

Genetic Counselling and Testing

Advanced maternal age is associated with an increased risk of chromosomal abnormalities and certain genetic conditions. As individuals age, the likelihood of chromosomal nondisjunction during egg development increases, which raises the risk of aneuploidy in the embryo. These risks include conditions such as trisomy 21 (Down syndrome), trisomy 18, and trisomy 13.

Refer to [SOGC Guideline No. 456: Prenatal Screening for Fetal Chromosomal Anomalies](#) and [SOGC Guideline No. 346-Advanced Reproductive Age and Fertility](#).

Preconception care provides an opportunity to discuss these risks before pregnancy occurs and to offer appropriate counselling regarding genetic screening and reproductive options. Early conversations allow patients and couples to make informed decisions about pregnancy timing, fertility planning, and available testing options.

Why Genetic Counselling Matters Before Pregnancy

Age-related reproductive risks increase gradually beginning in the mid-30s and continue to rise with advancing age. Preconception genetic counselling allows clinicians to:

- Discuss age-related chromosomal risks
- Review personal and family genetic history
- Identify individuals who may benefit from genetic carrier screening
- Provide information about reproductive options and available testing

These discussions support informed decision-making and help individuals plan pregnancies with greater awareness of potential risks.

Key Preconception Considerations

Providers should consider discussing genetic counselling with patients who are:

- 35 years or older at anticipated time of delivery
- Individuals with a family history of genetic disorders
- Patients with a prior pregnancy affected by chromosomal or genetic conditions
- Couples planning assisted reproductive technologies

Assessment should include both maternal and paternal factors, as paternal age may also influence certain genetic risks.

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Available Genetic Testing and Screening Options

During preconception counselling, clinicians may review potential options that could be offered before or during pregnancy, including:

Carrier screening

- Identifies whether individuals carry genes associated with inherited conditions (e.g., cystic fibrosis, spinal muscular atrophy)
- Can be performed before pregnancy to support informed reproductive planning

Prenatal genetic screening

- Screening tests performed during pregnancy to estimate risk of chromosomal abnormalities
- May include non-invasive prenatal screening (NIPS) or other screening approaches

Diagnostic testing

- Invasive diagnostic tests such as chorionic villus sampling (CVS) or amniocentesis can confirm chromosomal abnormalities during pregnancy

Preconception counselling helps patients understand these options before pregnancy begins.

Referral to Genetic Counselling

Referral to a genetic counsellor may be appropriate when:

- There is a family history of genetic disorders
- Patients request expanded carrier screening
- There has been a prior pregnancy affected by genetic conditions
- Patients are planning assisted reproductive technologies
- Complex genetic risk assessment is required

Genetic counsellors can provide detailed risk assessment, discuss testing options, and support shared decision-making.

Clinical Pearl Preconception genetic counselling allows patients to understand age-related genetic risks and consider screening and testing options before pregnancy occurs, supporting informed reproductive planning.

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Quick Clinical Actions

During visits with patients of advanced reproductive age (≥ 35 years):

- Discuss age-related fertility and genetic risks
- Review family genetic history
- Offer carrier screening where appropriate
- Provide information about prenatal screening options
- Refer to genetic counselling when indicated

Early counselling helps patients make informed decisions about pregnancy timing, fertility options and prenatal testing.

Who Should Be Referred for Genetic Counselling?

Consider referral to a genetic counsellor for individuals or couples with any of the following:

- Age ≥ 35 years at anticipated time of delivery
- Family history of a genetic disorder or inherited condition
- Previous pregnancy affected by a chromosomal or genetic condition
- Known carrier status for a genetic condition (patient or partner)
- Interest in or request for expanded carrier screening
- Consanguinity (partners who are biologically related)
- Recurrent pregnancy loss or infertility with suspected genetic etiology
- Planning assisted reproductive technologies (ART)
- Abnormal genetic screening results (if already available)
- Uncertain or complex family history requiring detailed risk assessment

Clinical Tip: Early referral in the preconception period allows more time for counselling, testing and consideration of reproductive options.

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Resources for Health Care Providers

To support preconception genetic counselling and informed decision-making, the following resources provide clinical guidance, education, and referral support.

SOGC Resources

- [SOGC Guideline No. 456: Prenatal Screening for Fetal Chromosomal Anomalies](#)
- [SOGC Guideline No. 346-Advanced Reproductive Age and Fertility](#)
- [SOGC Committee Opinion No. 406: Prenatal Testing After IVF With Preimplantation Genetic Testing for Aneuploidy](#)

Canadian Genetics Resources

- [Canadian College of Medical Geneticists](#) (CCMG)
- [Genetics Education Canada - Knowledge Organization](#) (GECKO)
- [Prenatal Screening Ontario](#) (PSO)

Patient Resources

- [MedlinePlus Genetics](#)