

Clinician Quick Reference Table

Domain	Key Considerations	Why It Matters	When to Discuss / Screen	When to Refer
Maternal Age	Fertility declines with age, especially ≥ 35 years	Reduced ovarian reserve and increased pregnancy complications	Discuss reproductive goals with patients ≥ 30 -35 years	Fertility specialist if infertility risk or delayed conception
Chromosomal Risk	Risk of aneuploidy increases with maternal age (e.g., trisomy 21, 18, 13)	Age-related chromosomal nondisjunction increases embryo risk	Discuss genetic screening options with patients ≥ 35 years	Genetic counselling for patients with concerns or risk factors
Carrier Screening	Identifies carriers of inherited genetic conditions	Helps identify risk for recessive genetic diseases	Offer preconception carrier screening (e.g., cystic fibrosis, SMA)	Refer if both partners are carriers or high-risk ancestry present
Family Genetic History	Family history of genetic disorders, congenital anomalies, or inherited diseases	May indicate elevated genetic risk for offspring	Review detailed family history during preconception visits	Genetic counsellor for risk assessment
Prior Pregnancy History	Previous pregnancy affected by chromosomal abnormality or genetic condition	Recurrence risk may be increased	Review obstetric and genetic history	Genetics referral recommended
Paternal Age	Advanced paternal age associated with increased risk of certain genetic mutations	Linked to rare dominant conditions and neurodevelopmental disorders	Include paternal age in reproductive counselling	Consider genetics referral if concerns
Prenatal Screening Options	Non-invasive prenatal screening (NIPS), first trimester screening	Early identification of chromosomal abnormalities	Discuss during preconception counselling for older patients	Refer for counselling if abnormal results
Diagnostic Testing Options	Chorionic villus sampling (CVS), amniocentesis	Confirm chromosomal abnormalities	Explain availability during pregnancy	Refer to maternal-fetal medicine or genetics

Quick Clinical Triggers for Genetic Counselling

Consider referral to genetic counselling if patients have:

- Maternal age ≥ 35 years at expected delivery
- Family history of genetic disorders or congenital anomalies
- Prior pregnancy with chromosomal or genetic abnormality
- Known genetic carrier status in one partner
- Consanguinity (biological relationship between partners)
- Interest in expanded carrier screening or reproductive technologies
- Increased prevalence of specific genetic conditions

Clinical Pearl

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

Quick Clinical Actions

During visits with older reproductive-age patients:

- Ask about pregnancy intentions
- 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