



Reasonable options for T21 screening in South Africa, given limited resources and limited access to more advanced screening and testing for all pregnant women.

Reality check: The ideal strategy of NIPT AND expert scans in all three trimesters is NOT achievable in our country for the foreseeable future.

The patient is a priori considered to have a low risk for fetal aneuploidy

Discuss options at the first antenatal visit

Offer first and/or second trimester serum screening for T21 + second trimester MSAFP + first AND second trimester scan by the general obstetrician (level 2 i.e. not basic obstetric scan but with standard review of fetal anatomy)

If any serum screening result indicates T21 risk > 1:1000, refer for:

- A: Expert assessment for risk adjustment, detailed ultrasound assessment (preferably still in the first trimester), genetic counselling and possible NIPT or invasive testing depending on the findings – highly recommended if serum risk > 1:300
- B: If option A is NOT available OR not accepted by the parents: offer NIPT

If serum MSAFP indicates increased risk for open neural tube defects or any scan finding of concern:

Refer for expert scan

If serum-based risks < 1:1000 AND both scans are apparently normal:

Inform parents that the risk is low but not zero – further testing is generally not advised but is available if parents insist (this should rather have been decided before screening was started)

The patient is a priori considered to have a high risk for fetal aneuploidy (previous aneuploidy or age > 35)

A: Request first trimester serum screening AND strongly recommend referral for an extended NT scan by an expert, for combined first trimester screening and early risk assessment with detailed ultrasound evaluation, with genetic counselling and possible NIPT or invasive testing depending on the findings

B: If option A is NOT available OR not accepted by the parents: offer NIPT

Authorship

These guidelines were drafted by a clinical team from Mediclinic and were reviewed by a panel of experts from SASOG and the BetterObs clinical team in 2019 and revised by the scientific subcommittee of BetterObs in 2022. All attempts were made to ensure that the guidance provided is clinically safe, locally relevant and in line with current global and South African best practice. Succinctness was considered more important than comprehensiveness.

All guidelines must be used in conjunction with clinical evaluation and judgement; care must be individualised when appropriate. The writing team, reviewers and SASOG do not accept accountability for any untoward clinical, financial or other outcome related to the use of these documents. Comments are welcome and will be used at the time of next review.

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