compensate for the stress of living in a poor environment; and recreational facilities are not as accessible for people with no transport or who have small children.¹⁷ These factors might need to be modified first if the promotion of healthier lifestyles is to be successful.

The new contract requires general practitioners to provide checks every three years for patients aged 16-74 years. It is thus of some concern that our findings reinforce those of other studies suggesting that unselective health checks might be an inefficient use of valuable resources and might promote further inequalities in health. Despite their simplicity and economic attractiveness health checks may have only a limited part to play in any comprehensive plan for health promotion as they cater primarily for the fitter and better motivated members of the public. The global strategy required to combat ischaemic heart disease must incorporate political will, in the form of public education and strong price and advertising controls to reduce the consumption of saturated fats and tobacco, as well as enthusiasm, organisation, and vigilance by primary health care teams. The medical profession should not be cajoled into accepting complete responsibility for health promotion and the prevention of ischaemic heart disease, so absolving the government of its obligations.

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Screening in Practice

Maternal and fetal screening for antenatal care

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The chief aim of antenatal care is to maintain and improve the health of both mother and fetus during pregnancy so that they are in optimum condition when parturition occurs. Subsidiary objectives are the early diagnosis of variations from the norm and the detection of asymptomatic, potentially threatening conditions that may arise. Screening is, in fact, a term that broadly covers this process of detection and for many years it has been accepted as an integral part of good maternity care. The procedures used were at first mainly directed at preserving the wellbeing of the mother but, particularly during the last two decades, innovative techniques have been introduced that enable indirect but sophisticated monitoring of the fetus.

Screening before conception

Preparation for pregnancy can sometimes be combined with the provision of contraceptive advice. For example, rubella state should be determined, and if the woman is not immune she can be immunised before contraception is abandoned. Many women are increasingly concerned about other infective hazards to the fetus, such as toxoplasmosis, so a comprehensive serum TORCH screen (toxoplasmosis, rubella, cytomegalovirus, herpes virus) may be appropriate, at least from the viewpoint that a mother shown to be immune to these conditions can be reassured that reinfection during pregnancy is unlikely. At the same time the woman's ABO blood group and rhesus factor should be determined (if they are not already known)

to warn of the possibility of isoimmunisation during pregnancy. Similarly, haemoglobinopathies should be looked for—for example, sickle trait in Afro-Caribbean women and thalassaemia in those of Mediterranean or Asian origin.

A family history should also be obtained regarding possible genetic anomalies, and if any are present formal genetic counselling should be arranged before conception occurs. This is especially necessary where there is a history of X-linked or autosomal defects, chromosomal rearrangement, or any major developmental abnormality. A considerable advance in preconceptual counselling has been the development of a technique for gene tracking in prospective parents and their families. It uses DNA probes and markers and allows assessment for the risk of such conditions as Huntington's chorea, cystic fibrosis, muscular dystrophy (Becker, Duchenne, or myotonic), neurofibromatosis, and familial polyposis coli as well as haemophilia A and B and the haemoglobinopathies. Guidelines on multistage screening and tests for carrier states were published in 1987 in the King's Fund consensus statement on screening for fetal and genetic abnormality.¹

Screening during pregnancy

Screening procedures during pregnancy can conveniently be associated with the key points during antenatal care when the mother would be seen by her doctor² rather than by her midwife. These are at

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Screening before and during pregnancy

	Urine	Blood	Chorionic villus sampling	Amniotic fluid	Ultrasonography	Vaginal swabs
Preconception	Diabetes Nephropathy	Rh state Rubella state Haemoglobinopathy Carrier states				
8-10 weeks	Bacteriuria	Syphilis Hepatitis (HBsAg)	Karyotype Gene defects			
16-18 weeks		Anaemia Rh or ABO antibodies α Fetoprotein		Karyotype α Fetoprotein Acetylcholinesterase	Multiple pregnancy Gestational age Macrosomal defects	
28-30 weeks		Anaemia Rh or ABO antibodies Gestational diabetes		Haemolytic disease	Intrauterine growth retardation Placental site	
34-36 weeks	Pre-eclamptic toxemia	Anaemia Rh or ABO antibodies Pre-eclamptic toxemia		Haemolytic disease	Intrauterine growth retardation	Herpes simplex virus Group B streptococcus

booking (8-10 weeks), at 16-18 weeks, at 28-30 weeks, and at 34-36 weeks of gestation. The range of tests by gestational stage is summarised in the box.

At 8-10 weeks

This is the time when most women present for confirmation of pregnancy. If preconceptual screening has not been undertaken the appropriate investigations and counselling should be carried out.

A urine sample should be tested to exclude diabetes or nephropathy, and if pregnancy is not clinically certain this sample can be used for a pregnancy test. A clean catch sample should also be examined to exclude asymptomatic bacteriuria.

Blood samples should be taken for full blood count, electrophoresis (in appropriate ethnic groups), ABO and rhesus grouping, determination of rubella state (if not already known), testing for toxoplasma if requested, and serology for syphilis. Screening for hepatitis B carrier state (HBsAg) is already routine in some areas and seems justifiable as early passive-active immunisation of babies born to seropositive mothers has been shown to be effective.³ It seems likely that HIV state will also be a mandatory screen in future.

Chorionic villus sampling should be offered to any women aged over 35 and those who have experienced a previous pregnancy with chromosomal abnormality or are known to have a single gene disorder for which the pregnancy is at high risk and which can be detected prenatally. Amniocentesis at a later stage (16 weeks) may, however, be preferable as it is believed to carry a lower risk of miscarriage and has the advantage that screening for neural tube defect can be done at the same time. Genetic defects can be detected more rapidly with chorionic villus sampling, however, and should termination of pregnancy be an option it can be undertaken with less distress to the patient if performed before the end of the first trimester.

At 16-18 weeks

Blood should be sent for determination of α fetoprotein concentrations to screen for neural tube defect. Gestational age should be confirmed by ultrasonography and women with α fetoprotein concentrations more than 2.5 times the mean for gestational age should be offered a high resolution ultrasound scan; if this is indeterminate they should be offered amniocentesis.

An ultrasound scan should be arranged routinely to exclude multiple pregnancy, confirm gestational age, and define the site of the placenta. If there is a history of previous macrosomal defects (cardiac or renal tract disorders, limb reduction defects, osteogenesis imperfecta, etc) a high resolution anomaly scan should be done at 18-20 weeks' gestation.

Amniocentesis should be offered to women over 35 or with a history of chromosome or gene defects who have not been screened by chorionic villus sampling or who have raised serum α fetoprotein concentrations. A raised α fetoprotein concentration in amniotic fluid may be disregarded if there is no concomitant rise in acetylcholinesterase activity and if an anomaly scan is normal. A low serum α fetoprotein concentration may be associated with Down's syndrome, particularly in older women,⁴ and if this is found then karyotyping of fetal cells from the liquor amnii is also indicated.

At 28-30 weeks

This assessment is to provide a baseline for the third trimester. Developmental anomalies are unlikely now but patients should be screened for such conditions as gestational diabetes, ABO and Rh isoimmunisation of the mother, the development of pre-eclampsia, and growth retardation in the fetus.

Blood should be taken for a full blood count, to exclude iron deficiency or macrocytic anaemia. In Rh negative women the blood group should be checked and isoimmunisation excluded. If anti-D antibodies are detected the test should be repeated within two weeks. If a rising titre is found amniocentesis may be indicated to confirm haemolytic disease and to assist in decisions regarding the timing of the delivery. A random plasma glucose concentration should be estimated in obese patients and women who have previously had very large babies or who have had glycosuria. This should preferably be done more than two hours after the last meal.⁵ If glucose concentration is found to be raised (>5 mmol/l) a fasting sample should be checked, and if it is still abnormal a formal glucose tolerance test should be performed.

Ultrasound examination should be performed if at the previous examination at 16-18 weeks the placenta had been encroaching on the lower segment of the uterus; as the uterus enlarges the placenta is usually drawn up away from the cervix but it is important to establish that no degree of praevia persists. If the fetus seems clinically to be small for gestational age then growth

parameters should be established and a further scan arranged three or four weeks later to check that fetal growth is not retarded.

At 34-36 weeks

Urine testing is routine at every antenatal examination, but as the pregnancy approaches term proteinuria is more likely to signify pre-eclampsia, so other causes such as contamination of the sample with vaginal secretion or a urinary tract infection must be positively identified. This is done with a carefully collected midstream specimen that is examined microscopically and sent for culture.

Blood is again checked at this visit for evidence of iron deficiency or other anaemias. The blood group should be checked and ABO or rhesus antibodies sought. If there are symptoms or signs of pre-eclampsia then raised concentrations of plasma urea and urate, and activity of aspartate aminotransferase provide confirmation and have prognostic value.

Ultrasound examination is again indicated if there is clinical evidence of fetal growth retardation or if, in the very obese patient, there is doubt regarding the presentation of the fetus.

Radiological examination (a single lateral view of the pelvis taken with the patient standing) is advisable in nulliparous patients when the fetus is in breech presentation. This allows the true conjugate measurement of the pelvic brim to be obtained, which together with the shape of the sacral curve may influence decisions regarding the appropriate method of delivery.

Vaginal swabs should be taken at this point (a) if the woman complains of symptoms suggesting candidiasis or other genital infection—after a pathogen has been identified appropriate treatment should be given before the onset of labour; the hazard to the fetus in the birth canal can thus be reduced; (b) if primary herpes genitalis has occurred during the pregnancy, swabs in virus medium should be sent for culture (management of this situation is somewhat contentious,⁶ so if swabs prove positive the advice of a virologist should be sought); (c) if a group B streptococcus has previously

been cultured from the vagina. If the result is still positive (as it may be in up to 15% of women) the neonate should be observed with special attention in the first few days of life and itself be screened for bacteria and given prompt chemotherapy in the event of positive culture or fever (streptococcal septicaemia causes perinatal death in about two babies per 1000, an avoidable tragedy).⁷

Conclusion

Advances in molecular biology, immunology, and genetics have now enabled more than one hundred biochemical defects to be identified, either in the prospective parents as carriers or in an affected fetus. Perhaps the most exciting development is the possibility of screening for cystic fibrosis⁸: one in 20 of the white population carries the newly identified defective gene; the incidence of this distressing and potentially fatal condition is approximately one in 2000 live births. Now that the means is available, the ethics and logistics of carrier screening in reproductive populations for this and similarly identifiable recessive conditions will need to be considered.

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