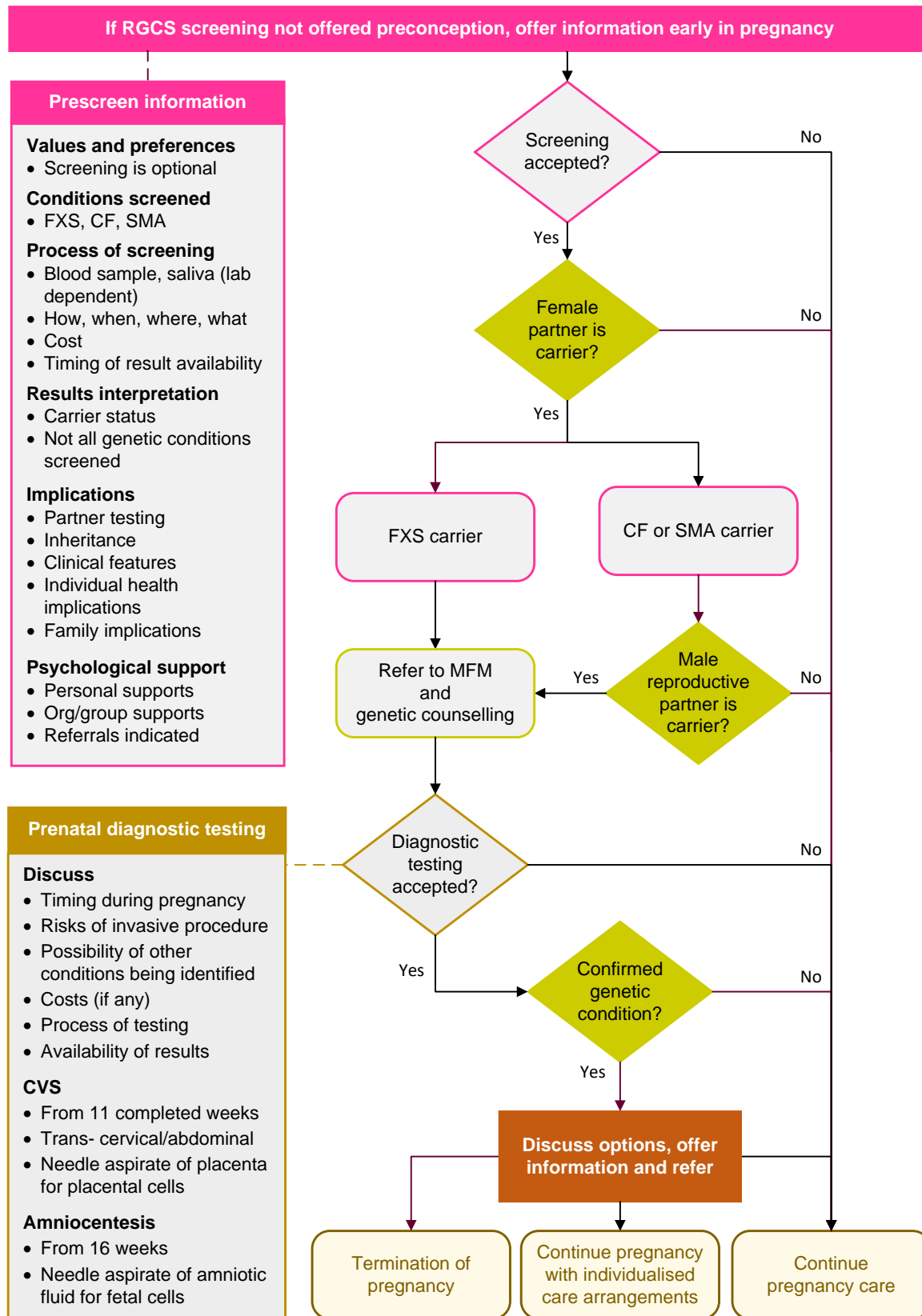


Reproductive genetic carrier screening during pregnancy



Prenatal diagnostic testing

Discuss

- Timing during pregnancy
- Risks of invasive procedure
- Possibility of other conditions being identified
- Costs (if any)
- Process of testing
- Availability of results

CVS

- From 11 completed weeks
- Trans- cervical/abdominal
- Needle aspirate of placenta for placental cells

Amniocentesis

- From 16 weeks
- Needle aspirate of amniotic fluid for fetal cells

CF: cystic fibrosis, **FXS:** fragile X syndrome, **MFM:** maternal fetal medicine, **NPV:** negative predictive value, **Org:** organisation, **PPV:** positive predictive value, **RGCS:** reproductive genetic carrier screen, **SMA:** spinal muscular atrophy
Individualised care arrangements: may include (as relevant to individual circumstances) increased antenatal surveillance, increased ultrasound surveillance, preparation for palliative care, or kinship or formal adoption, or foster care arrangements

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Queensland Clinical Guideline. Preconception and prenatal genetic screening. Flowchart F24.36-2-V1-R29