

## Trisomy 21 screening with FMF algorithm in South African private practice

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### Objective

As part of a bigger audit to determine the test performance of antenatal screening for trisomy 21 using alpha and Fetal Medicine Foundation (FMF) software respectively in South African private practice, the test characteristics of the different components of the FMF screening were determined.

### Methods

Screening data from 2010 to 2015 were retrospectively linked with pre- and postnatal genetic testing to assess screen positive and detection rates. The screen positive rate was determined from this database for FMF screening with nuchal translucency (NT) alone, combination screening with NT and biochemistry (BC), and combination screening with the addition of the nasal bone (NB) and either tricuspid regurgitation (TR) or ductus venosus flow (DV). The detection rate for each was calculated by determining the adjusted risk in 100 cases with trisomy 21 screened with the FMF algorithm.

### Results

The screen positive rate for NT screening was 7.2%, combined screening 5.4%, combined screening with addition of the NB 3.2%, and combined screening with addition of the NB and either TR or DV 3.8%. The screen positive rate for ultrasound only screening with NT, NB and either TR or DV was 8.0%. The detection rate of trisomy 21 with NT screening was 88%, combined screening 90%, combined screening with addition of the NB 88%, and combined screening with addition of the NB and either TR or DV 96%. The detection rate of trisomy 21 with ultrasound only screening with NT, NB and either TR or DV was 96%.

### Conclusion

In practice, the addition of more ultrasound markers tend to increase the detection rate, while the addition of biochemistry tends to decrease the screen positive rate.

Trisomy 21 screen positive rate (from audit data) and detection rate (calculated post hoc)

	NT	BC+NT	BC+NT+NB	BC+NT± NB + DV or TR	NT ± NB + DV or TR
Screen positive (audit data)	404/5534 7.3%	272/5049 5.4%	598/18929 3.2%	442/11651 3.8%	50/623 8.0%
DR (calc data)	88/100 88%	90/100 90%	69/78 88%	43/45 96%	43/45 96%