

SOUTH AUSTRALIAN MATERNAL SERUM ANTENATAL SCREENING PROGRAMME

Department of Chemical Pathology,
The Women's and Children's Hospital, Adelaide

Update 7 - May 1999

What's in an "increased risk of Down syndrome" report?

A report from the SAMSAS Programme stating "increased risk of Down syndrome" is always a matter for anxiety, especially after the dating of the pregnancy has been confirmed.

For those helping manage pregnancies it is also frustrating when, after all the counselling, ultrasonography, amniocentesis and anxiety, the karyotype on most of those declared at increased risk of Down syndrome is reported as normal. The question inevitably arises "what are we achieving here?"

The chances of an affected pregnancy

Between 1st January 1991 and 31st December 1997 the SAMSAS Programme screened 65328 pregnancies of South Australian mothers. After corrections of misdatings, 3481 (5.3%) were finally reported as being "at increased risk of Down syndrome".

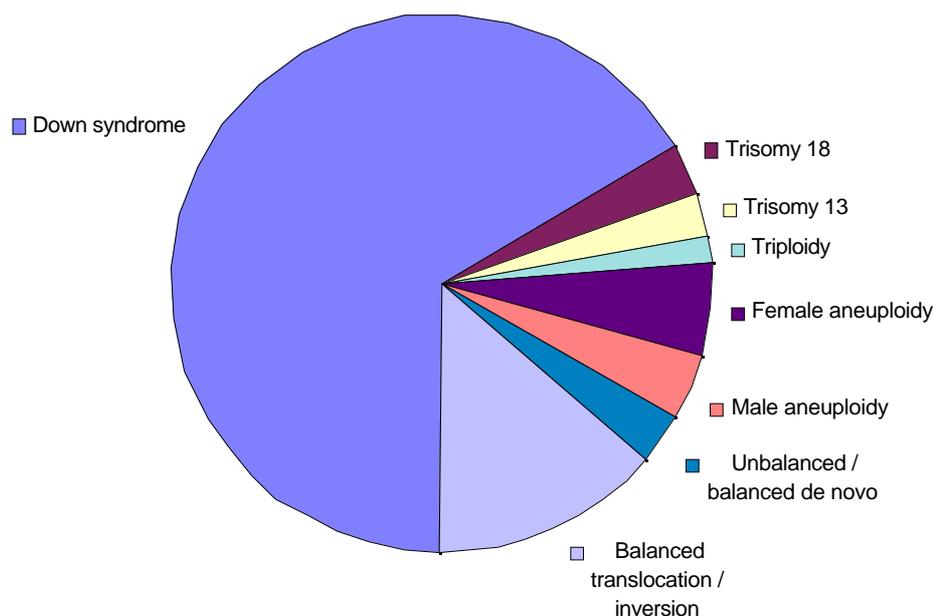
2748 of these 3481 mothers (78.9%) had amniocentesis and fetal karyotyping performed. 126 of these were carrying a fetus with an abnormal karyotype.

There was thus a confirmed abnormal fetal karyotype in 126/3481 (or 1 in 28) pregnancies reported by the SAMSAS Programme as being "at increased risk of Down syndrome".

Of the 733 unkaryotyped pregnancies reported "at increased risk of Down syndrome"

- early fetal loss occurred in 28
- 651 resulted in a liveborn infant who had no subsequent genetic investigations; these children are presumed to be genetically, or at least phenotypically, normal
- a live birth was not recorded in South Australia for the remaining 54 pregnancies.

Abnormal karyotypes found



The majority (84/126) of the abnormal fetal karyotypes found were, not surprisingly, Down syndrome. **1 in 41 pregnancies reported “at increased risk of Down syndrome” by the SAMSAS Programme were of a fetus with Down syndrome.** (This is the real figure behind the <1:50 risk appearing on our reports).

Also found were:

- 4 pregnancies with Trisomy 18
- 3 pregnancies with Trisomy 13
- 2 pregnancies with Triploidy (69,XXX)
- 7 pregnancies with Female Sex chromosome abnormalities
- 5 pregnancies with Male Sex chromosome abnormalities

all of which are associated with significant clinical abnormalities.

Four unbalanced/balanced *de novo* karyotypes, which have a 10% chance of resulting in a phenotypically abnormal child, were also present.

The remaining 17/126 abnormal karyotypes found were inherited balanced translocations or inversions which could be expected to result in phenotypically normal children.

In summary, **1 significant chromosomal abnormality was found for every 32 amniocenteses recommended by the SAMSAS Programme.** With over 80% of pregnancies affected by fetal Down syndrome being detected annually for the past three years, the SAMSAS Programme is continuing to be an effective part of obstetric care in South Australia.

Richard G. Ryall, PhD MAACB

on behalf of the South Australian Maternal Serum Antenatal Screening Programme