MATERNITY CONSUMER NETWORK | Down Syndrome Queensland | Prenatal Screening | Queensland Government

FACTSHEET

Prenatal screening

What is prenatal screening?

Prenatal screening is offered in pregnancy to look for certain chromosomal variations.

These screens and tests look for **trisomy** and **sex chromosome conditions**.

Trisomy is a broad term used to describe conditions with extra chromosomes.

A common trisomy is Down syndrome.

Sex chromosome conditions happen when full or partial deletions or duplications of sex chromosomes occur.

Turner syndrome and Klinefelter syndrome are two types of sex chromosome conditions.

What are my options?

Prenatal screening should always be offered by your provider.

It is your decision whether or not to have prenatal screening tests and/or diagnostic tests.

You are in charge of what happens to your body.

The final decision is yours.

The ***Screening Choices Tool*** (right) may help you to understand choices and make decisions.

If opting out of prenatal screening, your choice should be respected, and your provider should still offer you standard antenatal care.

Screening Choices Tool

FIRST CHOICE–SCREENING TESTS

- Screening **no**
- Screening **yes**

High chance | **Low** chance

SECOND CHOICE–DIAGNOSTIC TESTS

- More information needed?
- Testing **no**
- Testing **yes**

Confirmed | **Not confirmed**

THIRD CHOICE–WHAT'S NEXT

- Continue pregnancy
- Adoption/other
- Terminate pregnancy

Prenatal screening

Prenatal tests can be either screening or diagnostic. Each test is offered at a specific stage of pregnancy.

Screening tests are used to establish whether there is a low or high chance of a chromosomal condition. Examples of screening tests are blood tests and ultrasounds.

Diagnostic tests are usually offered after a high-chance screening test result, to verify whether a chromosomal condition is present. An example of a diagnostic test is an amniocentesis.

You can opt out of testing or change your mind at any time.

Your provider will discuss with you the best time to have each test during your pregnancy, as well as any costs and how you can access these tests – especially if you are in a rural or remote location.

Common chromosome conditions

CHROMOSOME CONDITION	CHANCE*
Trisomy 21 Down syndrome	1 in 1100
Trisomy 18 Edward syndrome	1 in 1600
Trisomy 13 Platau syndrome	1 in 3400
SEX CHROMOSOME CONDITION	**CHANCE***
Turner syndrome – Monosomy X or 45, X	1 in 2500 females
Klinefelter syndrome – 47, XXY	1 in 450 males
Triple X syndrome – 47, XXX	1 in 1000 females
Jacob syndrome – 47, XYY	1 in 1000 males

*chance dependant on age

Prenatal screening results

If it is determined that your baby may have a chromosomal condition, your provider should arrange a private consulting area in which to explain the screening results to you.

You can choose to bring a support person with you to your appointment and your provider. will arrange an interpreter if needed.

Providers should:

- be supportive and respectful when giving results
- allow you time to absorb information, discuss your needs and concerns and ask questions
- talk with empathy and answer truthfully.

Chromosome conditions:

- occur for parents of all ages, cultures and backgrounds
- are not caused by anything the parents have done before or during pregnancy.

Your provider will offer you referral to further support, services and resources. You can request to be connected to people living with chromosomal conditions and their families, who can offer you a unique insight and perspective.

What's next?

Upon receiving results confirming chromosomal conditions you have many options.

You may choose to:

- continue the pregnancy (raise the baby)
- continue the pregnancy (arrange adoption/other)
- terminate the pregnancy.

Your provider must be supportive of your choice.

Further information and support can be found at **www.prenatalscreening.org.au**

INFORMATION AND RESOURCES

prenatalscreening.org.au
genetic.org/variations/about-xyy
understandingturnersyndrome.org
genetic.org/variations/about-47xxy
trisomy.org/about-trisomy/trisomy-13
trisomy.org/about-trisomy/trisomy-18
genetic.org/variations/about-trisomy-x
downsyndrome.org.au/about-down-syndrome/what-is-down-syndrome/
safetyandquality.gov.au/consumers/working-your-healthcare-provider/australian-charter-healthcare-rights
downsyndrome.org.au/qld/services-and-supports/pre-natal-diagnosis/

MATERNITY CONSUMER NETWORK

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