

GENETIC COUNSELLING & PRENATAL TESTING

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Some sobering facts

- Chance of a baby being born with a serious physical or intellectual handicap 1:50
- Chance a baby will be born with an abnormality of some kind 1:30
- Chance a pregnancy will end in miscarriage 1:4

Genetic Counselling - Referrals

- AMA / LMA : ≥ 35 yrs at delivery
- Chromosomal abnormalities eg., Down syndrome
- Chromosomal rearrangement
- PHx of abnormal sib or child / stillborn / NND
- Known AD, AR, XL or multifactorial disorders
 - ◆ Thalassaemia
 - ◆ neural tube defect
 - ◆ cardiomyopathy
 - ◆ cystic fibrosis (CF)
 - ◆ Huntington disease (HD)
 - ◆ FRAXA
- Consanguinity
- Abnormalities on ultrasound scan
- Multiple miscarriage
- Paediatric eg., dysmorphism, FTT, developmental delay
- Adult referrals eg., peripheral neuropathies

Genetic Counselling

- Multifaceted communication process
- May involve the diagnosis of a genetic disorder
- Seeks to enhance understanding
- Provides support
- Involves facilitation of decision making
'personal values, beliefs, abilities and lifestyle'

Reproductive Genetics

- can be emotionally charged
- grief from previous losses



Late Maternal Age (≥ 35 years)

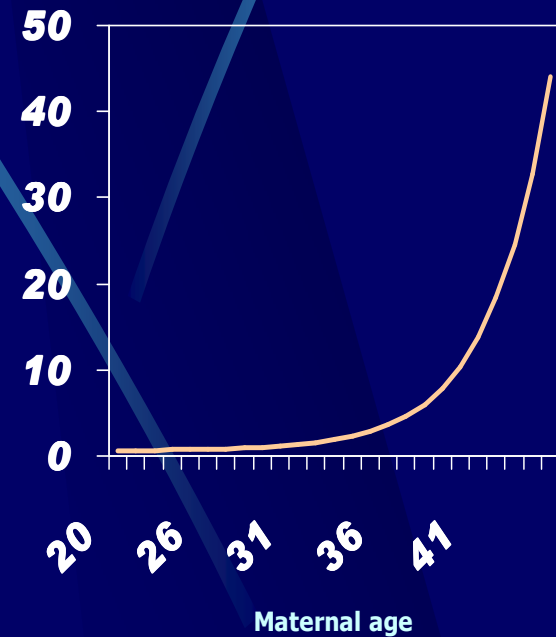
- most common indication for prenatal testing
- trisomy in the fetus
- chief condition is Down syndrome

Late Maternal Age (LMA) Advanced maternal age (AMA)

- maternal age effect in trisomies eg., Down syndrome
- predisposes to non-disjunction during meiosis
- 90% of trisomies are of maternal origin
- different chromosomes involved, most miscarry spontaneously
 - ◆ live birth possible with
 - 47,+21
 - 47,+13
 - 47,+18
 - 47,XXX
 - 47,XXY

Trisomy 21 or Down syndrome

- Population incidence 1/650
- Incidence very low if <25 yrs
- Risk rises above popⁿ risks >32yrs
- Numerical in ~95%
- Robertsonian translocation ~4%
- Recurrence risk ~1%



Types of testing

Screening

- ultrasound scan
- nuchal translucency
- combined first trimester
- maternal serum triple test (MSTT)

Diagnostic

- ultrasound scan
- chorionic villus sampling (CVS)
- amniocentesis

Screening Tests

- originally designed to detect high risk pregnancies within the low risk population
- at some age point tests provide no new information
- may have value for older women in assisting decision making
- optional
- informed consent
- require pre & post test counselling

Screening Tests

Nuchal Translucency Measurement

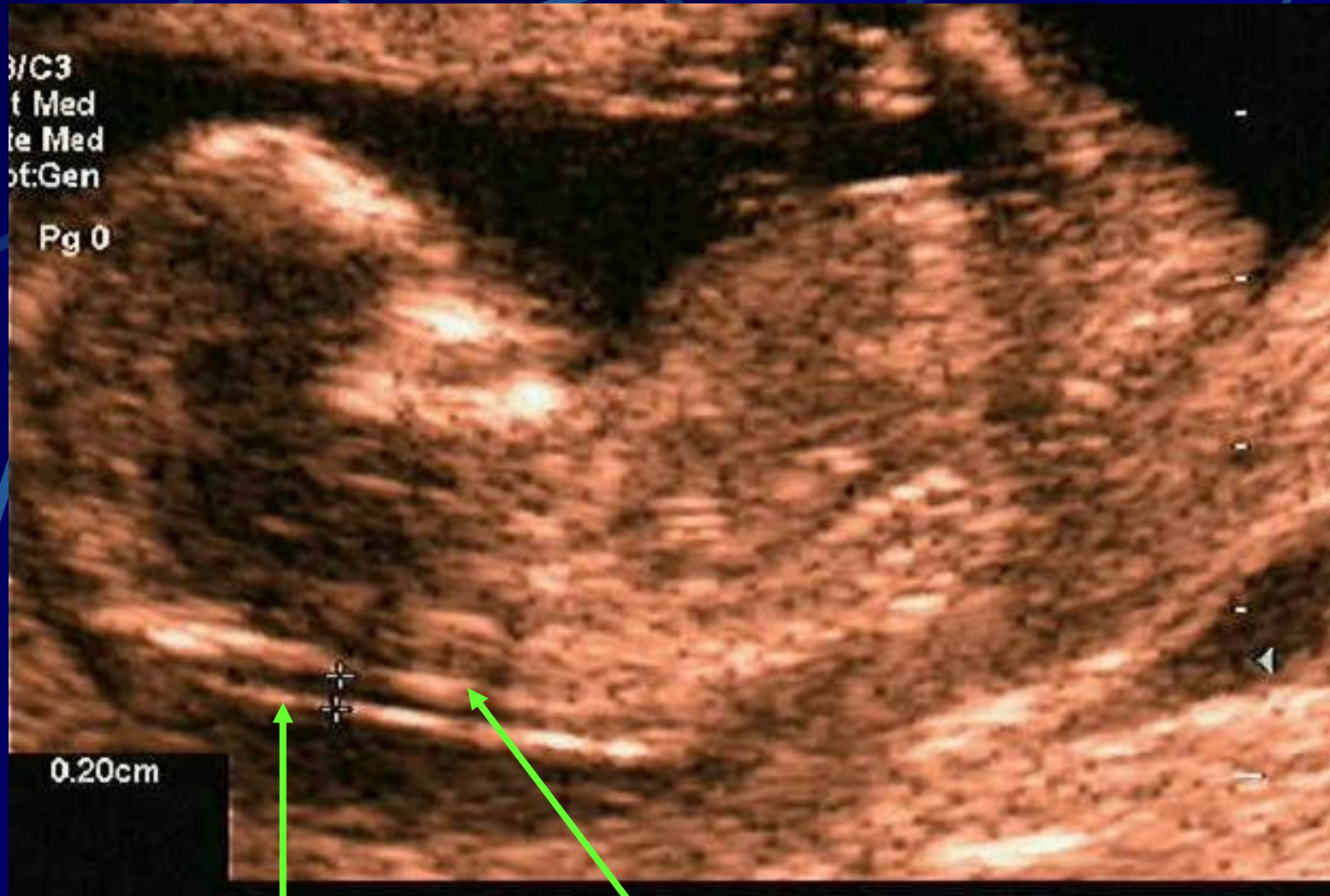
- 11⁺⁴ – 13⁺⁶ weeks
- 70 – 80% of trisomy 21
- 75 – 75% of other major trisomies
- 5% false positive risk
- age, gestation, NT, β hCG & PAPP A, *

Combined with

Serum Screen at 10 – 13 weeks

- 80 – 90% trisomy 21
- 5% false positive risk

Nuchal translucency



skin line

underlying tissue

Maternal Serum Triple Test

- 15 – 17 weeks
- 3 proteins + maternal age + gestation
uE, β hCG, AFP
- 60% of trisomy 21
- 5% false positive risk

Ultrasound Scan

Pre-test counselling

- why testing is available
- what conditions involved
- how / when is it done
- what test can & can't do – only a *risk*, not definitive
- is it important to know now if baby has Down syndrome ?
- would knowing the baby has Down syndrome change your decisions about the pregnancy?
- feelings about invasive testing?
- feelings about TOP for these conditions?

Counselling keys (both pre & post test)

- what do risk figures mean to the woman / couple?
 - ❖ priorities
 - ❖ beliefs & values
- definite vs screening test
 - ❖ weighing up options, balancing risks
- if an abnormal / high risk result
 - ❖ normalising feelings/thoughts they consider inappropriate
 - ❖ assist decision making

Nuchal Translucency

- high risk > 1/300

Results

LOOK AT THE ENTIRE REPORT!

- how does result compare to background (age) risk?
- why this risk?
- high risk does **not** mean the baby is affected!!

- \uparrow NT will \uparrow risk for T₂₁
- \uparrow β hCG + \downarrow PAPP A → \uparrow risk for T₂₁
- normal β hCG + \uparrow NT + \downarrow PAPP A → \uparrow risk for T₂₁
- \downarrow β hCG + \downarrow PAPP A → \uparrow risk for T_{13/18}

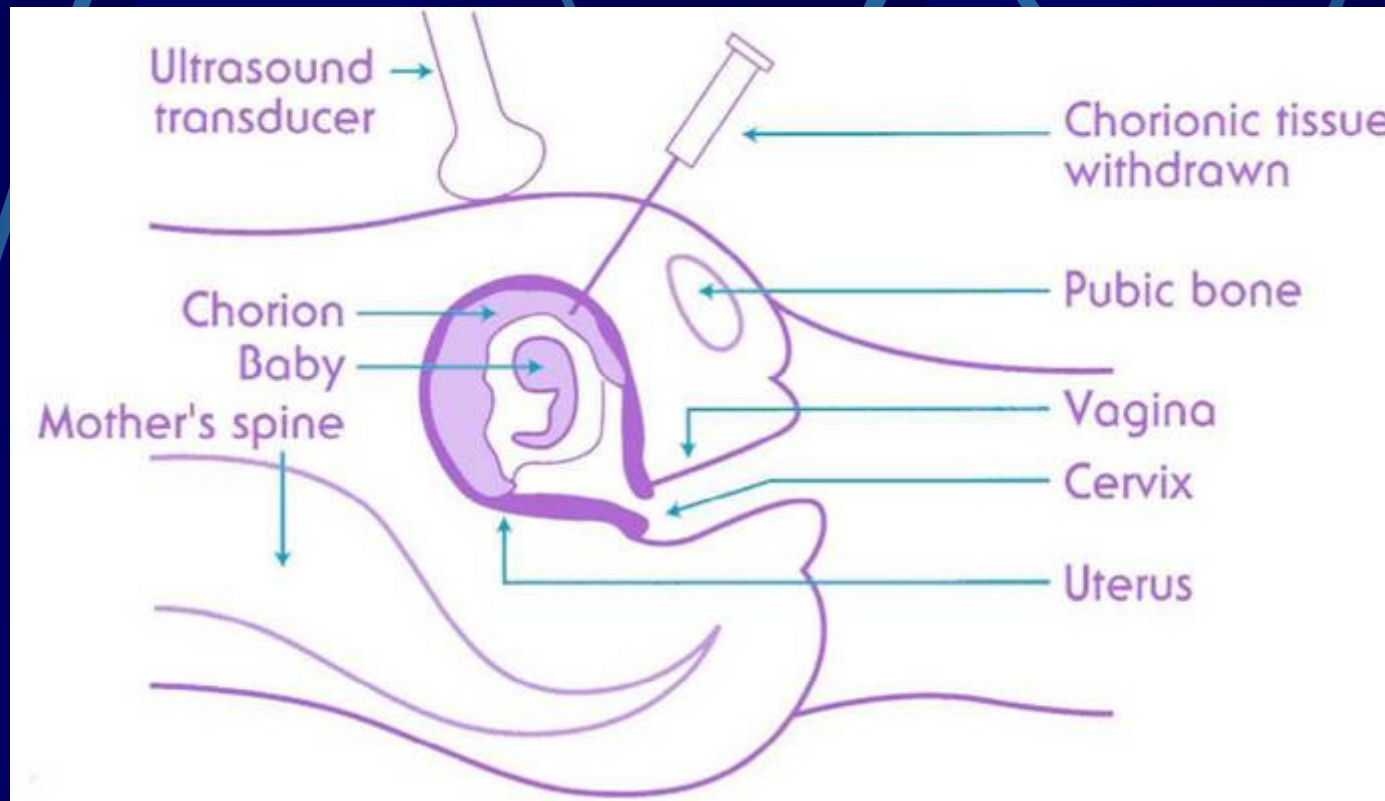
Anything else?

- NT > 3mm + normal chromosomes → fetal echo
→ 3° 19/40 USS
- PAPP A < 0.4 → USS around 32 – 34 weeks

Diagnostic Tests

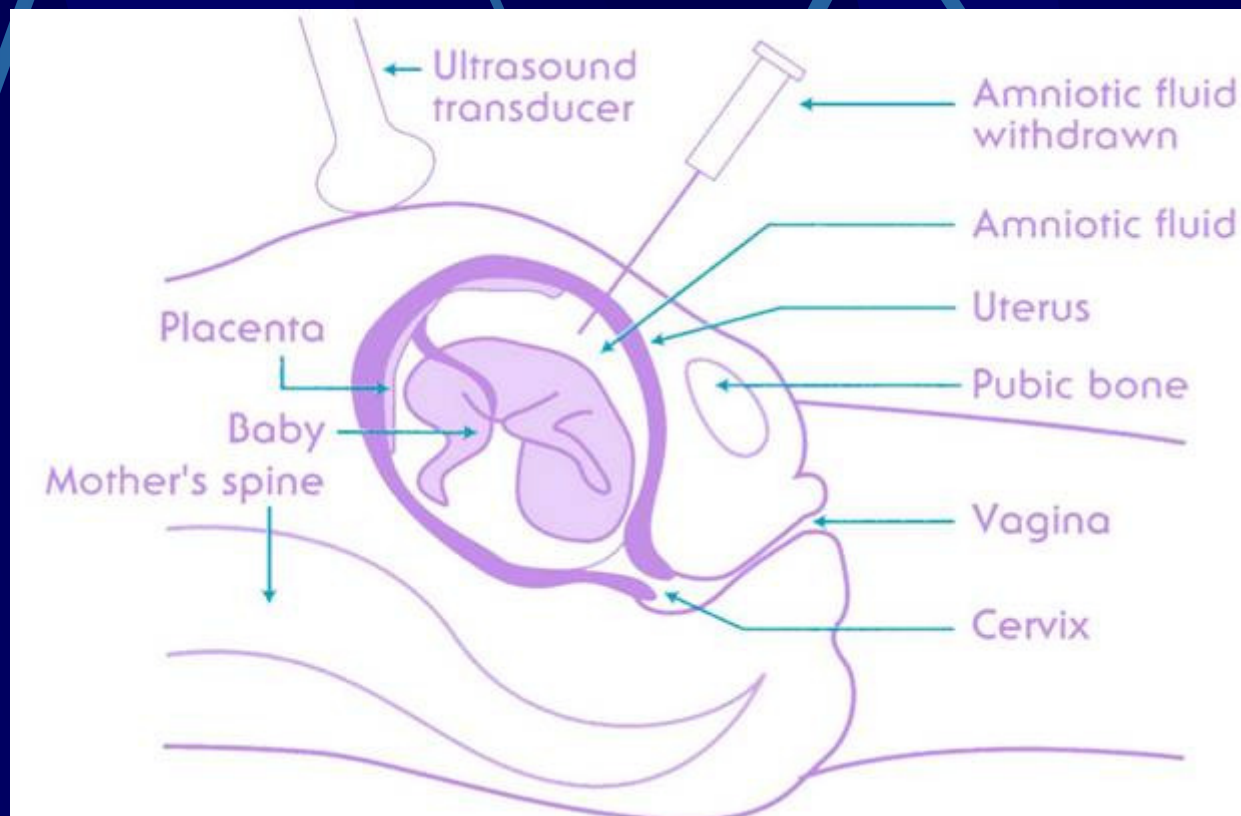
CVS

- 11 – 13 weeks
- miscarriage risk \sim 1% (100)
- accuracy \sim 99%

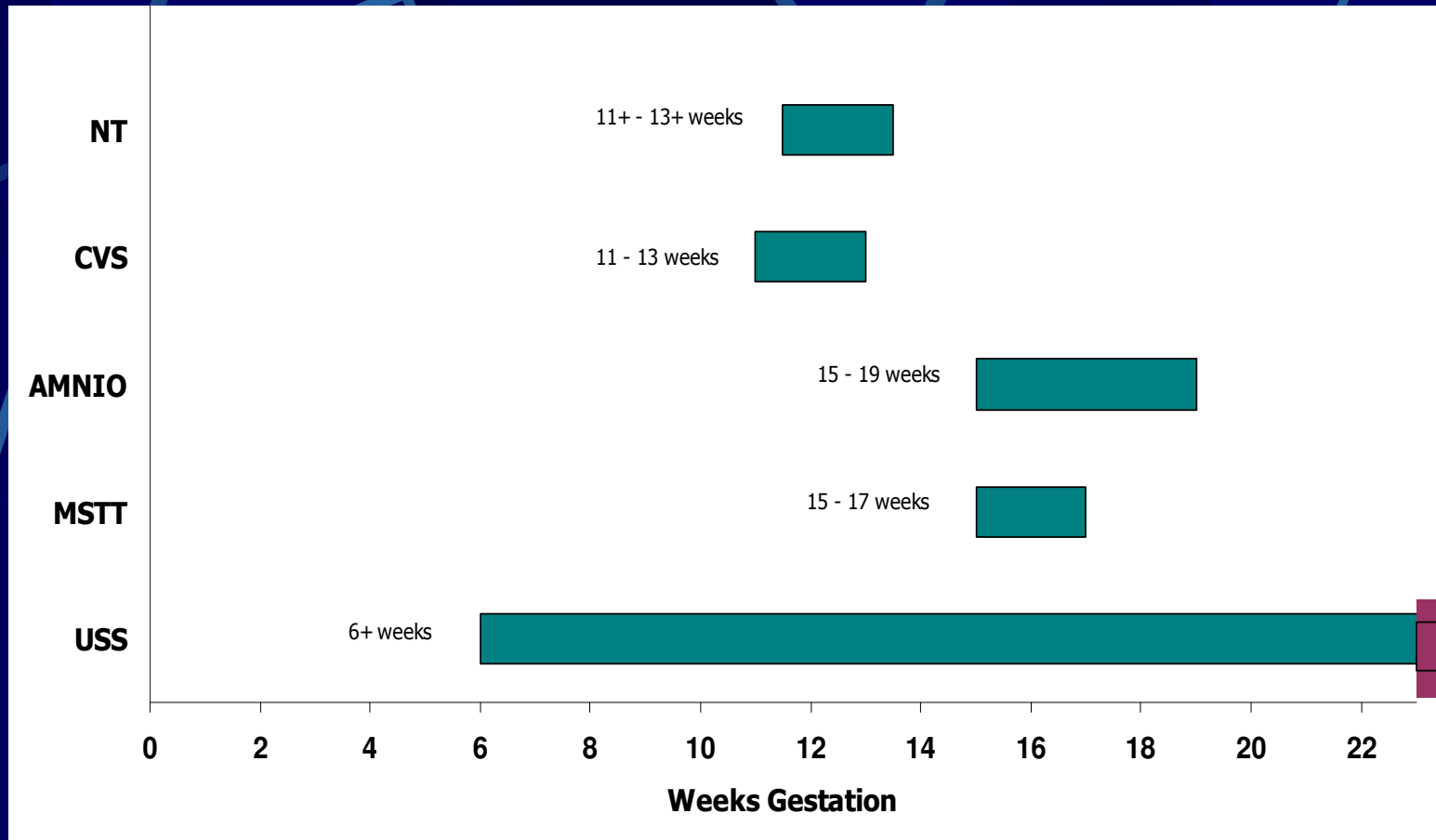


Amniocentesis

- 15 – 19 weeks
- miscarriage risk $\sim 0.5\%$ (1/200)
- accuracy $\sim 99\%$



Prenatal testing time line



Difficulties faced

- language *jargon vs everyday*

abnormality or problem, issue, difficulty
normal or as usual, as expected

- emotions - grief, aggression
- shy, embarrassed

- client not understanding

- culturally inappropriate

- time

Decision Making

- involves evaluation of alternatives and making choices between them
- simple strategies focus on a few facets of the options



Methods

- compensatory model - features 'weighed' for personal value
- elimination model - alternatives are considered & eliminated

Deciding between alternatives takes time!

Risky Decisions

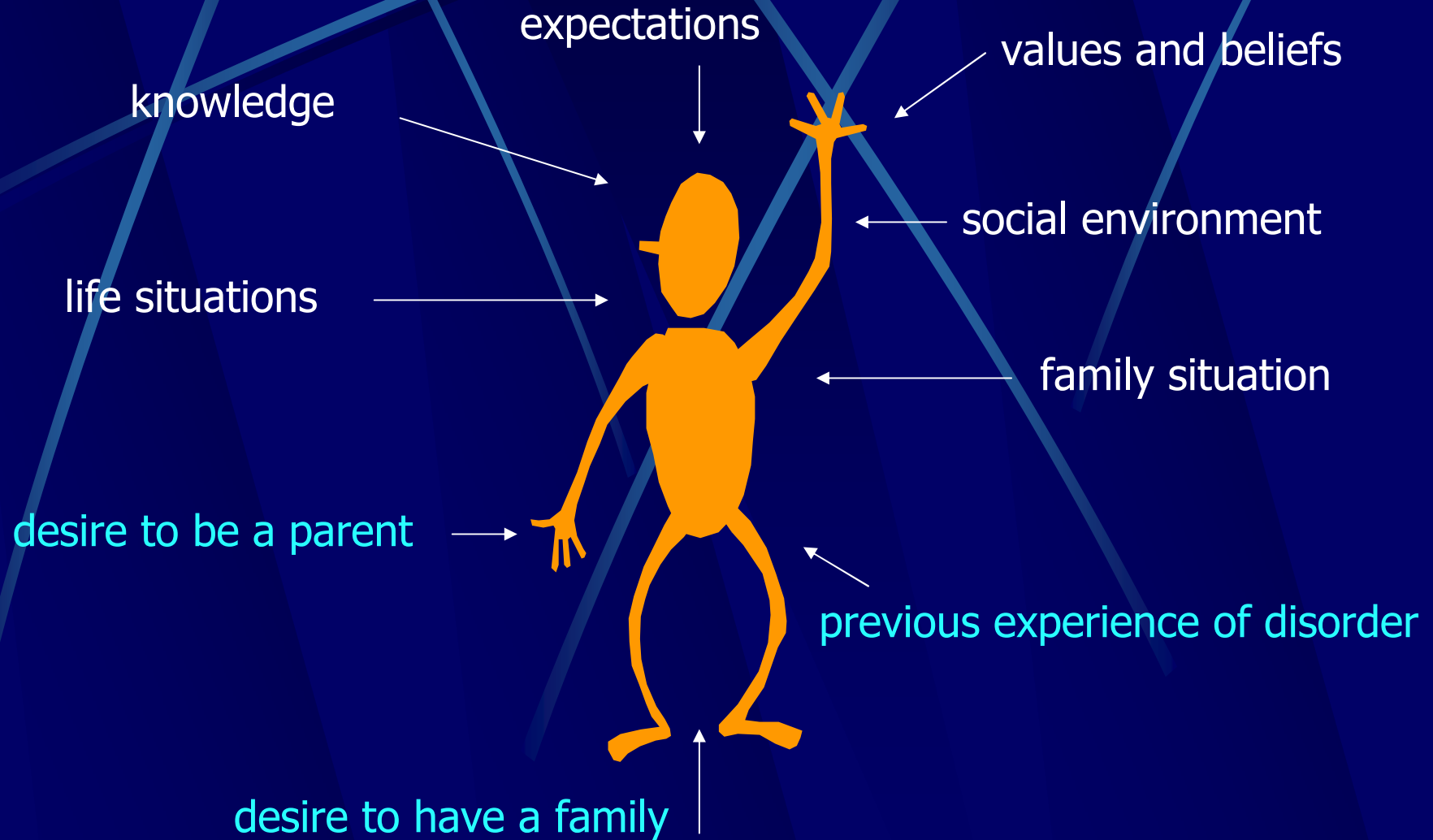
- conditions or outcomes are uncertain
- only probabilities are known

- personal worth of an outcome is a *major* factor



*Personal estimates of probabilities
are important*

Factors Impacting Prenatal Decisions



technical information

experience

beliefs

instincts

inner feelings

"transformative,
interpretive &
integrative
process"

mine



Not technical information vs personal ideas

Four processes

- the personalisation of maternal age
- a re-calculation of maternal age and its relation to Down syndrome
- a redefinition of the notion of fetal health
- the use of Genetic Counselling / GP to confirm their own knowledge claims



Personalisation of maternal age

- how does a women feel her age?
- how does she feel about her age?

Re-calculation of maternal age

“...didn't need an amniocentesis last pregnancy only 18 months ago”

“... my mother & sister both had babies >35 & they're OK, not Downs”

Notion of fetal health

- what does healthy mean?
- "...inner feeling everything will be OK..."
"...looked good on the USS..."

Using Genetic Counselling

- gain her Doctor's approval & put the matter to rest

women consider their own **feelings** about the pregnancy to be at least **as important** as any technical information



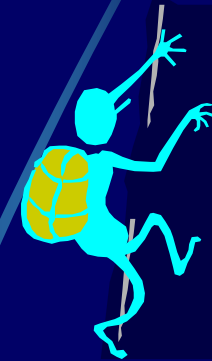
... choose an **approach consistent** with their own 'embodied' knowledge...

Counselling skills assist this process

Personal Control

Two main mechanisms

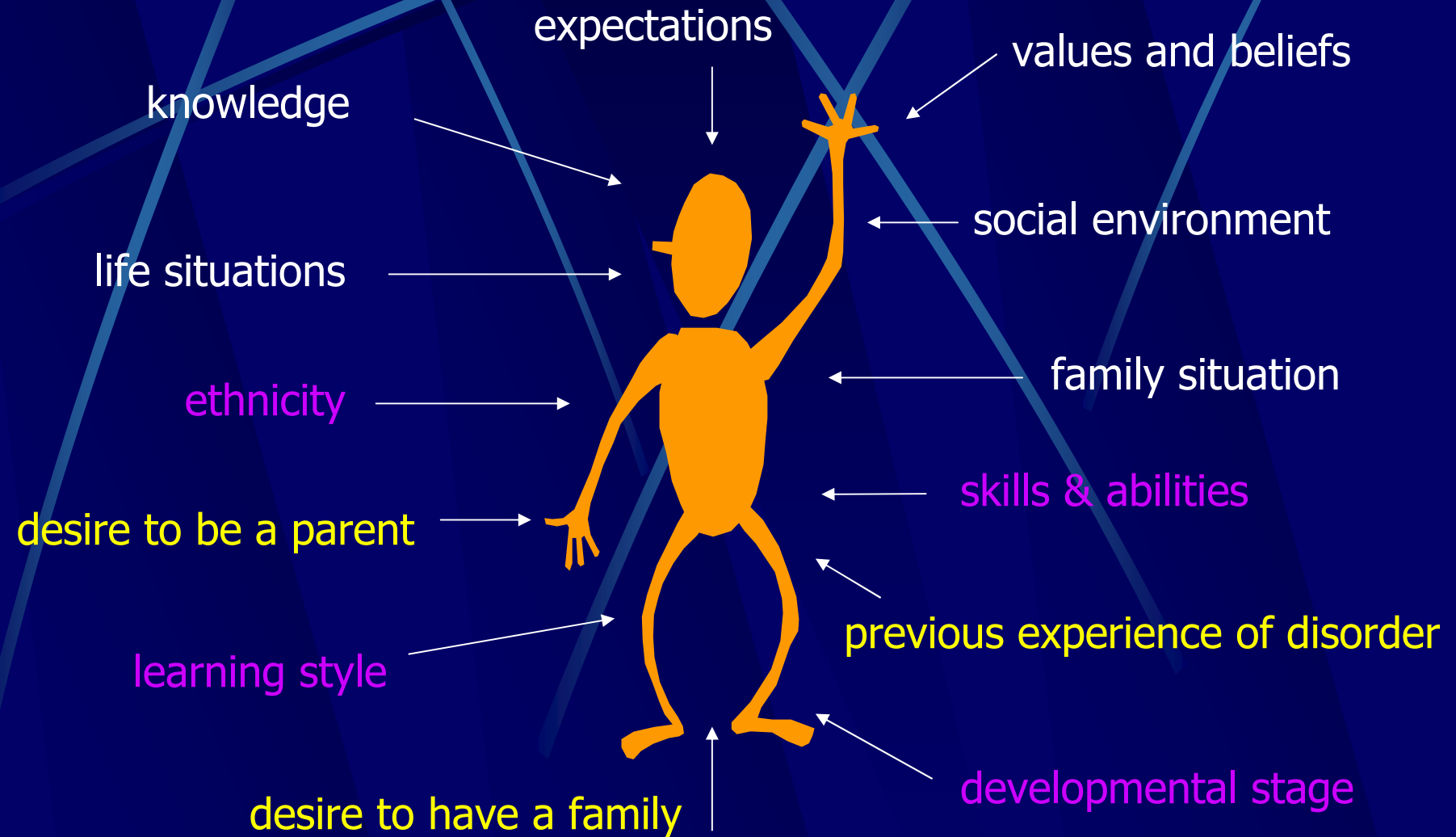
- search for information or understanding
- search for meaning



Averill's typology

Elements	Examples
behavioural control	prenatal testing
cognitive	never happened
decisional control	specific choices

Factors Impacting Prenatal Decisions



Implications

- organisation of information
- language
- style of presentation
- framing of information / issues
- personalisation of information
- time