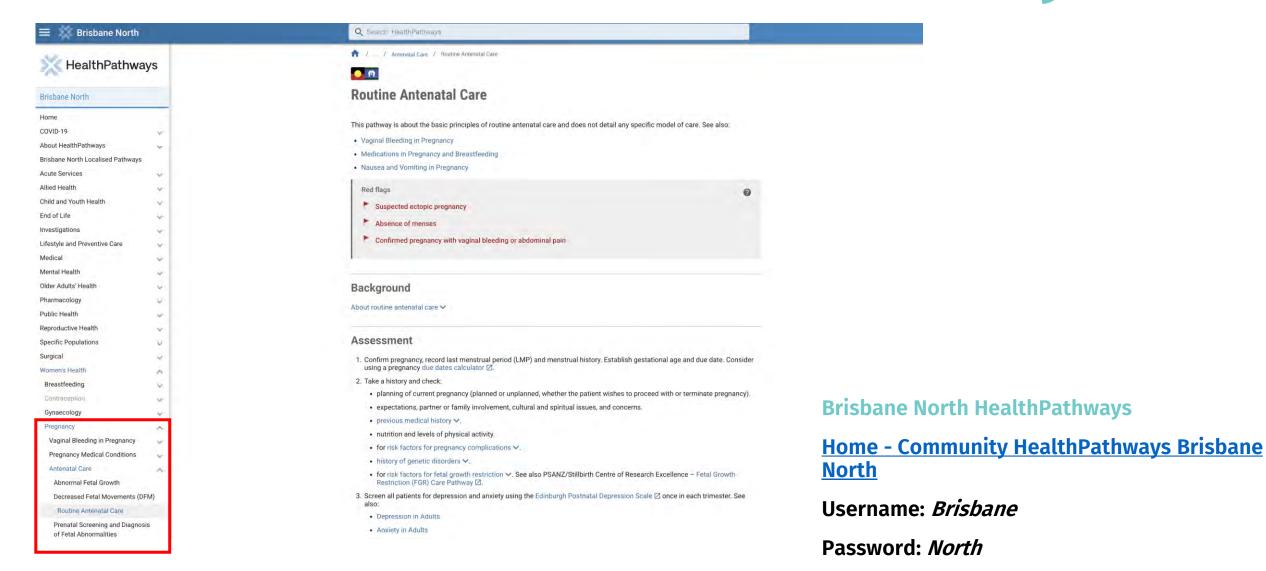
Refer Your Patient



Metro North Antenatal and Maternity Referral Guidelines
Antenatal and Maternity | Metro North Health



Brisbane North HealthPathways



Metro North Antenatal Shared Care

Pre-conception

- Folate and iodine supplementation
- Rubella serology +/vaccination
- Varicella serology if no history +l- vaccination
- COVID-19 vaccination
- Influenza vaccination
 Cervical Screening Test if
- Chlamydia if age <30
- Smoking cessation
- Alcohol cessation
- Discuss genetic carrier screening
- If significant medical, genetic, psychological illness that impacts preconception, gestation or birth refer to preconception clinic &/or genetics service

First GP Visit(s)

(may require more than one consultation)

- Confirm pregnancy and dates
- Scan if dates uncertain or risk of ectopic (previous ectopic, tubal surgery)
- · Folate and iodine supplementation for all
- Review medical/surgical/psych/family/obstetric history,
 madications, allegates at a conductor CR records.
- medications, allergies etc + update GP records
 Identify risk factors for pregnancy
- Discuss genetic carrier screening
- Order first trimester screening tests
- Perform physical examination as per Pregnancy Health Record (PHR)
- Weight, BMI discuss healthy weight gain, nutrition and physical activity
- Discuss smoking, alcohol, other drugs, Listeria, Toxoplasmosis etc.
- COVID-19 vaccination
- Influenza vaccination
- Discuss models of care
- Complete referral early indicate if high risk, you wish to share care, or preference for Birth Centre (RBWH) or Midwifery Group Practice
- Send GP Smart Referral or eReferral to Central Patient Intake (CPI)
- · Ask woman to complete online registration

First Trimester screening tests (GP)

(cc ANC on all request forms) – all requests to be reviewed and actioned by referring clinician

- FBC, ferritin, blood group and antibodies, Rubella, Hep B, Hep C, HIV, syphilis serology + dry swab (PCR) if lesions/chancre present, MSU (treat asymptomatic bacteriuria)
- Chlamydia if <30 or area of high prevalence.
- If risk factors for GDM, OGTT (or HbA1c if OGTT not tolerated)
- ELFTs, TFT, Vit D for specific indications only
 Varicella serology (if no history of Varicella or vaccination)
- . Cervical Screening Test if due
- · Discuss/offer genetic carrier screening
- . Discuss/offer prenatal screening
 - 1. Nuchal translucency scan + first trimester screen (free B-hCG, Papp-A) K11-13+6 or
 - 2. Triple test (AFP, estriol, free B-hCG) K15-20 if desired or if presents too late for first trimester testing (not twins or diabetes)
- NIPT > K10 (not Medicare funded); anatomical scan at K13 still recommended
- Discuss and refer for CVS/amniocentesis if appropriate

Uncomplicated Pregnancy

- Rh D NIPT to predict fetal RhD status in non-alloimmunised RhD negative pregnant women from K15
- 18-20 week morphology scan including cervical length measurement
- Arrange to see woman after scan
- Cervix length if TA cervix length <35mm, a TV USS should be performed. If TV cervix length <25mm, commence vaginal progesterone (200mg nocte from 16-36 weeks) and refer to MFM.
- First ANC visit with midwife K16-20
- Obstetrician review if required
- All investigations to be reviewed and followed up by referring clinician
- · Other referrals if applicable

GP visits

- Schedule as per PHR or specific facility
- . More frequent if clinically indicated
- · Record in PHR
- Assessment/education as per PHR
 K24-28: OGTT, (if + refer to ANC), FBC. If Rh negative: blood group/antibodies screen; offer Anti-D
- . K26-28: repeat syphilis serology
- Pertussis vaccine K20-32 in each pregnancy
- K28-36: RSV vaccine
- . K34: If Rh neg offer Anti-D
- . K36: FBC, repeat syphilis serology
- Dry swab (syphilis PCR) at any stage if lesions/chancre present

ANC visits

- K36
- K41

Contacts	RBWH	Caboolture	Redcliffe
For referral or advice			
GP Liaison Midwife	3647 3960 3646 1305	5433 8800	3049 2301
O&G Registrar on call	3646 8111	5433 8120	3883 7777
Obstetric Medicine Registrar	3646 8111	-	-
Perinatal Mental Health (Metro North)	3146 2525 or perinatal-mental-health@health.qld.gov.au		
Pregnancy complications			
<20 weeks: Care of complications e.g. bleeding, pain, threatened or incomplete miscarriages	3646 8111 O&G Registrar on call	5433 8120 O&G Registrar on call	3883 7777 Early Pregnancy Assessment
<20 weeks: haemodynamically unstable women	3646 8111 DEM	5433 8888 ED	3883 7777 ED
>20 weeks: complications (RBWH > K14)	3647 3931 Obstetric Review Centre	5433 8670 Birth Suite	3883 7714 Birth Suite

Additional Information

High risk for GDM?

- Previous GDM or baby >4500g or >90th centile; previous elevated BGL; PCOS; FHx; BMI >30; maternal age ≥40; previous perinatal loss; multiple pregnancy; high risk ethnicity; medications: corticosteroids, antipsychotics
- First Trimester OGTT or HbA1C
- Post bariatric surgery OGTT not suitable. First trimester HbA1c or fasting BGL if diabetes history or risk feature.
- Urgent hospital ANC referral if abnormal
- Specify reason and include results in referral. Send GP Smart Referral or eReferral to CPIU

Rh negative?

- RhD NIPT to predict fetal RhD status through Lifeblood or SNP
- Offer Anti-D:
- 28 and 34 weeks
- Sensitising events
- For details and dosages, refer to https://www.blood.gov.au/quidelineprophylactic-use-rh-dimmunoolobulin-pregnancy-care.

Medical condition or obstetric complications? Early/urgent hospital ANC

- GP referrals are promptly triaged.
- Please specify urgency and reasons in referral
- Send GP Smart Referral or eReferral to CPIU



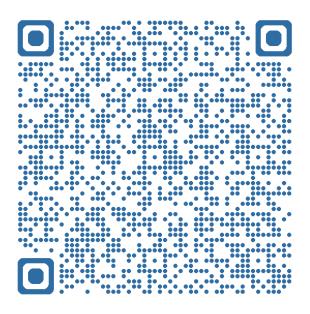


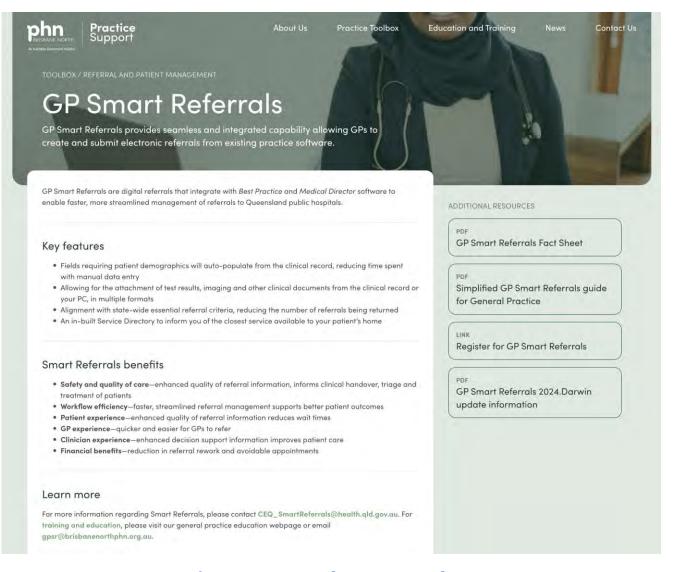


Modified by Brisbane North PHN, MNHHS and Mater Mothers' Hospital from an original created by Drs Michael Rice, Mano Haran and Heng Tang.
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Version 7 Effective: 02/2025 Review: 02/2026

GP Smart Referrals





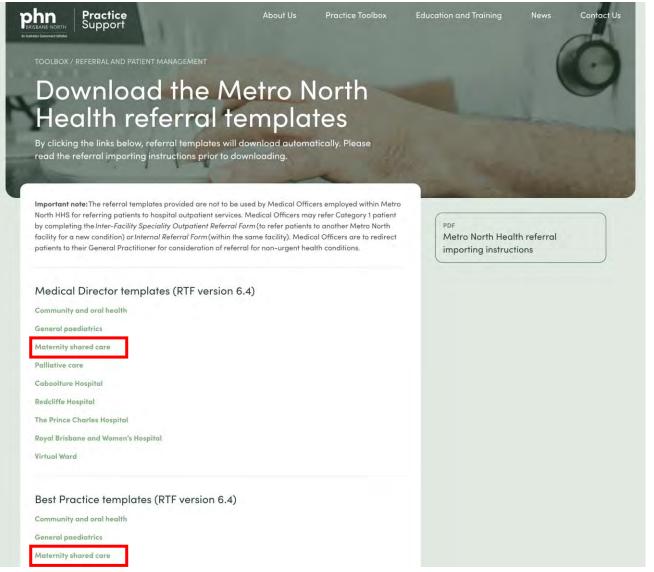
Practice Support | Toolbox | GP Smart Referrals

GP Smart Referrals

Condition and Specialty	Midwifery and Maternity - Antenatal (Antenatal) (Adult) HealthPath	ways ►	
Suitable for Telehealth?	Yes No		
Are you the patient's usual GP?	Yes No		
Request recipient		-	
* Service/Location	Please select		
Specialist name	Please select 💌		
Organisation details			
Condition specific clinical information		-	
Show emergency referral criteria	Show Hide		
Minimum Referral Criteria			
* Minimum referral criteria	Antenatal care requiring review within 30 days Antenatal care requiring review within 90 days Request clinical override of minimum referral criteria		
Clinical Details - Current Pregnancy			
Woman's preferred MOC	GP shared care Midwifery care Obstetric care		
Current pregnancy	Single Multiple		
Date of LNMP	[7]		
Estimated date of birth (EDB)			
Blood group	A		
Rhesus status	Rh negative		
BMI			
Blood pressure			
Cervical length (after 16 weeks, if known)			

Practice Support | Toolbox | GP Smart Referrals

Metro North eReferral Template



Practice Support | Toolbox | GP Referral Templates

Antenatal Referrals

- Confirm pregnancy and EDB
- Confirm Medicare eligibility
- Indicate preferred Maternity Care Option on referral
 - if requesting Birth Centre (RBWH) or Midwifery Group Practice, include in referral
- Send referral to CPI
 - GP Smart Referral
 - eReferral
 - enquiries 1300 364 938

Antenatal Referrals

- Include copies of available results with referral
- <u>All</u> pathology & USS results must be <u>reviewed and</u> <u>actioned</u> by requesting practitioner
- Advise woman to follow-up results with you and attend regularly for antenatal visits (every 4 weeks in Trimesters 1 & 2)

Antenatal Referrals

- Advise woman to visit Hospital websites for more information regarding maternity services
 - RBWH | Pregnant what to do next
 - Redcliffe Hospital | Pregnant what to do next
 - Caboolture Hospital | Pregnant what to do next
- Online registration is available at all Metro North Maternity Facilities

- First Appointment
 - "booking-in" appointment will be completed prior to 18 weeks

Pregnancy Health Record

Pregnancy Health Record Clinician's section	(Affix identification URN: Family name: Given name(s): Address: Medicare number: Date of birth:	on label here)		
Attach ADR Sticker ALLERGIES AND ADVERSE DRUG REACTIONS (ADR) Nil known Unknown (tick appropriate box or complete details below)	Model of care (complete details page	e a10):	Rh D negati Yes N See page a10 Rh D Immuno	lo for
Drug (or other) Reaction / Date Initials	Medicare ineligible – comments:			
	Religious, ethnic or cultural conside			natal
Sign: Date:	Religious, ethnic or cultural considerate (e.g. birth practices, blood productions)			natal
Sign: Date:		cts, dietary, et		natal
Woman's Information Preferred name:	care (e.g. birth practices, blood produc	cts, dietary, et	c.): arital status;	natal
Woman's Information Preferred name: Country of birth: Australia Other:	care (e.g. birth practices, blood produc	cts, dietary, et	c.): arital status; sustralia?	
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Woman's Information Preferred name: Country of birth: Australia Other: Do you have refugee status experience? Interpreter required? Yes No	Age: If Other, what year did	years Ma	c.): arital status; sustralia?	
Woman's Information Preferred name: Country of birth: Australia Other: Do you have refugee status experience? Interpreter required? Yes No If Yes, Language.	Age: If Other, what year did	years Ma	c.): arital status; sustralia?	N
Woman's Information Preferred name: Country of birth: Australia Other: Do you have refugee status experience? Interpreter required? Yes No If Yes, Language: Do you have any problems reading English and understand Are you of Aboriginal and/or Torres Strait Islander origin?	Age: If Other, what year did Ethnicity: ing the content of this Pregnancy Healt Occupation.	years Ma	c.): arital status; sustralia?	N

Clinical Excellence Queensland | Pregnancy Health Record

Pregnancy Health Record Initial Physical Examination (PHR Page a8 of 20)

		URN: Family name: Given name(s): Addrass: Medicare number; Date of birth:	(Affix identification label here)	
Initial Physical E	xamination weight if known, otherwi	se use first weight taken	To be completed by a Medical Officer / Midwife	
Date: / / Sooking-in weight.	Pre-pregnancy weight:		Breasts / Nipples:	
kg	kg	meigni:	Cardiovascular:	
Pre-pregnancy BMI:	Underweight (s18.5) Normal (18.5-24.9) Overweight (25-29.9) Obese I (30.0-34.9)	Referral to Medical Officer Dietilan for review Physio for review	Respiratory:	
6 week kg/BMI: kg/ BMI	Obese II (35.0-39.9) Obese III (≥40) Underweight (≤18.5) Normal (18.5-24.9) Overweight (25-29.9)	Referral to Medical Officer Dietilan for review	Abdominal:	
Dental:	Obese II (30.0–34.9) Obese II (35.0–39.9) Obese III (>40)	Physio for review	Skeletal:	
Last appointment:	1_1_		Thyroid:	
Name:		Designation:	Signature:	

Clinical Excellence Queensland | Pregnancy Health Record

Responsibility of referring GP regardless of woman's requested maternity care option

Routine Antenatal Tests

Metro North Antenatal Shared Care

Pre-conception

- Folate and iodine supplementation
- Rubella serology +/vaccination
- Varicella serology if no history +/- vaccination
- COVID-19 vaccination
 Influenza vaccination
- Cervical Screening Test it
- Chlamydia if age <30
- Smoking cessation
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- Discuss genetic carrier screening
- If significant medical, genetic, psychological illness that impacts preconception, gestation or birth refer to preconception clinic &/or genetics service

First GP Visit(s)

(may require more than one consultation)

- · Confirm pregnancy and dates
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 Review medical/surgical/psych/family/obstetric history,
- medications, allergies etc + update GP records
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- Discuss genetic carrier screening
- Order first trimester screening tests
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- Discuss smoking, alcohol, other drugs, Listeria,
- Toxoplasmosis etc.
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- Influenza vaccination
 Discuss models of care
- Complete referral early indicate if high risk, you wish to share care, or preference for Birth Centre (RBWH) or Midwifery Group Practice
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- · Cervical Screening Test if due

vaccination)

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- Discuss/offer prenatal screening
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- · K34: If Rh neg offer Anti-D
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ANC visits

- K36
- K41

Contacts Redcliffe For referral or advice GP Liaison Midwife 3647 3960 5433 8800 3049 2301 3646 1305 3883 7777 O&G Registrar on call 3646 8111 5433 8120 Obstetric Medicine Registrar 3646 8111 Perinatal Mental Health 3146 2525 or perinatal-mental-health@health.qld.gov.au (Metro North) **Pregnancy complications** <20 weeks: Care of 3646 8111 5433 8120 3883 7777 complications e.g. bleeding, pain, O&G Registrar on O&G Registrar on Early Pregnancy threatened or incomplete call Assessment miscarriages <20 weeks: haemodynamically 3646 8111 5433 8888 3883 7777 DEM ED ED unstable women >20 weeks: complications 3647 3931 5433 8670 3883 7714 (RBWH > K14) Obstetric Review Birth Suite Birth Suite Centre

Modified by Brisbane North PHN, MNHHS and Mater Mothers' Hospital from an original created by Drs Michael Rice, Mano Haran and Heng Tang

Additional Information

High risk for GDM?

- Previous GDM or baby >4500g or >90th centile; previous elevated BGL; PCOS; FHx; BMI >30, maternal age 240; previous perinatal loss; multiple pregnancy, high risk ethnicity; medications: corticosteroids, antisoxybotics
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- Post bariatric surgery OGTT not suitable. First trimester HbA1c or fasting BGL if diabetes history or risk factors
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- Specify reason and include results in referral. Send GP Smart Referral or eReferral to CPIU

Rh negative?

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- · Offer Anti-D:
- 28 and 34 weeks
- Sensitising events
- For details and dosages, refer to https://www.blood.gov.au/quidelineprophylactic-use-rh-dimmunoqlobulin-pregnancy-care.

Medical condition or obstetric complications?

Early/urgent hospital ANC referral?

- · GP referrals are promptly triaged
- Please specify urgency and reasons in referral
- Send GP Smart Referral or eReferral to CPIU



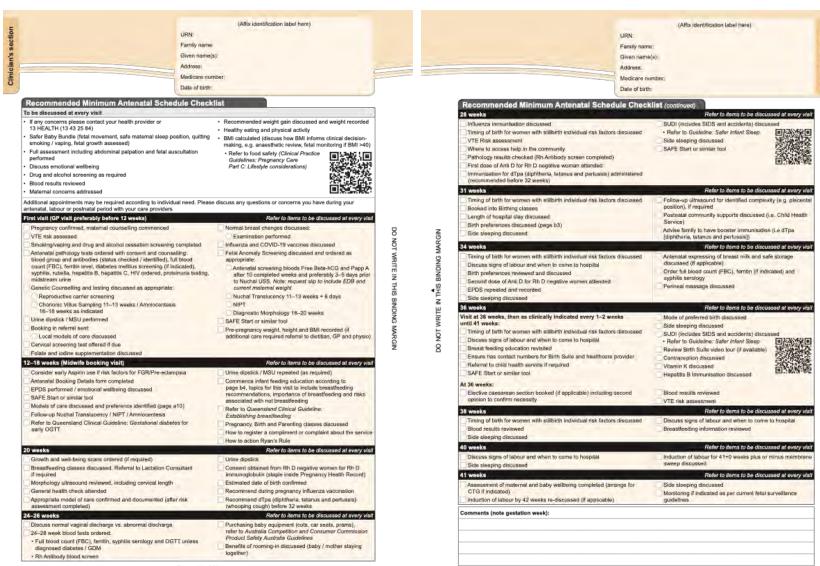




Metro North Antenatal Shared Care

This is a joint initiative between Metro North Hospital and Health Service and Brisbane North PHN

Antenatal Visit Schedule



Metro North Perinatal Mental Health

- Metro North HHS Perinatal Mental Health Service (Non-Acute)
 - https://metronorth.health.qld.gov.au/hospitalsservices/mental-health-services/perinatal-mental-health
 - P: 07 3146 2525
 - F: 07 3146 2314
 - E: perinatal-mental-health@health.qld.gov.au
 - Perinatal Psychiatrist Dr Anastasia Braun fax referral
 07 3646 2314
- 1300 MH CALL (1300 64 2255) (Acute)



Home / Healthcare Services / Maternity Services

Maternity Services



Pregnancy

Choosing an option for maternity

Maternity Services Referral Catchment

Tests and scans

<u>Learning about pregnancy, birth and baby</u>

Pregnancy problems



Having your baby

Preparing for labour

Labour and birth

When complications occur

Care after birth

While you're in hospital

Newborn Bloodspot Screening



Think you might be in labour?

Call (07) 3647 3931 and speak to a midwife before you come to hospital

Contact us

Maternity outpatient

appointments

Location: Ground floor, Ned Hanlon

Building

Phone: (07) 3646 7182
Email: rbwh_maternity.
@health.qld.gov.au

Open: Monday-Friday 8.00am-

4.00pm

Birth Suite and Birth Centre

Location: Level 5, Ned Hanlon

Building

Phone: (07) 3646 8516 or

(07) 3646 8317

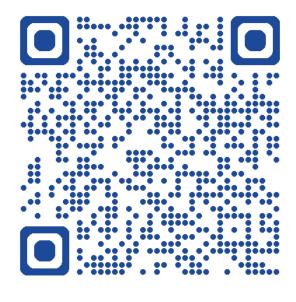
Women's Obstetric Review Centre

Location: Level 5, Ned Hanlon

Building

Phone: (07) 3647 3931

Private practice appointments



RBWH | Maternity Services



Home / Healthcare Services / Maternity Services / Maternity Services Referral Catchment

Maternity Services Referral Catchment

To facilitate supporting families closer to home, from October 2021 the RBWH will not be accepting referrals from Brisbane Metro South and West Moreton. This will apply to all models of care currently offered with the exception of the below.

The exclusions include:

- The acceptance of all referrals for Aboriginal and Torres Strait Islander women (i.e. Ngarrama) who would like maternity care at RBWH to support the 'Closing the Gap' initiative
- · Women requiring tertiary care at RBWH due to pre-existing medical conditions which are currently managed at RBWH
- · Complex maternal cardiac conditions occurring in pregnancy
- · Women under the care of Private Practice Midwives credentialled at RBWH; and
- General medicine / Obstetric medicine telehealth referrals







Home / Healthcare Services / Maternity Services / Choosing an option for maternity care

Choosing an option for maternity care

All <u>options for maternity care (PDF)</u>are delivered by caring and dedicated health professionals in partnership with you and your support people. Your general practitioner (GP) or midwife will discuss these options at your <u>first appointment</u>.



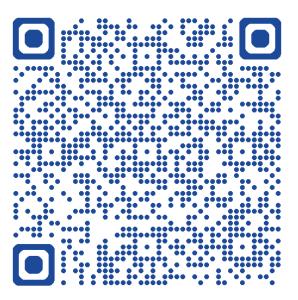
Choosing the best option for you will depend on your own personal preferences, and sometimes, your medical history. We offer three main options for maternity care for your pregnancy, birth and after your baby is born:

- Midwifery care
- · GP shared care
- Specialist care

All care options have the opportunity for discharge home at **6 hours** after birth, if you have a normal birth and you and your baby are well. If you need to stay longer, you can expect to be discharged around **24 hours** following a normal birth or within **72 hours** after a caesarean birth.

Maternity outpatient appointments Location: Ground floor, Ned Hanlon Building Phone: (07) 3646 7182 Email: rbwh maternity @health.gld.gov.au Open: Monday-Friday 8.00am-4.00pm **Private practice appointments** Phone: (07) 3646 3395 Refer a patient Maternity outpatient Complete the Maternity booking in referral form (PDF) and forward it to Metro North Central Patient Intaketo refer your patient.

Contact us



RBWH | Choosing an option for maternity care



Home / Healthcare Services / Maternity Services / Learning about pregnancy, birth and baby

Learning about pregnancy, birth and baby

Learning about your pregnancy, birth and baby can help build your confidence and prepare you for the weeks ahead. The RBWH has resources and experienced staff available to ensure you're supported throughout your journey.

Nurture Your Bump - Workshop



Unsure of what foods you need to avoid during pregnancy or if you need a pregnancy multivitamin? Our 2-hour Nurture Your Bump workshop, is run by our experienced maternity dietitian and will provide you with all the building blocks needed to grow a healthy baby. Book your workshop instantly online or call RBWH Maternity Outpatients Department on (07) 3646 7182.

Register or refer now >

GLOW (online resource)

<u>GLOW (PDF)</u> is a free online resource, full of helpful and factual information about pregnancy, breastfeeding, birth and going home with a newborn. Access to GLOW is offered for all women having their baby at RBWH and includes the following topics:

Contact us

Maternity Outpatients

Location: Ground floor, Ned Hanlon

Building

Appointment enquiries

Phone: (07) 3646 7182 Email: rbwh maternity @health.qld.gov.au

Open: Monday-Friday 8.00am-

4.00pm

Private practice appointments

Phone: (07) 3646 3395

Refer a patient

Complete the <u>Maternity booking in</u> referral form (PDF) and forward it to <u>Metro North Central Patient Intake</u> to refer your patient.

RBWH | Learning about pregnancy birth baby

Learning about pregnancy, birth and baby



- Free online resource for women having their baby at RBWH
- Women opt-in at bookingin visit
- Access 24/7 from home computer, tablet or smartphone



Other RBWH Services

Early Pregnancy Assessment Unit (EPAU)	Obstetric Review Centre (ORC)
Pregnancy, birth & baby education	Gestational Diabetes Mellitus midwives
Postnatal in-home visiting following discharge	Complex Case Manager (Inc. Obstetric Medical Team)
Gynaecology, Urogynaecology, Gynaecology Oncology, Adolescent Gynaecology 14-18yrs	Specialist Clinics including Anaesthetics, Cardiac and Endocrine
Social Work including Child Protection Liaison Officer	Centre for Advanced Prenatal Care (Maternal Fetal Medicine)
Allied Health	Fertility
Perinatal Mental Health	OASIS (Obstetric Anal Sphincter Injuries)
Lactation Service	Centre for Breast Health
Grantley Stable Neonatal Unit	



Home / Healthcare Services / Maternity services

Maternity services



Pregnancy

Pregnant? What to do next

Choosing an option for your maternity care

Tests and scans

<u>Learning about pregnancy, birth and baby</u>

Valledan

Your appointments



Having your baby

Preparing for labour

Labour and birth

When complications occur



Complete the <u>online</u>
registration form to
start the booking
process

Contact us

Antenatal Clinic

Location: Rear of the hospital,

access via Silvyn Street

Phone: (07) 3883 7802

Birth Suite

Location: Level 3, Main Building,

Redcliffe Hospital

Phone: (07) 3883 7714

Childbirth and Parenting

Education

Location: Education Centre, Redcliffe

Hospital

Phone: (07) 3883 7802

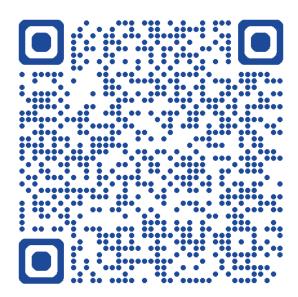
Open: Please call 1.00pm-4.00pm

Monday-Friday

Home Maternity Service

Phone: (07) 3883 7709







Home / Healthcare Services / Maternity services / Choosing an option for your maternity care

Choosing an option for your maternity care

All options for maternity care are delivered by our caring and dedicated health professionals in partnership with you and your support people. Your GP or midwife will discuss these options with you.

Maternity care options

Choosing the best option for you will depend on your own personal preferences, and sometimes, your medical history.

- Midwives clinic + CRIB Clinic complex MH & psychosocial issues Redcliffe & Deception Bay
- Midwifery Group Practice AMITY
- Private Practice Midwives
- Aboriginal and Torres Strait Islander Maternity Service Ngarrama Redcliffe & Deception Bay
- Young Parent Group
- Obstetric led care with Doctors and Midwives
- GP Shared Care

Contact us

Location: Antenatal Clinic, Redcliffe

Hospital

Phone: (07) 3883 7802



Complete the antenatal online registration form



Home / Healthcare Services / Maternity services / Learning about pregnancy, birth and baby

Learning about pregnancy, birth and baby

Learning about your pregnancy, birth and baby can help build your confidence and prepare you for the weeks ahead. Redcliffe Hospital has resources and experienced staff available to help you throughout your pregnancy.

Childbirth and Parenting Education

We offer classes with experienced staff, who will answer any of your questions about your pregnancy, birth and parenting. If you have any questions outside of these classes, please ask your health care provided.

To book these classes please ring (07) 3883 7802 between 1,00pm-4,00pm Monday-Friday.

Birth and parenting classes

Evening classes

When: Monday or Thursday evenings from 6.30pm-8.30pm. You can choose which evening to attend.

Located: Education Centre, Redcliffe Hospital

Saturday classes

When: Saturday 9.00am-2.30pm (please note that these classes are on two consecutive Saturdays each month)

Located: Education Centre, Redcliffe Hospital or North Lakes Health Precinct.

Young Parent Group (YPG)

When: Every second Tuesday from 1.00pm-3.00pm

Located: Community Health, Anzac Avenue, Redcliffe

Emotional preparation for parenthood classes

Emotional health is just as important as physical health. A combined team of health professionals and peers outline some of the emotional challenges of pregnancy, birth and adjustment for parenthood. Information is provided about practical resources to support your own and your partner's emotional wellbeing during this time.

Breastfeeding classes

Breastfeeding classes are recommended for all women who are having their first baby or have experienced problems breastfeeding. Classes are run by a midwife and are designed to provide consistent advice and support for you to make an informed decision about breastfeeding. The class teaches you practical skills and tips to successfully breastfeed your baby. Partners and a support person (friend, mum, sister, aunt) are encouraged to attend.

When: Monday or Thursday 6.30pm-8.30pm Located: Education Centre, Redcliffe Hospital

Physiotherapy classes

Come along to the physiotherapy class to help with problems such as leaking when you cough or sneeze, difficulty sleeping, general aches and pains in your pregnancy or if you are interested in safe exercise.

When: Monday 6.30pm-8.30pm Located: Education Centre, Redcliffe Hospital

Contact us

Childbirth and Parenting

Education

Hospital

Location: Education Centre, Redcliffe

Phone: (07) 3883 7802

Maternity tour

Location: Birth Suite, Level 3, Main

Building, Reddiffe Hospital Phone: (07) 3883 7714

Resources

Raising Children

Mutrition while pregnant

Emotional support

Pregnancy, birth and early parenthood are times of great change. Most women experience various emotional reactions. It helps if you can talk about your concerns openly with your partner or close friend. We are able to provide support and treatment through our expert clinicians.

There are also many organisations that offer support. If you are experiencing signs of depression, there are a number of places you can go for help. A list of these services are below:

Perinatal mental health

- Post and Antenatal Depression Association (PANDA) 1300 726 306
- beyondblue's Just Speak Up 1300 22 4636
- White Cloud Foundation 1300 726 306

Mental health support

- . Black Dog Institute
- Lifeline 13 11 14 (24 hours a day)
- Sane Australia 1800 18 7263
- MensLine Australia 1300 789 978 (24 hours a day)

Pregnancy support

- · Pregnancy, Birth and Baby
- 13 HEALTH 13 43 25 84
- Women's Health Queensland Wide
- Pregnancy Helpline 1800 090 777 (if you have an unplanned pregnancy and want to discuss your options with a qualified counsellor)
- · Perinatal Mental Health

Alcohol

The safest choice for your baby is to not drink any alcohol. This is advice from the <u>National Health and Medical Research Council</u> of Australia.

Smoking

It is recommended to stop smoking during pregnancy. We offer support for you to stop or reduce the amount you smoke. Ask your doctor or midwife during your antenatal appointment. You can also call Quittine on 13 78 48.

Smoking while pregnant increases your risk of

- ectopic pregnancy
- miscarriage

Redcliffe Hospital | Learning about pregnancy birth baby

Other Redcliffe Hospital Services

Early Pregnancy Assessment Unit (EPAU)	Antenatal Day Assessment Service (ANDAS) Obstetric Review Centre (ORC)
Pregnancy, birth & baby education	Gestational Diabetes Team Credentialed Diabetes Educator
Home Maternity Services - postnatal in- home visiting	Complex Case Manager (Inc. Obstetric Medical Team)
Gynaecology	Specialist Clinics including Anaesthetics and Endocrine
Social Work including Child Protection Liaison Officer	Neonatal Unit from 32 weeks
Allied Health	Lactation Service
Perinatal Mental Health	Paediatrics

Metro North GP Alignment Program



Home / Healthcare Services / Maternity services

Maternity services



Pregnancy

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Preparing for labour

Labour and birth

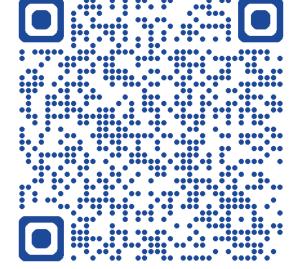
When complications occur

Newborn Bloodspot Screening



Complete the <u>online</u>
<u>registration form</u> to
book an appointment





Resources



Home / Healthcare Services / Maternity services / Choosing an option for maternity care

Choosing an option for maternity care

All options for maternity care are delivered by caring and dedicated health professionals in partnership with you and your support people. Your general practitioner (GP) or midwife will discuss these options at your <u>first appointment</u>. Choosing the best option for you will depend on your own personal preferences, and sometimes, your medical history.

Maternity care options

Caboolture Hospital offers a range of care options that vary to suit your individual needs.

- Midwives clinic
- Midwifery Group Practice Continuity of Care
- Private practice midwives
- The Lotus Circle (TLC)
- Aboriginal and Torres Strait Islander Maternity Service Ngarrama North
- + Kilcoy Outreach Clinic
- Obstetric led care with doctors and midwives
- GP shared care

Antenatal Clinic Location: Outpatient Services, 120 McKean Street Caboolture Hospital Phone: (07) 5433 8701 Ngarrama Maternal Health Ngarrama Maternal Health is also located at Caboolture, Kallangur and Bribie Island Satellite Health Centres. For all enquires please call Caboolture Hospital Outpatient Services on (07) 5433 8701

Caboolture Hospital | Choosing an option for maternity care

MEN!	Queensland Governme	ent		Contact us	Research	Newsroom	Get involved	☎ Resize font	A Print
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企	Healthcare services	Patients & visitors	Health professionals	Careers	About u	s			

Home / Healthcare Services / Maternity services / Learning about pregnancy, birth and baby

Learning about pregnancy, birth and baby

Learning about your pregnancy, birth and baby can help build your confidence and prepare you for the weeks ahead. Caboolture Hospital has resources and experienced staff available to help you throughout your journey.

Classes

We offer classes with our experienced staff, who will answer any of your questions about your pregnancy, birth and parenting. If you have any questions outside of these classes, please ask your health care provider.

- Becoming a family
- Evening class
- Saturday class
- Breast feeding classes

Class timetable

Bookings are essential for all classes

y 9.00am-3.30pm
ay evening 6:00pm – 8:30pm
ur midwife
ır Midwife
ons twice a month. Friday 9.00am–12.00pm and 1.00pm-
sic

Contact us

Antenatal Clinic

Location: Outpatient Services, 120 McKean Street, Caboolture Hospital Phone: (07) 5433 8474

Caboolture Hospital | Learning about pregnancy birth baby

Caboolture Complex Maternity Midwife Navigator

Caboolture catchment

Refer by

- Email: <u>CABHMidwifeNavigator@health.qld.gov.au</u>
- Phone:0436 937 527

Eligibility:

- Mental Health
- Domestic and Family Violence
- Child Safety
- Substance use
- History of poor engagement with care

Caboolture Young Mothers for Young Women





YPIC - Young. Pregnant. In Control.

Young Mothers for Young Women | Micah Projects

Caboolture Young Mothers for Young Women 19 Morayfield Road Caboolture South P: 07 5294 9600





GENETIC SCREENING

Pauline McGrath
Principal Genetic Counsellor | Children's Health Queensland





Screening and Diagnosis in Pregnancy Care

Pauline McGrath
Principal Genetic Counsellor
Queensland Children's Hospital

1st March 2025





Overview

- Reproductive Carrier Screening
- First trimester screening
 - cFTS
 - NIPT
- Second trimester screening
 - MSS
- Ultrasound scanning
- Genomic Medicine

- All women should be offered reproductive carrier screening
- Genetic Carrier Screening (C-Obs 63) (ranzcog.edu.au)

- Hundreds of inherited genetic conditions that can affect human health
- Most are very rare
- All these inherited conditions are considered together affect up to 1 in 400 people
- The majority of children born with such conditions are born into families with no other affected family members
- The two major types of inheritance that can lead to a healthy couple having children with serious genetic conditions are autosomal recessive and X-linked recessive

Mackenzie's Mission

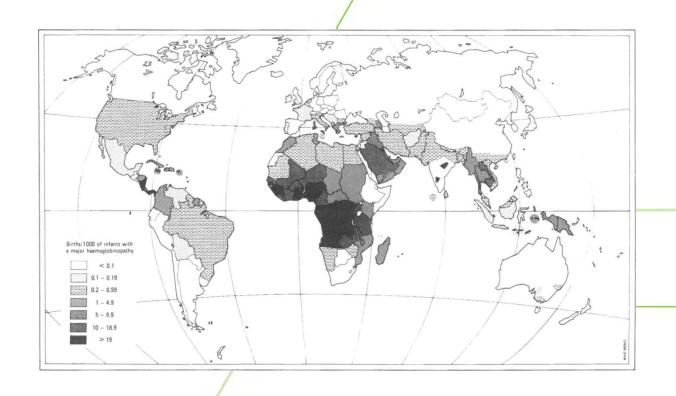
- Tested reproductive couples for pathogenic variants in at least 1281 genes associated with approximately 750 serious, childhood-onset autosomal recessive or X-linked conditions
- 1.9% of reproductive couples had a newly identified increased chance of having a child with one of these conditions
- Most of these couples have since chosen a reproductive option with the aim of avoiding having a child with the condition
- There was a high level of engagement with the study, positive attitudes toward screening, and minimal decisional regret
 - Nationwide, Couple-Based Genetic Carrier Screening | New England Journal of Medicine (nejm.org)

Reproductive Carrier Screening - Options

- 1. Having a child naturally and testing after birth to see if the child is affected
- 2. Conceiving naturally and having diagnostic testing during pregnancy to determine if the fetus is affected. This is usually performed with an invasive test (amniocentesis or chorionic villus sampling)
- 3. Conceiving the pregnancy by in vitro fertilisation (IVF) and testing embryos by preimplantation genetic diagnosis (PGD). Unaffected embryos would then be selected for achieving pregnancy. Using donor sperm, egg or embryo from unaffected individuals
- 4. Adoption
- 5. Not having children

- All couples intending to have children, or who are pregnant, should have a family history taken with a view to identifying relatives with heritable genetic disorders, as well as the presence of consanguinity or Ashkenazi Jewish heritage
- Those identified with a family history of a specific inherited disorder should be offered referral to a
 genetic counselling service for information about carrier testing and prenatal diagnosis/
 preimplantation genetic diagnosis for the condition
- All pregnant women should be offered basic screening for thalassaemia carrier status by a full blood examination initially
- Screening with specific assays for haemoglobinopathies (such as HPLC or EPG and haemoglobinopathy DNA testing) should be considered in high probability ethnic or population groups

Prevention and control of haemoglobinopathies*
M. Angastiniotis, B. Modell, P. Englezos, & V.
Boulyjenkov



Bulletin of the World Health Organization, 1995, 73 (3): 375-386

Genetic Carrier Screening (C-Obs 63) (ranzcog.edu.au)

Condition	Carrier	Affected	Main clinical features of the condition Recurrent lung infections, malabsorption, shortened life span	
Cystic fibrosis	1 in 35	1 in 4925*		
Spinal muscular atrophy Fragile X syndrome 1 in 332		1 in 9917*	Severe muscle weakness, death usually during childhood	
		1 in 7143 males^	Intellectual disability, autism	

^{* =} inferred from the carrier frequency

Item 73451 (*Updated 1 November 2024*)

- Testing of a patient (who is pregnant or planning pregnancy) to identify carrier status for pathogenic or likely pathogenic variants in a gene mentioned in paragraph (a), (b) or (c), to determine:
 - (a) for the cystic fibrosis transmembrane conductance regulator (CFTR) gene—reproductive risk of cystic fibrosis;
 - (b) for the survival motor neuron 1 (SMN1) gene—reproductive risk of spinal muscular atrophy;
 - (c) or the fragile X mental retardation 1 (FMR1) gene—reproductive risk of fragile X syndrome;

(other than a service associated with a service to which item 73300, 73305, 73345, 73346, 73348, 73348, 73349 or 73350 applies)

One test per lifetime

The intent of MBS item 73451 is to test an asymptomatic female chromosomal sex patient who is either planning a pregnancy or is already pregnant

Item 73452

Testing of the reproductive partner of a patient who has been found to be a carrier of a
pathogenic or likely pathogenic variant in the CFTR or SMN1 gene identified by testing under item
73451, for the purpose of determining the couple's reproductive risk of cystic fibrosis or spinal
muscular atrophy

One test per condition per lifetime

The intent of MBS item 73452 is to test an asymptomatic male chromosomal sex patient who is the reproductive partner of the patient planning pregnancy or already pregnant and has been tested under item 73451

• MBS item numbers were also approved for individuals who identify as being of Ashkenazi Jewish descent to access screening for up to nine autosomal recessive conditions more commonly present in this population (MBS items 73453, 73454 and 73455)

- Many commercially available options
- Expanded genetic carrier testing all offer different gene panels for different costs
- Women with a family history of CF, SMA or FXS should be referred directly to a clinician with genetics expertise rather than offered a "3-gene" screening panel, as they may require specialised testing
- If one reproductive partner is found to be a carrier it may also be wise to refer to a genetic service for testing of the other reproductive partner ie cystic fibrosis

- All couples found to have a higher chance of having a child with one of the conditions screened for should be referred for genetic counselling to be informed of available reproductive options
- The preconception period is the preferred timing for carrier screening
- If a couple are found to have an increased chance during pregnancy, genetic counselling and prenatal diagnosis should be offered
- Diagnostic testing (usually involving amniocentesis or chorionic villus sampling) may allow couples
 to prepare for the birth of a child with a genetic condition, to consider the option of terminating
 an affected pregnancy
- Regardless of the timing of diagnosis, it may be appropriate to refer the couple to see a physician
 with expertise in the condition ie referral of a couple found have a high chance of having a child
 with SMA to a paediatric neuromuscular physician

Aneuploidy Screening

All women should be offered aneuploidy screening

• <u>Screening and diagnosis of fetal structural anomalies and chromosome conditions (C-Obs 35)</u> (ranzcog.edu.au)

Aneuploidy Screening

More accurate than agerelated risk alone Screening in first trimester enables diagnostic testing

Reduction of invasive tests

Highest detection rate

- NIPT 99% detection rate for trisomy 21
- Combined first trimester screen 85-90% detection rate

Aneuploidy Screening

Source: https://www.ranzcog.edu.au

Test	Down Syndrome Detection Rate	Screen positive rate
Non-Invasive Prenatal Testing (NIPT)	99%	0.1%
Nuchal translucency scan (NTS)	70%	5%
Combined NTS, Serum testing (B HCG, PAPP-A)	85-90%	5%
Second trimester serum test (Free B HCG, oestriol, AFP +/- Inhibin)	65-70%	5%
Morphology scan	20-50%	10-15%

Combined First Trimester Screening

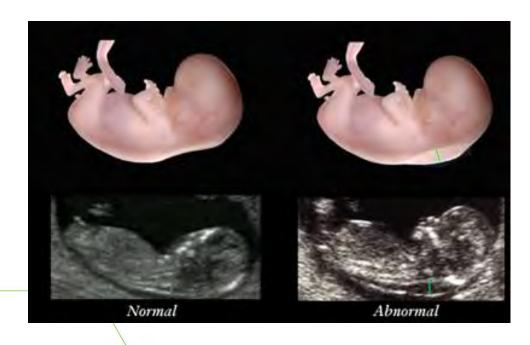


Image source: http://www.fetal.com



Image source: Woman's and Newborn Services RBWH

Combined First Trimester Screening – Nasal bone

• The presence of the nasal bone increased screening accuracy



Combined First Trimester Screening – Nasal bone

- Absent nasal bone
 - Delayed ossification of the nasal bone
 - Reassure women the baby will have a nose



Combined First Trimester Screening – Nasal bone

- At 11-13 weeks gestation, ~1-2% of normal fetuses have an absent nasal bone
- ~60% of fetuses with trisomy 21 have an absent nasal bone
- Overall effect on screening is increased detection and reduced screen positives

Combined First Trimester Screening

Indication:

1st Trimester screening.

History:

Maternal age: 33 years, pre-pregnancy weight 62.0 kg, height 170.0 cm, BMI 21.5, blood group: O, (Rh D): Rh +ve. Conception spontaneous. Non-smoker.

Obstetric History: Gravida: 5. Para: 2. CMV infection.

EDD by ultrasound: 7 January 2011. Gestational age: 13 weeks + 3 days

First Trimester Ultrasound

Transabdominal US with Voluson E8. Ultrasound view: good.

Fetal heart action present. Frequency 149 bpm

Crown-rump length (CRL) 75.0 mm 50th%

Nuchal translucency (NT) 1.92 mm

Nasal bone present

Fetal anatomy: skull/brain appears normal, heart not examined, spine appears normal, abdomen appears normal, stomach visible, bladder visible, hands both visible, feet both visible.

Additional Markers for Risk Assessment: Ductus Venosus (a-wave): positive. Placenta: posterior, structure normal. Amniotic fluid: normal. Cord: 3 vessels.

Cervix length 46 mm.

Summary of ultrasound findings: normal intrauterine pregnancy.

Size agrees with dates. I could not see any fetal abnormality on today's scan. Ultrasound is unable to detect all fetal abnormalities.

Maternal Serum Biochemistry:

Sample taken on 30 June 2010.

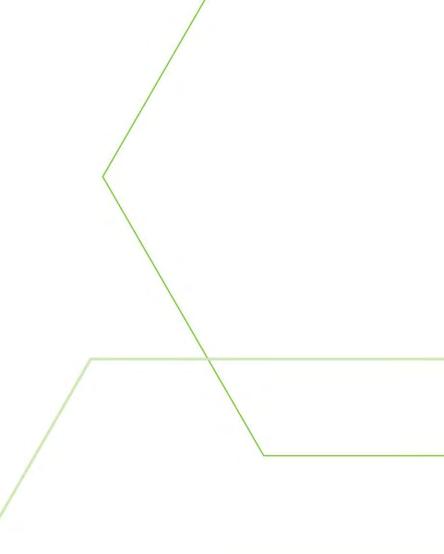
No. of fetuses: A. Maternal weight: 62.0 kg. Non-smoker. Ethnic origin: White. Parity > 0. Manufacturer:

Free beta hCG: 99.000 IU/I, equivalent to 2.7078 MoM. PAPP-A: 2.000 IU/I, equivalent to 0.5254 MoM.

Estimated risk for chromosomal abnormalities:

Trisomy 21 Trisomy 18 Trisomy 13

Background risk: 1:360 1:924 1:2886 Adjusted risk: 1:110 1:18484 1:57726



Combined First Trimester Screening – when to refer

Nuchal translucency	% Chromosomal defects	% Normal karyotype – fetal death usually prior to 20 weeks of gestation	% Normal karyotype – major fetal abnormalities	% Normal karyotype – alive and well
< 95th centile	0.2	1.3	1.6	97
3.5 – 4.4mm	21.1	2.7	10.0	70
4.5 – 5.4mm	33.3	3.4	18.5	50
5.5 – 6.4mm	50.5	10.1	24.2	30
> or equal to 6.5mm	64.5	19.0	46.2	15

Image source: Snijders et al 1998;2001;2005; Michailidis et al 2001

Combined First Trimester Screening –when to refer

Increased nuchal translucency (>3.5mm)

- cardiac malformations, genetic syndromes
- Recommend tertiary morphology scan 18-20 weeks gestation

Low PAPP-A (<0.4 MoM)

- associated with preeclampsia, growth restriction & stillbirth
- fetal growth & uterine artery doppler assessment at 22-24 weeks gestation

Non-invasive Prenatal Testing

 All women should be offered a first trimester anatomy scan even if they are choosing to have NIPT as a primary screening test

Non-invasive Prenatal Testing

Fetal cell-free DNA found in plasma of pregnant women from 10 weeks gestation

Testing of fetal DNA in maternal blood poses no risk to pregnancy

Not a diagnostic test, abnormal results should be confirmed via invasive testing

Cost approximately \$400

- Mother with chromosomally normal fetus the proportion of fragments will be in a narrow normal range
- If fetus has abnormal chromosomes the fetal contribution for that chromosome will be abnormal and distort the overall proportion



- Benefits of NIPT
 - Highest sensitivity and specificity
 - Reduces invasive testing
 - Beneficial for women unable to access cFTS or later gestation
 - Low false positive rate
 - Early as 10 weeks
 - Noninvasive
 - Large RATs may be detected by some platforms

- Limitations of NIPT
 - No Medicare rebate, costs vary
 - Abnormal results may require confirmation by invasive testing
 - Complex false positive and negative results
 - Failure rate of NIPT 0.1-3%
 - More likely to fail in
 - High BMI
 - Patient using anti-coagulant therapy
 - A quarter of CNV go undetected
 - Which one to choose?

When is NIPT not a good option

- Abnormalities on USS
 - NT > 3.5mm
 - Ventriculomegaly
 - Cardiac anomalies
- 8% of women who have fetal abnormality detected with have an abnormal chromosome micro array test
- Screening results > 1:100 (minimise delay)

• Chromosomal abnormalities detected by chromosomal microarray analysis and pregnancy outcomes of 4211 fetuses with high-risk prenatal indications | Scientific Reports

- Use a risk calculator
- NIPT Predictive Value Calculator



The estimated prevalence of Trisomy 21 (Down syndrome) at 16 weeks gestation for women who are 25 at EDD is 1 in 1040. Where does this number come from? See the FAQs from the menu above for details.

Sensitivity: Specificity: 99.2 99.91

The default performance metrics for Trisomy 21 (Down syndrome) are set at a sensitivity of 99.2 and specificity of 99.91 based on the weighted and pooled data from a meta-analysis by Gil et al (2015). The user may wish to change these inputs to reflect the performance statistics provided by the referral laboratory.

The prevalence of Trisomy 21 (Down syndrome) at 16 weeks gestation for a woman who is 25 at EDD is 1 in 1040.

The probability that result is a **true positive** (the fetus **is affected**). **PPV**:

51%

Probability that it is a **false positive** (the fetus is **not affected**).

49%

PPV (not rounded): 51.47631155622404%

PPV = (sensitivity x prevalence) / ((sensitivity x prevalence) + (1 – specificity)(1 – prevalence))

 $PPV = (0.992 \times 0.0009615384615384616) / ((0.992 \times 0.0009615384615384616) + (1 - 0.9991)(1 - 0.0009615384615384616))$

Please note: the post-test probability for an individual patient may differ based on other factors that influence her unique prior risk to have an affected pregnancy, such as gestational age of the patient, ultrasound findings and biochemical screening.

- Refer repeated failed NIPT for specialist care
- False positive NIPT
 - Placental
 - Confined placental mosaicism
 - Fetal
 - Vanishing twin early demise of aneuploid twin
 - Maternal
 - Sex chromosome aneuploidy (SCA) mosaic or non mosaic
 - Other aneuploid or structural mosaicism
 - Benign or malignant tumour
 - Bone marrow or organ transplant

Maternal Serum Screening

- Rarely used
- Blood test at 15-20 weeks gestation
- fβhCG + Oestriol + alpha fetoprotein (AFP)
- Detection rate 70%
- Provided risk assessment for open neural tube defects (AFP)
- Used 1 in 250 cut-off for high risk for chromosomal abnormalities
- Provides an option for screening later in gestation

Appropriate Diagnostic Test

High Risk Result	CVS	Amnio
T21		
T18		
T13		\square
XO		
XXX		
XXY		
XYY		

^{*} CVS would be appropriate for inc risk T13 and XO in the context of an abnormal ultrasound

CVS and Amniocentesis

Risks associated with CVS

- The risk of pregnancy loss due to a transabdominal CVS is between 0.5 and 1%
- The risk of pregnancy loss due to a transcervical CVS is up to 2%
- In 1%of cases, a CVS result may be difficult to interpret due to mosaicism. This uncertainty may be resolved by performing an amniocentesis
- In rare cases a result can not be provided from a CVS and repeat sampling may be required

Risks associated with amniocentesis

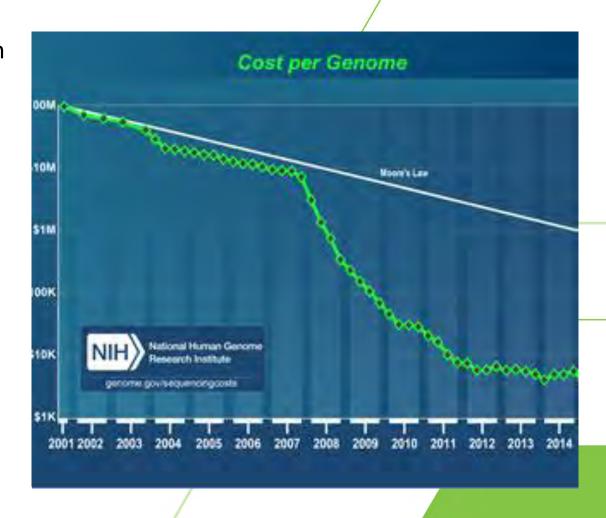
- The risk of pregnancy loss due amniocentesis is one in 900 procedures.
- Infection following amniocentesis is very rare and occurs in less than one in 1000 procedures performed
- In rare cases a result can not be provided from amniocentesis and repeat sampling may be required
- Risk of miscarriage following amniocentesis or chorionic villus sampling: systematic review of literature and updated meta-analysis PubMed

Ultrasound Scan – First Trimester Scan 11-14 weeks

- Advantages
 - Early Detection: Can identify major structural abnormalities earlier
 - Nuchal Translucency Measurement: Helps assess the risk of chromosomal abnormalities like Down syndrome
 - Accurate Dating: Provides accurate gestational age, to assist managing the pregnancy
 - Can detect multiple pregnancies (twins, triplets, etc.) early
 - Placental position
- Disadvantages:
 - Limited Detail: Some abnormalities may not be visible or fully developed at this stage
 - False Positives/Negatives: Early scans can sometimes lead to false positives or negatives, requiring follow-up scans
 - Anxiety: Discovering abnormalities at this stage can cause significant anxiety for expectant parents

Genomics in Pregnancy

- More than 10 years ago the 'reference' human genome sequence was published
- Approximately 20,000 human genes
- The smaller than expected number hinted at the hidden complexity of the human genome



Genomics in Pregnancy – Prenatal Testing

- Banded karyotype to determine translocations, used if testing for familial translocation or in the presence of an abnormal FISH result for trisomy 13 and 21
- Chromosome microarray
- Routine Genetic testing for known familial conditions
 - TAT 2-4 weeks ie CF, DMD, Fragile X
- Targeted Panel testing ie skeletal dysplasia panel
- Whole Exome Sequencing approximately 2% of the genome, analyses manageable data sets
- Whole Genome Sequencing includes coding, non-coding and mitochondrial DNA can detect novel genomic variants (structural, single nucleotide, insertions, deletions and CNV)

Genomics in Pregnancy – Prenatal Testing

Fetal phenotype group	Definition	Total cases	Solved cases (diagnostic yield)
Skeletal malformations	Evidence of skeletal abnormalities in ultrasound, such as shortened tubular bones, multiple fractures, achondroplasia, thanatophoric dysplasia, other skeletal dysplasias	63	33 (52%)
Complex malformations	≥2 organ systems affected in ultrasound, incl. Facial dysmorphias	122	54 (44%)
Urogenital malformations	Renal agenesis, renal dysplasia, polycystic kidneys	25	11 (44%)
Brain malformations	Lissencephaly, corpus callosum agenesis, holoprosencephaly, hydrocephalus, ventrikulomegalia	79	34 (43%)
Increased nuchal transparency	Nuchal transparency >3 mm, nuchal edema, hygroma colli	72	24 (33%)
IUGR (intrauterine growth retardation)	<10th percentile	27	7 (26%)
Heart defects	Ventricular septal defect, hypoplastic left heart syndrome, tetralogy of Fallot	50	12 (24%)
Eye anomalies	Anophthalmie, cataracts	10	2 (20%)
Arthrogryposis	Arthrogryposis	10	2 (20%)
Abnormalities of internal organs	Intestinal malformations (e.g., microcolon), megacystis, malformations of the liver	21	4 (19%)
Other	For example, abnormal biochemical parameters such as PAPP-A, β-hCG; akinesia, generalized edema, harlequin ichthyosis	21	6 (29%)

Trio exome sequencing is
highly relevant in prenatal
diagnostics - Gabriel - 2022 Prenatal Diagnosis - Wiley
Online Library

Thank you

- <u>Statements and guidelines directory RANZCOG</u>
- Guideline: Preconception and prenatal genetic screening



EPILEPSY IN PREGNANCY

Dr Lata Vadlamudi Staff Specialist | Neurology, RBWH



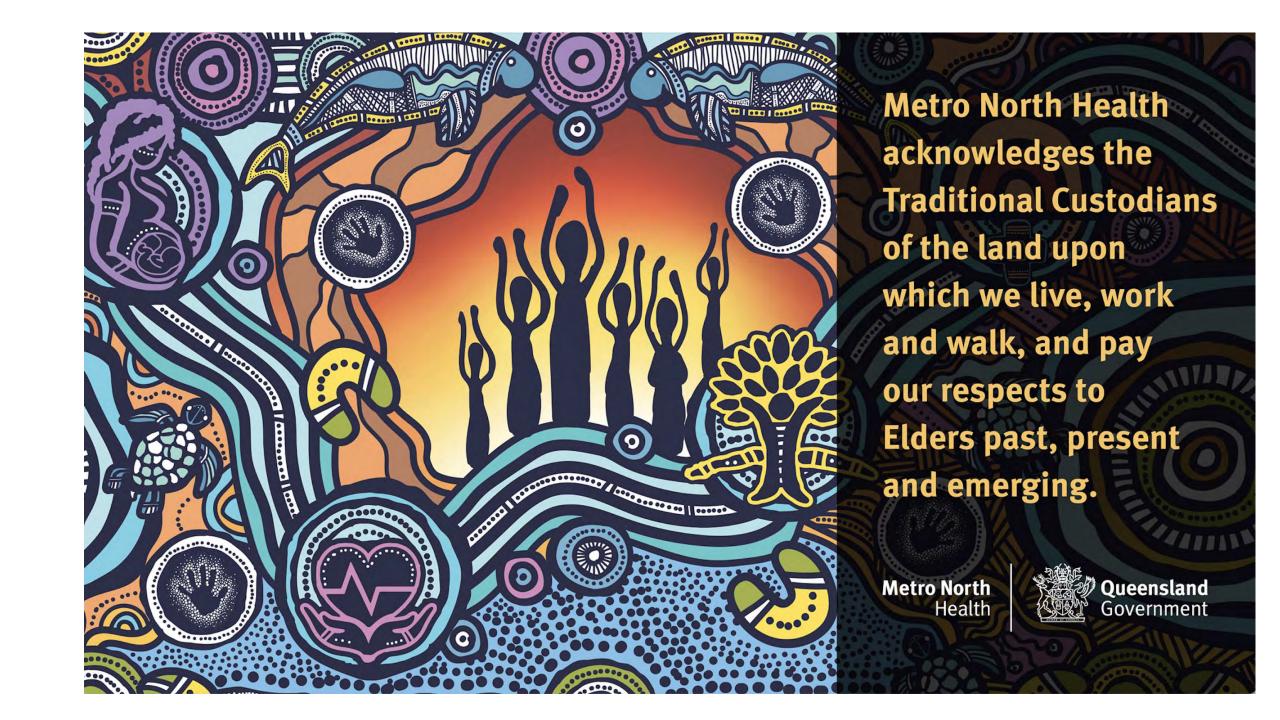


Epilepsy and Pregnancy 2025

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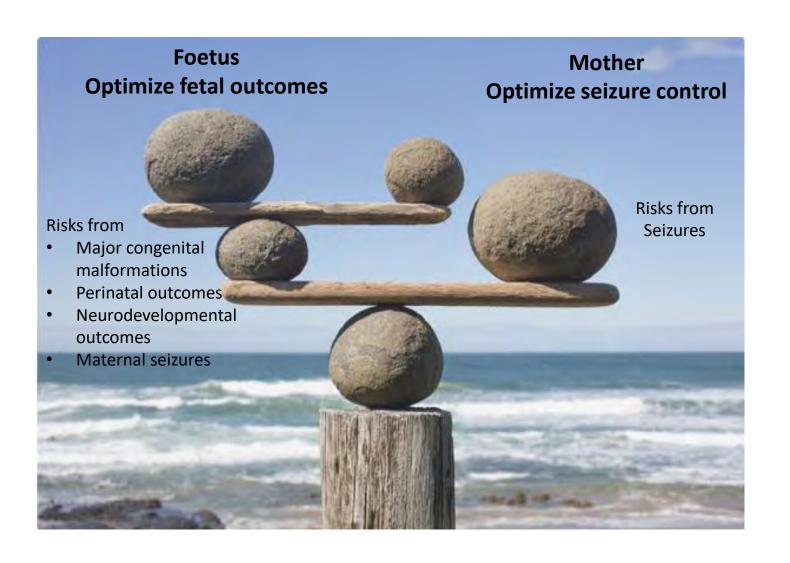
Metro North GP Maternity Workshop Saturday 1 March 2025





Epilepsy and Pregnancy

Optimizing fetal outcomes and maternal seizure control



- Vast majority of women with epilepsy have uneventful pregnancies and give birth to healthy children.
 - Planning will improve outcomes for mother and baby
 - Shared decision-making with your GP, neurologist and up-to-date knowledge is the key

Maternal and Foetal Risks of Seizures

Maternal

- Injury
- Status epilepticus
- Sudden unexplained death with epilepsy
 - More than 10 times higher
- Adverse outcomes- preterm labour, preeclampsia, postpartum haemorrhage
- Prolonged hospital admissions

Foetal

- Trauma from uterine injury
- Adverse outcomes low-birth weight baby and premature baby
- Prolonged bilateral tonic-clonic seizures (BTCS) associated with foetal hypoxia
- Stillbirth
- Frequency of BTCS in the mother may be a risk factor for low IQ in their children

Original Investigation

Mortality and Morbidity During Delivery Hospitalization Among Pregnant Women With Epilepsy in the United States

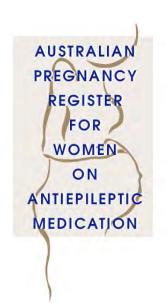
JAMA Neurol. doi:10.1001/jamaneurol.2015.1017 Published online July 6, 2015.

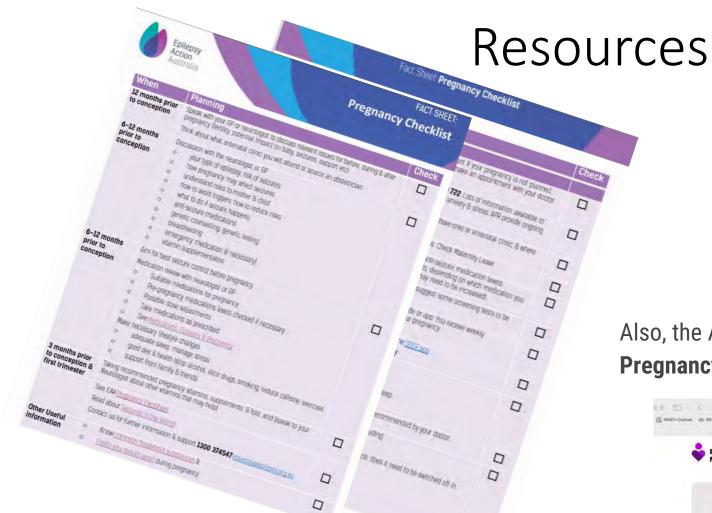
Sarah C. MacDonald, BSc; Brian T. Bateman, MD, MSc; Thomas F. McElrath, MD, PhD; Sonia Hernández-Díaz, MD, DrPH



Pre-Pregnancy planning tips

- Regular neurologist review (there is a Women with Epilepsy clinic at RBWH)
 - Review medication choices
 - Ensuring the most optimal medication for the epilepsy type and for pregnancy
 - Review the dose- aim for lowest, safest dose needed to control seizures
- Shared decision regarding use of anti-seizure medications if falls pregnant
- Discuss general health, diet and well being
- Review their support network for after the baby is born
- Screen for anxiety and depression
- Start folic acid supplements- at least 0.4 mg/day
- Obtain pre-pregnancy anti-seizure medication levels
- Talk about the Australian Pregnancy Register and other resources
- Assisted reproductive technology- watch seizure control if on Lamotrigine
- Ob Med pre-conception clinic at RBWH, with pharmacist support







For pregnant women with epilepsy and/or pregnant women taking antiepileptic meds for other conditions

1800 069 722 | www.apr.org.au | apr@mh.org.au

Also, the APR are currently recruiting for the **Lacosamide and Pregnancy** study. Call the APR for more information.





1300 37 45 37

epilepsy@epilepsy.org.au www.epilepsy.org.au

Fertility in women with epilepsy

Epilepsy

- Higher rate of menstrual disorders
 - around 30% versus 12-14 % in the general population
- Higher rate of polycystic ovary syndrome
 - 10-25% compared with 4-7% in the general population
- Higher rate of sexual dysfunction
- Higher risk of premature ovarian failure (< 42 years)
 - 14% compared with 1% population risk
- Poorly controlled seizures may lead to earlier age of menopause

Anti-seizure medications (ASM)

- Enzyme-inducing ASM can contribute to sexual dysfunction, menstrual irregularities, fertility issues
- Valproate is associated with polycystic ovarian syndrome, weight gain, anovulatory cycles and menstrual irregularities
- Levetiracetam not found to have endocrine or sexual dysfunction effects
- Lamotrigine possible improvement in sexual function in women
- No data on newer anti-seizure medications

Genetics and pre-pregnancy care

- There is no single test that will diagnose all epilepsies with a genetic basis
 - Copy number variation or single gene mutation can cause some epilepsies
- Referral to a clinical geneticist for discussion
 - If there is a known copy number variation or single gene causing the epilepsy
 - If there is a history of major congenital malformations with a previous pregnancy
 - If there is a strong family history of epilepsy, unknown cause for epilepsy, drug resistant epilepsy and neurodevelopmental comorbidities
- Routine testing for all epilepsy patients is not undertaken
- We know certain epilepsy types run in families, but have a low yield for a single gene on testing, likely multiple genes of small effect
- Neurogenetics clinic- referral to Genetic Health Queensland

Risk of epilepsy to offspring

- If you have generalised epilepsy the chance your child will develop epilepsy is around 1 in 12
- If you have focal epilepsy the chance your child will develop epilepsy is 1 in 50

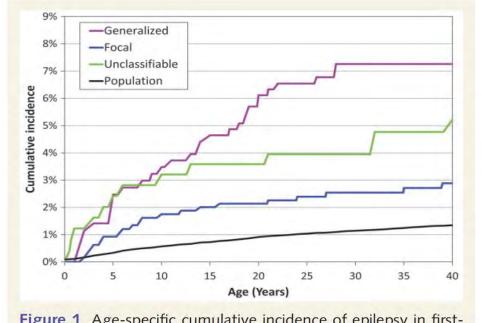


Figure 1 Age-specific cumulative incidence of epilepsy in first-degree relatives of probands with epilepsy, by proband epilepsy type.

Maternal effect- higher risk of epilepsy in children born to mothers with epilepsy compared with fathers

(Peljto et al Brain 2014;137:795-805)

Contraception and interactions with anti-seizure medications

Anti-seizure medications can reduce effectiveness of the contraception

Contraception can reduce anti-seizure medication levels

Enzyme-inducing anti-seizure medications increase activity of the CYP3A4 system and increase sex hormone-binding globulin.

Increase the metabolism of estrogen and progesterone, decreasing their levels

Lamotrigine, in particular (also valproate and oxcarbazepine).
Estrogen induces glucuronidation (enzyme

UGT1A4) and increases lamotrigine elimination

STRONG INDUCERS	WEAK INDUCERS	NON-INDUCERS
Carbamazepine	Topiramate (higher doses)	Levetiracetam
Oxcarbazepine	Perampanel (higher doses)	Zonisamide
Phenytoin	Lamotrigine*	Briviracetam
Phenobarbital	Felbamate	Lacosamide
Primidone	Rufinamde	Ethosuximide
Clobazam		Valproate (inhibitor)
Cenobamate		Clonazepam

* Lamotrigine
No change to
estrogen levels,
modest decrease
in progestins

Mirena and Kyleena

- Contraception of choice for women with epilepsy
- levonorgestrel- releasing intrauterine device (IUD)
- Mirena (52 mg), releases 21 mcg/day
- Kyleena (19.5 mg), releases 9 mcg/day suited for younger women without children
- Local hormone release-thickens cervical mucous and thins endometrium Disadvantages **Advantages**
 - User independent
 - Effectiveness Mirena 99.8%, Kyleena 99.75%
 - Low risk of complications, lighter periods
 - Mirena duration extended from 5 to 8 years Kyleena up to 5 years
 - Rapid return to fertility after removal
 - No oestrogen
 - No expected change in contraception effectiveness with any anti-seizure medications
 - No effect on anti-seizure medication levels

- Some hormonal side effects possible-bloating, mood swings
- Temporary pain with insertion
- Expulsion or removal pain, increased bleeding
- Risk of pelvic infection



Medroxyprogesterone acetate intramuscular injection (Depo Provera)



Inhibits ovulation

Advantages

- User-independent
- Effectiveness 99.8%
- Can cause loss of periods
- No estrogen
- No change in contraception effectiveness with any anti-seizure medications
- No effect on anti-seizure medication levels

Disadvantages

- reinjection every 3 months
- can cause irregular bleeding, mood changes
- Once given can not be removed
- Not immediately reversible, return to fertility can be delayed up to 18 months
- Can reduce bone density
- Prolonged use (> 3 years) and association with increased risk of brain meningioma



Use of progestogens and the risk of intracranial meningioma: national case-control study

Noémie Roland, ¹ Anke Neumann, ¹ Léa Hoisnard, ² Lise Duranteau, ³ Sébastien Froelich, ⁴ Mahmoud Zureik, ^{1,5} Alain Weill ¹

Implanon

Slow release Etonogestrel 3 cm soft, flexible rod, sits under the skin in the upper arm

Advantages

- Effective 99.95%
- User independent
- Lasts up to 3 years
- No estrogen
- No effect on anti-seizure medications

Disadvantages

- Reversible with removal
- Pain and complications at the insertion site
- Bleeding irregularities
- Enzyme inducing anti-seizure medications can reduce contraception effectiveness



Oral contraceptive (OC) pill

- Combined pill contains estrogen and progesterone
- Apart from dropirenone, progesterone only pills do not reliably inhibit ovulation

Advantages

- Non-invasive
- May shorten periods
- No impact on fertility after cessation
- Effectiveness 99.7% if taken correctly

Disadvantages

- User dependent
- If missed pills, effectiveness can drop to 91%
- Contains estrogen- can increase risk of thrombosis
- Enzyme-inducing anti-seizure medications reduce the contraception effectiveness
- Combined OC pill reduces lamotrigine levels, can also reduce valproate and oxcarbazepine levels
- Dropirenone may decrease lamotrigine levels

Contraception Barriers- Condoms

Advantages

- Condoms protect against sexually transmitted infections
- Useful in conjunction with other contraception (dual protection)
- No change in contraception effectiveness with any antiseizure medications
- No effect on anti-seizure medication levels

Disadvantages

- User dependent
- Effectiveness around 85%

Predictors of seizure control

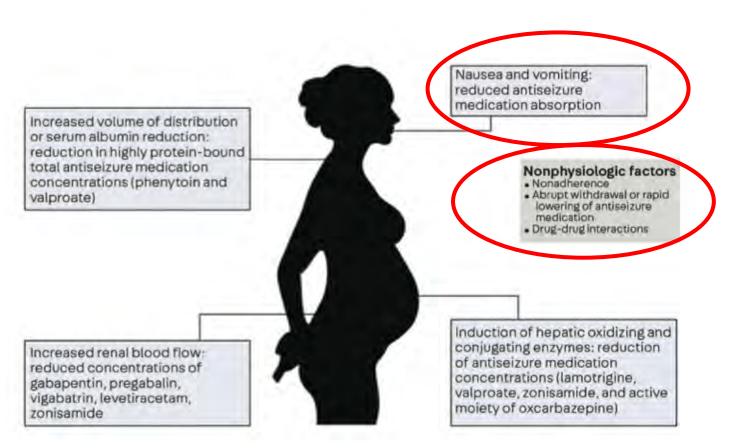
- Seizure occurrence prior to the pregnancy is the most important predictor of seizures during pregnancy
 - If seizure in the month prior to pregnancy- 15x greater risk of seizure during pregnancy
 - If seizure-free the year before conception, more than 80% remain seizure-free
- Other predictors of worsening seizures
 - Focal epilepsy syndrome in particular frontal lobe epilepsy (Voinescu et al Neurology 2022)
 - Sleep deprivation
- Changes to antiseizure medication levels

Changes in antiseizure medication (ASM) levels

ASM dependent and substantial inter-individual variability

Decline in serum concentration > 35% from pre-pregnancy optimal concentration is

associated with increased risk of deterioration in seizure control



Defined therapeutic window for effective seizure control

Decreases can occur at any point during the pregnancy

Many ASM – in particular, lamotrigine and levetiracetam

Pregnancy

SPECIAL ARTICLE

Teratogenesis, Perinatal, and Neurodevelopmental Outcomes After In Utero Exposure to Antiseizure Medication

Practice Guideline From the AAN, AES, and SMFM

Alison M. Pack, MD, MPH, Maryam Oskoui, MD, MSc, Shawniqua Williams Roberson, MEng, MD, Diane K. Donley, MD, Jacqueline French, MD, Elizabeth E. Gerard, MD, David Gloss, MD, MPH&TM, Wendy R. Miller, PhD, RN, CCRN, Heidi M. Munger Clary, MD, MPH, Sarah S. Osmundson, MD, MS, Brandy McFadden, Kaitlyn Parratt, MBBS (Hons 1), Page B. Pennell, MD, George Saade, MD, Don B. Smith, MD, Kelly Sullivan, PhD, Sanjeev V. Thomas, MD, DM, Torbjörn Tomson, MD, Mary Dolan O'Brien, MLIS, PMP, Kylie Botchway-Doe, Heather M. Silsbee, MWC, and Mark R. Keezer, MDCM, PhD

Neurology® 2024; 102: e209279. doi:10.1212/WNL.0000000000209279

- Correspondence American Academy of Neurology guidelines@aan.com
- Systematic review by a multidisciplinary panel up to August 2022
- Total articles included = 82
- Summary of the key findings

- a) Prevalence of major congenital malformations across specific antiseizure medications (ASM)
- b) Perinatal outcomes
- c) Neurodevelopmental outcomes after in utero exposure of ASM
- d) Impact of folic Acid

Prevalence of major congenital malformations (MCM)

- Without epilepsy prevalence of MCM in the general population 2.4-2.9%
- No independent effect of polytherapy
- Data from >1000 exposures lowest prevalence of MCM
 - Lamotrigine 3.1%,
 - Levetiracetam 3.5%,
 - Oxcarbazepine 3.1%
- Highest prevalence- Valproate 9.7%
- Types of malformations
 - Valproate -neural tube defects (1.4%), urogenital (1.2%), renal (1.4%)
 - Phenobarbitone- cardiac malformations (4.4%), oral and cleft palate (2.2%)
 - Topiramate- oral and cleft palate (1.4%)

Neurodevelopmental and Functional Outcomes Following In Utero Exposure to Antiseizure Medication

A Systematic Review

Eliza Honybun, MPsych, Emily Cockle, MPsych, Charles B. Malpas, MPsych, PhD,
Terence J. O'Brien, MB, BS, MD, FRACP, FRCPE, FAHMS, FAES, Frank J. Vajda, MB BS, MD, FRCP, FRACP,
Piero Perucca, MD, PhD, FRACP,* and Genevieve Rayner, BA (Hons), MPsych, PhD*

Neurology® 2024;102:e209175. doi:10.1212/WNL.0000000000209175

Correspondence Ms. Honybun eliza,honybun@ unimelb.edu.au Systematic review of 43 studies from 1990 to 2023

- Focus on the social, emotional, behavioural and adaptive domains
- Valproate- 2-4 x increased risk autism spectrum disorder (ASD), 2-5 x increased risk intellectual disability (ID) and poor adaptive functioning
- Topiramate- growing evidence 2x increased risk ASD, 3-4 x increased risk ID
- These outcomes are dose dependent
- Levetiracetam linked to increased attention deficit hyperactivity disorder and anxiety
- Carbamazepine variable neurodevelopmental outcomes
- Lamotrigine seems to be "safe" in terms of post natal development
- Evidence for other anti-seizure medications is lacking

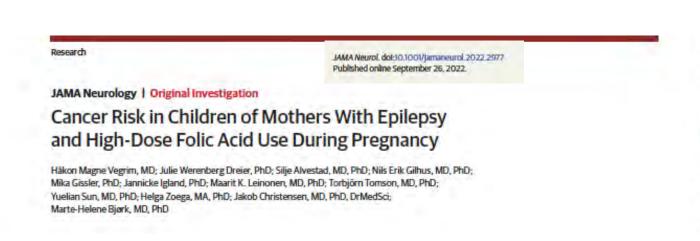
Folic acid benefits

- No benefit specific benefit for prevention of major congenital malformations for people with epilepsy, general childbearing evidence
- Possibly associated with better neurodevelopmental outcomes
- Reduced autistic traits and language delay at 3 years (Norwegian Mother and Child Cohort Study)
- Likely associated with with higher global IQ at 6 years in children exposed to ASM in utero (NEAD study Group)

- Bjork M, Riedel B, Spigset O, et al. JAMA Neurol. 2018;75(2):160-168.
- Husebye ESN, Gilhus NE, Riedel B, Spigset O, Daltveit AK, Bjork MH. Neurology. 2018;91(9):e811-e821. doi:10.1212/WNL.0000000000006073
- Meador KJ, Baker GA, Browning N, et al; NEAD Study Group. Lancet Neurol. 2013;12(3):244-252.doi:10.1016/S1474-4422(12)70323-X

What is the safe dose of folic acid?

- Association between high dose folic acid (defined at greater than 1 mg/day) and cancer risk in children of mothers with epilepsy
- Further population study- showed association between high dose folic acid (greater then 1 mg/day) in all women who have given birth
- At least 0.4 mg/day
- Adherence is generally poor
- Consider if they are also on another supplement (eg elevit- 800 mcg folic acid)





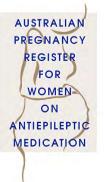
Unplanned pregnancy

- Seek neurological advice, urgent referral to the Comprehensive Epilepsy Program
- Scenario's- taking multiple ASM, on Valproate, poor seizure control, minimal work-up prior to pregnancy
- Caution in attempting to remove or replace ASM if it is effective, even if it is not optimal choice (Valproate)
- Ensure safety for mother and baby, seizure control is paramount
- Stress to the patient the importance of not ceasing ASM
- Commence folic acid supplements (at lease 0.4 mg/day)
- Generally, if medication change is warranted, we try to avoid in the first trimester (organogenesis), but best to obtain neurological advice

Pregnancy Management

ASM- anti-seizure medication

- Will be a high-risk pregnancy for obstetric care
- Caution in changing an ASM if effective seizure control, even if not optimal
- Folic acid supplements, review other supplements (elevit- 800 mcg folate)
- If hyperemesis gravidarum- re-dose ASM if vomiting occurs within 1 hour
- Discuss the Australian Pregnancy Register
- Screen for anxiety and depression
- Trough ASM levels
 - Ideal is monthly
 - In practice, at least every trimester, generally 3-4 levels (12 weeks, 20 weeks, 28 weeks, 36 weeks) and more frequently as required
- Aim to ensure decline in level is <35% of pre-pregnancy level
- Review for anxiety and depression
- Check on support networks for after the birth



Post-Partum Management

Develop plan for contraception

Maternal safety

- Risk of seizures increased and may persist for several months due to sleep deprivation
- Discourage from taking a bath alone/behind locked door
- Assess post-partum depression and anxiety
- Sleep hygiene
 - At least 6 hours of sleep, at least one 4 hours stretch/24 hours cycle
 - Early introduction of the bottle given by family member if breast feeding
 - eg shift approaches to ensure the mother has uninterrupted sleep

Newborn safety

- No driving, subject to seizures and sleep deprivation
- No bathing of the baby with the mother alone
- No co-sleeping with the mother
- Baby carrier if mother at risk of myoclonic jerks

Breast feeding

ASM- anti-seizure medication

- Generally encouraged due to established benefits
- Children of mothers treated with Valproate, IQ at 6 years higher compared if not breastfed
- Different ASM concentrations in breastmilk
 - low with Levetiracetam and medium with Lamotrigine
- If ASM has been increased during pregnancy, reduction but generally a bit higher than prepregnancy dose (balance seizure risk and sleep deprivation)
- Take ASM immediately after breastfeeding or before baby's longest sleep
- Adverse effects to the newborn appear to be rare (sedation, poor suckling and weight gain)
 - serum levels if symptoms, consider reducing amounts, combining with formula
- Importance of avoiding stress and sleep deprivation related to breast feeding

Summary

Pre-pregnancy

- Planning
- Shared decision-making
- IUD contraception of choice
- Folic acid- at least 0.4 mg/day folic pre-conceptionally and during pregnancy
- Baseline ASM drug levels
- screen for anxiety and depression

IUD- intrauterine device
ASM- anti-seizure medication
LTG-lamotrigine
LEV-levetiracetam
OXC-oxcarbazepine
TPM-topiramate
VPA- valproate

Pregnancy

- If unplanned-caution in changing if effective seizure control, Neurology review
- Discuss Australian Pregnancy Register
- VPA and TPM- dose dependent risks
- Consider using LTG, LEV, OXC
- LEV may affect post-natal neurodevelopment, at least at higher doses
- Breast feeding is encouraged
- Post-partum care of mother and newborn

Acknowledgements

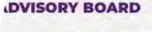
Women with epilepsy

A/Prof Cecilie Lander





For pregnant women with epilepsy and/or pregnant women taking antiepileptic meds for other conditions





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