

Down's Syndrome Screening

Factors Affecting Screening

Element E

Aims and Objectives

- To identify and understand factors which may affect the Down's syndrome screening test result
- To act accordingly on information given to reduce variance of results
- Give accurate up to date information to all those involved in screening for Down's syndrome

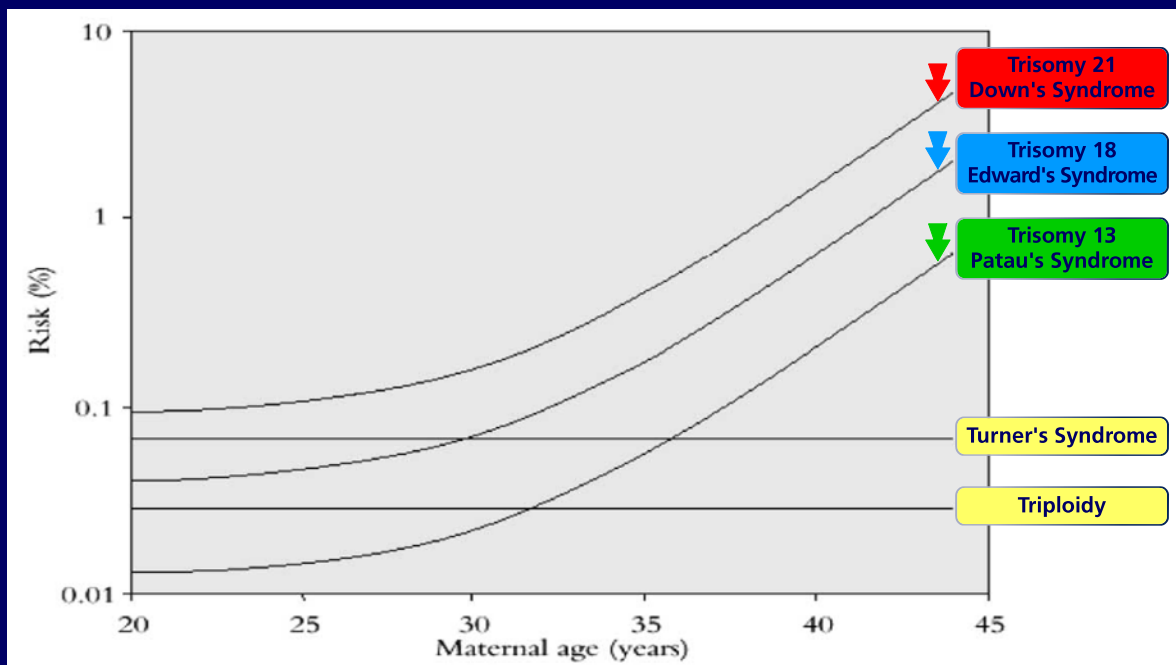
Factors Affecting Screening

- Maternal related
- Pregnancy related
- Programme related

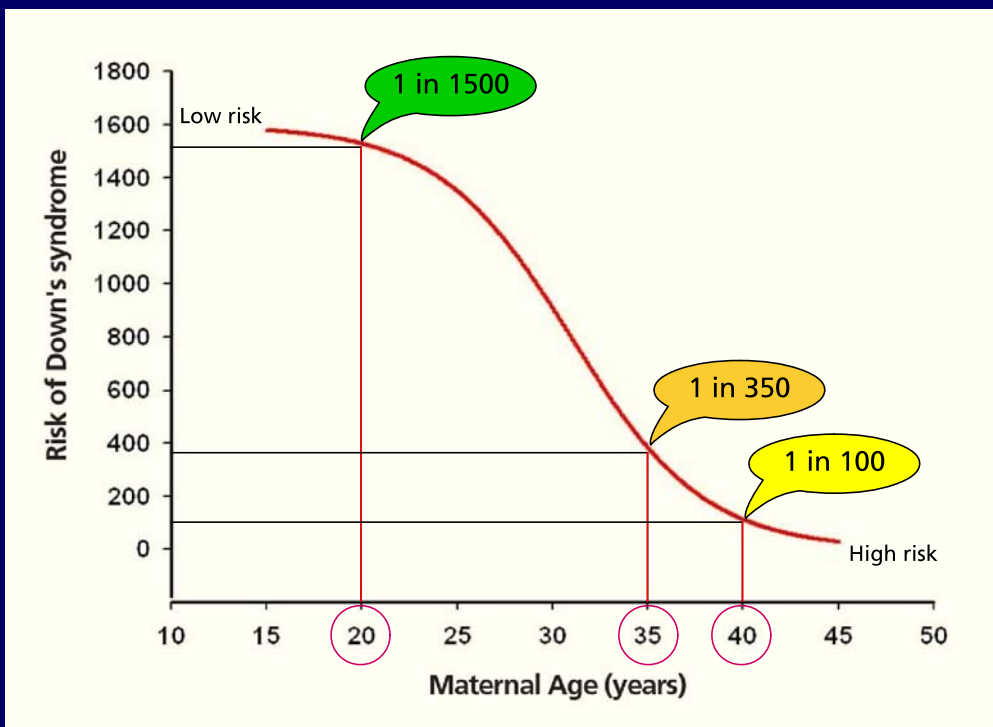
Maternal Factors

- Maternal age
- Weight
- Ethnic group
- Previous pregnancy with Down's syndrome
- Insulin Dependent Diabetics

Risk of Trisomy Increases with Maternal Age



Risk of Trisomy Increases with Maternal Age



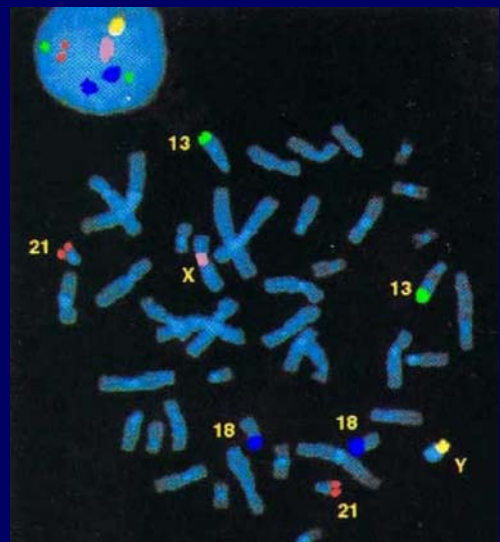
Ethnic Group

- There are slight differences in the average levels of the biochemical markers between different Ethnic origin
- Afro-Caribbean women in particular have higher levels of AFP and hCG when compared to Caucasian women
- It is important therefore to document the Ethnic origin

Wald et al 1997; HTA 1998; Vol.2: No 1

Previous Down's Syndrome Pregnancy

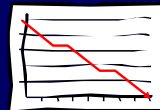
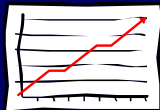
- There is an increased chance of a subsequent pregnancy being affected
- It is important therefore to document if there has been a previously affected Down's syndrome pregnancy and the type of Down's syndrome (i.e. inherited or non inherited)



Noble 1998; HTA 1998; Vol.2: No1

Maternal Weight

- There is an inverse relationship between biochemical marker levels and maternal weight
- So marker levels seem higher in lighter women and lower in heavy women



Maternal Weight

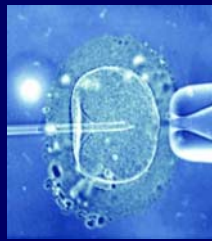
- Average weight is 63.4 kg
- Average decrease for each 20 kg increase in weight:-
 - Serum AFP $< 17\%$
 - Serum hCG $< 16\%$
 - Serum $UE_3 < 7\%$

Pregnancy Related Factors

- Gestational age



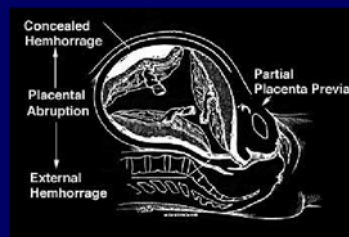
- Assisted conception



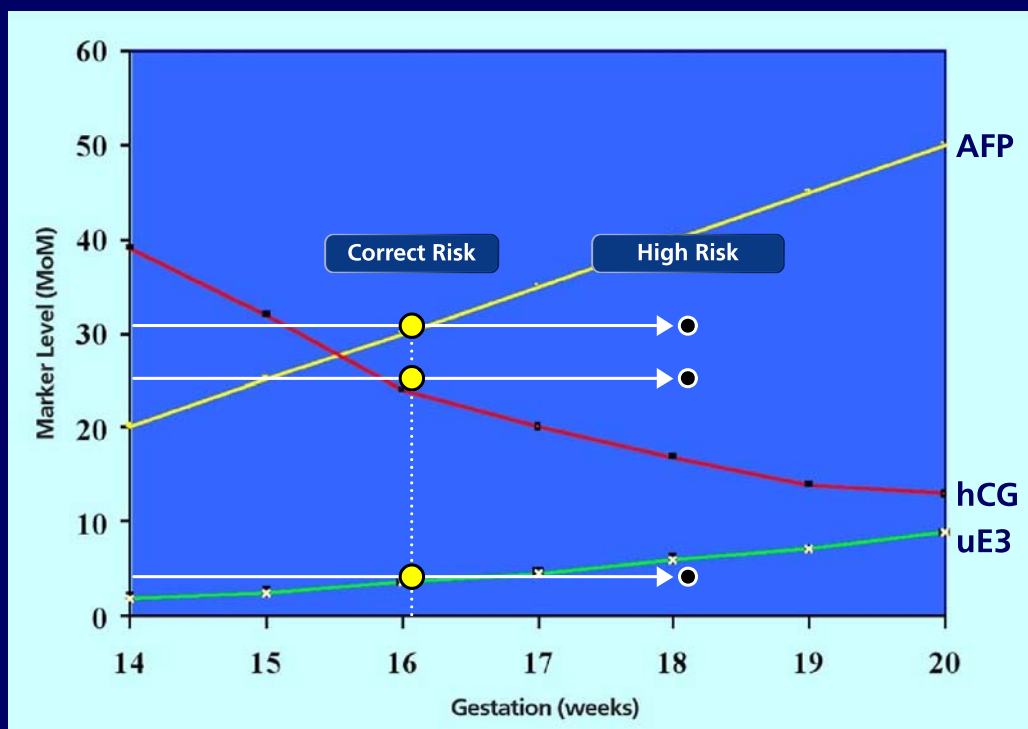
- Multiple pregnancy



- Bleeding in pregnancy



Gestational Age



Maternal Serum 'Markers' used in Antenatal Screening for Down's Syndrome

- May be referred to as 'Analytes'
- Tend to have low sensitivity when used alone
- Serum markers levels may be affected by maternal weight

Bleeding in Pregnancy

- Recent bleeding will increase AFP levels
- It is important therefore to document if any bleeding has occurred

Brennand JE; Cameron AD: Current methods of screening for Down's syndrome. The obstetrician and gynaecologist. October 2001: Vol.3: No 4

Multiple Pregnancies

Unaffected twin pregnancies

- We know biochemical marker levels are higher in twin pregnancies compared to singleton pregnancies at the same gestational age



Affected twin pregnancies

- We do not know with confidence what happens to biochemical marker levels in multiple pregnancies where one or more fetuses may be affected

Multiple Pregnancies

- Measurement of nuchal translucency as a screening marker in multiple pregnancies will enable the affected fetus to be identified



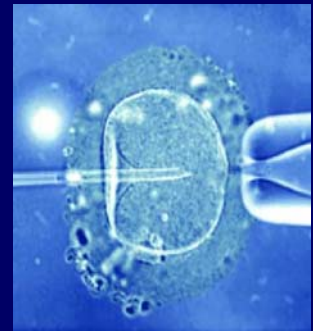
Assisted Conception

- **IVF** - In vitro fertilisation
- **ICSI** - Intra cytoplasmic sperm injection
- **GIFT** - Gamete intra fallopian transfer

- **Egg Donation**

It is essential that the age of the donor at the time of harvesting is clearly written on the screening request form

The risk of Down's syndrome is calculated on the age of the egg, not the woman's age



Assisted Conception

In pregnancies conceived by these methods

- hCG levels may be increased
- It is important therefore to document if a woman has undergone assisted conception

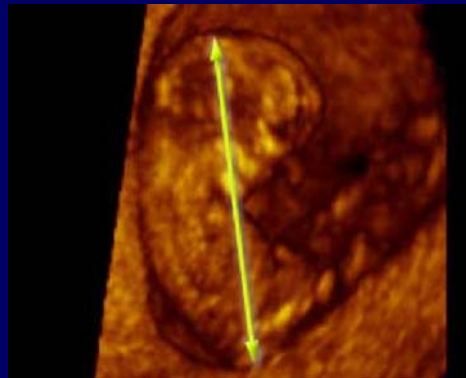
HTA 1998; Vol.2: No1
Donnenfeld AE et al 2002

Vanishing Twin

- If pregnancy started as a twin pregnancy and there has been a fetal demise this should be documented if serum screening is undertaken

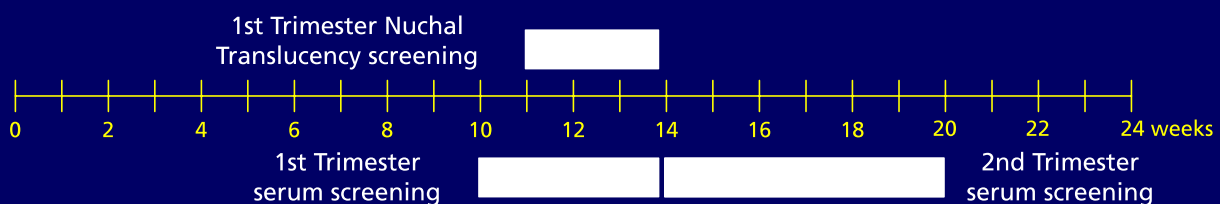
Programme Related Factors

- Timing of screening
- Accuracy of information
- Laboratory requirements
- Check your local policy



Timing of Screening

- 1st Trimester Nuchal Translucency screening should be undertaken between 11 weeks and 13 weeks + 6 days
- 1st Trimester serum screening should be undertaken between 10 weeks and 13 weeks + 6 days by ultrasound scan
- 2nd Trimester serum screening should be undertaken between 14 weeks and 20 weeks by ultrasound scan



Accuracy of Information

- **All** blood samples and forms **must** be clearly labelled
- Patients Name
Hospital Number
Date of Birth
Date of Sample
- Patients name, date of birth, hospital number and date of sample **must match** the details written on the sample tube
- Unlabelled samples **must not** be used and should be **disposed of**.

Accuracy of Information

- Correct gestation at time of sample following dating scan
- All information requested correctly documented
- All details should be checked by the person taking the sample

Guidelines for records and record keeping. NMC. 2002

Sample Collection

- Send to laboratory as soon as possible after collection
- Clotted blood sample in correct sample tube
- Ensuring sample is taken first to avoid contamination with EDTA
- Do not freeze the sample
- Audit and tracking of specimens



Examples of incorrect information, poor documentation and the effect this will have on screening results

Maternal Serum Screening	Clinical Information
Surname <u>Jones</u> D.O.B. <u>1.3.64</u> Forename <u>Sarah</u> Address <u>1 The Place</u> Hosp. No. <u>MAT 1234</u> <u>Anywhere</u> Hospital <u>St Elsewhere.</u> Other (please state).....	Date of Sample <u>17.2.03</u> Gestation calculated by (Please circle) USS LMP <u>14.10.02</u> Other 18/40 Weight <u>65kg</u> Sample: First ☒ Repeat ☐ Patient Insulin Dependant Diabetic? Yes ☐ No ☒
Investigation Requested	Infertility Information
(Please tick one only) AFP only ☐ AFP & Downs Risk ☒	IVF Pregnancy? No ☒ Yes ☐ Donor Egg? No ☒ Yes ☐ D.O.B of donor.....
Identification (must be completed)	
Signature <u>Annie Other</u> Print Name <u>Annie Other</u>	

Down's Syndrome Screening Report

Name:	Sarah Jones
Date of Birth:	01-03-1964
Date of sample:	17-02-2003
Age at EDD:	39 years
Gestation at sampling:	18 weeks and 0 days (by dates)
Weight:	65kg
AFP:	0.81 MoM
hCG:	1.73 MoM
uE3:	0.73 MoM
Risk of Down's Syndrome:	**HIGHER RISK** 1 in 85 (at term)

Down's risk based on maternal age alone is 1 in 150

Down's Syndrome Screening Report

Name:	Sarah Jones
Date of Birth:	01-03-1969
Date of sample:	17-02-2003
Age at EDD:	34 years
Gestation at sampling:	18 weeks and 0 days (by dates)
Weight:	65kg
AFP:	0.81 MoM
hCG:	1.73 MoM
uE3:	0.73 MoM
Risk of Down's Syndrome:	Higher Risk 1 in 240 (at term)

Down's Syndrome Screening Report

Name:	Sarah Jones
Date of Birth:	01-03-1969
Date of sample:	17-02-2003
Age at EDD:	39 years
Gestation at sampling:	16 weeks 0 days (by scan)
Weight:	65kg
AFP:	1.10 MoM
hCG:	1.11 MoM
uE3:	1.13 MoM
Risk of Down's Syndrome:	Lower Risk 1 in 3,100 (at term)