CASE 1

Short case number: 3_4_1
Category: Endocrine & Reproductive Systems
Discipline: O&G
Setting: General Practice
Topic: Pre-conception care

Case

Harpa Singh aged 38 yo presents to your general practice. She is known to you as an insulin-dependant diabetic. She has recently married and is hoping to conceive. She asks how she should prepare for pregnancy and in view of her age what are the risks of a chromosomal abnormality.

Questions

1. Describe the factors you would discuss that help assess the risks for this pregnancy for Harpa and for her baby.
2. Outline the general advice and the particular advice you would give.
3. Outline the examination you would perform and list the investigations you would recommend.
4. Harpa is worried that at age 38 she is at increased risk to have a baby with an abnormality. Describe to her the current screening and diagnostic tests available.
5. What are the current Australian recommendations for the use of alcohol and smoking in pregnancy?
6. How might your advice change if Harpa had epilepsy, hypertension, thyroid disease, or depression?

Suggested reading:

- Abbott, J., Bowyer, L., & Finn, M. (2014). *Obstetrics and Gynaecology: an evidence-based guide (2nd ed).* Australia, Elsevier. Chapters 11 & 14

- RANZCOG College Statements and Guidelines, Prenatal screening and diagnosis of chromosomal &genetic abnormalities in the foetus in pregnancy. (C-Obs-59)

ANSWERS

1. Preconception counselling – risks of pregnancy to patient and foetus.

Individual – age, ethnic background, family history
Social – partner and family support, work, access to health care
Health behaviours – smoking, alcohol, recreational drugs
Psych – depression, anxiety, stress, abuse
Reproductive Hx – pregnancies and outcomes
Previous medical history-any ongoing medical problems
Current medications
Vaccinations, particularly rubella, varicella zoster if seronegative, also Diphtheria, tetanus, pertussis, measles.
Nutrition – vegetarian, malabsorption,

Family history – If a high risk of a chromosomal or genetic disorder then pre-pregnancy genetic testing and counselling may enable couple to consider relative merits pre-implantation genetic diagnosis versus prenatal diagnosis in pregnancy.

2. General advice:

Nutrition – preconception folic acid, diet (meat and green vegetables for iron, 3 serves dairy products for 1000mg calcium, measures to avoid listeria. Increased fluid intake and adequate fibre to avoid constipation), supplement 150mcg iodine, role of multivitamins
Drugs – quit program for smoking, recommend avoid alcohol and recreational drugs
Sex – STI screen, hepatitis, HIV
Exercise
Weight control
Dental hygiene
Medication advice (e.g. anti-hypertensive, anti-epileptic, anti-depressant etc.)

Reassure that conception may take up to one year
Record dates of period on a calendar
General outline of antenatal care and first trimester screening
Early pregnancy symptoms (nausea, breast tenderness, fatigue, urinary frequency)
If she has a cat avoid contact with its faeces, risk toxoplasmosis.

Specific advice

Risk foetal congenital abnormality high (cardiac defects and neural tube abnormalities) and to prevent this
Strict diabetic control for conception and pregnancy. Monitor HbA1C and keep in normal range.
Insulin needs will increase through pregnancy.
Refer to endocrinologist for reassessment diabetes, renal and eye disease
Folic acid 5mg (usually 0.4mg) for one month pre-conception at least and until 12 weeks pregnant
5mg if: previous baby or family history neural tube defect, diabetes mellitus, epilepsy, BMI > 30, malabsorption,

3. Examination: Height and weight for BMI, Blood pressure, auscultation of heart and lung fields, Breast examination and abdominal palpitation. Check for lymphadenopathy and thyroid swellings. **Pelvic examination if relevant.**

4. Investigations:

- Cervical Screening test
- Rubella and varicella serology, FBC, ferritin if vegetarian or heavy menses, blood group, Vitamin D levels if appropriate.
- Genetic test if indicated
- Urine analysis

50% of babies with a chromosomal abnormality are born to women younger than 35. All pregnant women should receive counselling and be offered screening for chromosomal abnormalities, chromosomes 21, 18, 13.
Risk Down's syndrome 1 in 300 at age 35 and 1 in 100 at age 40.

Screening Tests
Combined first trimester screen incorporates maternal age, U/S measurement of the foetal nuchal translucency and maternal serum markers levels (Papp-A, Pregnancy-associated plasma protein A & betaHCG) to generate an overall risk figure for trisomy 21.
Cell-free DNA (cfDNA) NIPT Non-Invasive Prenatal Test. DNA from foetus found in maternal blood. May need repeat in obesity.
Morphology U/S at 18 weeks

Investigation Value	Gestation	Sensitivity	Specificity	Positive Predictive
Combined first trimester Mat age+NT+betaHCG+Papp-A	11-13^6 weeks	85%	95%	7-10%
Quadruple Test (late booking) Mat age+AFP+betaHCG+UE3+	15-20 weeks	75%	95%	2-3%
Cell-free DNA screening (NIPT)	>10 weeks	99%	99%	45%

Diagnostic Tests
All abnormal screening tests require confirmation.

Amniocentesis. Foetal cells are cultured from amniotic fluid aspirated from uterus. Performed under U/S guidance from 15 weeks. <14 weeks risk talipes.

Chorionic villus sampling. After 11 weeks aspiration under U/S of chorionic villi for genetic testing. Before risk transverse limb reduction defects.

Conventional (G-banded) Karyotyping use cultured foetal cells.
Rapid aneuploidy tests – FISH fluorescent insitu hybridization.
Adjunct to above for rapid assessment.
Chromosomal Microarray analysis. Assess foetal genome – if foetal abnormality detected at U/S.

5. Alcohol and smoking recommendation

Excessive alcohol can damage foetal development. The minimum or threshold level at which alcohol begins to pose a significant threat to pregnancy is not known. Avoidance of alcohol in pregnancy is recommended. Smoking increases the risks of infertility, miscarriage, adverse events in pregnancy,

respiratory disease in the baby and sudden infant death syndrome. Women should be screened for smoking and given counselling and behavioural support. This includes written information, referral to QUIT. Nicotine replacement therapy is not recommended in pregnancy.

6. How might your advice change if Harpa had epilepsy, hypertension, thyroid disease, or depression?

Some medications present increased risks for foetal abnormality or growth and need to be changed prior to conception. Always check in MIMS/ with the respective specialist that the current medication is safe in pregnancy. Referral for pre-natal foeto-maternal review may be indicated. Serum levels of thyroxin and anti-epileptic medications decline with the increasing haemodilution of pregnancy. Serum levels need to be checked throughout the pregnancy and dosage adjusted as indicated.

smoking cessation

4 Advising on smoking cessation in pregnancy

Why is this important?

Cigarette smoking by pregnant women can harm not only their own health, but also that of the foetus. Smoking during pregnancy doubles the risk of having a low birthweight baby.[1] It is also associated with a higher risk of spontaneous abortion, premature birth, stillbirth, sudden infant death syndrome (SIDS), cleft palate and cleft lip, and childhood cancers.[2]

In Australia, smoking rates for women are higher in younger age groups. The prevalence of smoking among women generally peaks between the ages of 20 and 24, remains high for those aged 25 to 29, then starts to decline.[3] High smoking rates happen to coincide with peak fertility, given that the average age of first-time mothers is 27 years and 29 years for mothers generally.[4] It should not be a surprise then that approximately 30 per cent of women are smokers when they become pregnant and about 20 per cent smoke during pregnancy.[5]

Best available evidence

There is compelling evidence from a large number of trials that smoking cessation programs can be effective in reducing smoking rates among pregnant women, and consequently in reducing the adverse effects on both mothers and babies.[6]

Pregnancy is a time when many women are motivated to make healthy lifestyle changes, including quitting smoking, and it is also a time when they have regular contact with health professionals.[7] In Australia, nearly all women who give birth attend some sort of antenatal care, whether it is provided by general practitioners, obstetricians, midwives, nurses, antenatal clinics or antenatal classes.

For pregnant women, psychosocial treatments (such as self-help materials and counselling) are preferred over pharmacotherapies such as nicotine replacement therapy, because the pharmacotherapies carry some risk to the foetus. The data show that extended psychosocial interventions, such as those involving more intensive counselling, work better than minimal advice.

Although stopping smoking early in the pregnancy will be of greatest benefit to the foetus and expectant mother, smoking cessation at any point during pregnancy is beneficial.[5]

Current practice

Generally there is limited information on whether antenatal care providers in Australia are identifying and counselling pregnant women who smoke. However, the available data suggest it is not routine practice. For example, a 2002 study found that no smoking cessation advice was given to almost three-quarters of women smokers who were 30 weeks into pregnancy and had contacted health care providers.[8]

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