# *Prenatal Screening*

## *What is prenatal screening?*

Prenatal screening can tell us which pregnancies might have a greater chance of certain genetic problems. Screening does not say that an unborn baby actually has the disorder—only prenatal testing can tell you that.

## *What common problems can be screened for?*

- Problems with a single gene that affect the blood, such as thalassemia or sickle cell anemia
- Problems that are more common in some family backgrounds, such as Tay-Sachs disease or cystic fibrosis.
- Some chromosome* problems, such as Down syndrome
- Problems of the spinal cord and brain (neural tube defects), such as spina bifida or anencephaly

* A chromosome is the carrier of hereditary characteristics (e.g., eye colour). A child receives a chromosome from each parent.

## *Who should be screened and when?*

Every woman, ideally *before* becoming pregnant, should talk to her doctor about:

- any previous problems with pregnancy, such as 3 or more miscarriages, a stillborn child, or a child born with a physical problem or a chromosome problem
- a child in your or your partner's family who died shortly after birth or within the first few years of life
- a member of either family with a genetic or chromosome problem

It's important to know that your background (i.e., your race, origin, or ancestry) could be a factor as well. For example, you might want to consider prenatal screening if you or your partner is: Pakistani, East Indian, Bangladeshi, Southeast Asian, Chinese, Filipino, Mediterranean, Middle Eastern, African (including African-American), Caribbean, or Ashkenazi (northeastern European) Jewish.

If any of the above apply to you or your partner, talk with your doctor about screening. The choice of having screening is entirely up to you.

**Note**: Your doctor may take your age into account, but we know that age alone is not a good way to predict your chances of having a baby with a genetic problem. Thus, *any* woman who is pregnant may be offered certain types of tests, such as maternal serum screening.

## *What tests does screening involve?*

Usually a simple blood test and/or an ultrasound.

## *What do the results mean?*

It depends on which disorder is being screened for.
For example, for Down syndrome:

- A positive result doesn't mean your baby has the disorder. It only means the chance of your baby being born with Down syndrome is higher than average.
- A negative result means the chance of your baby being born with Down syndrome is lower than average (but the chance isn't zero).

For a blood problem like thalassemia:

- A positive test means you carry one normal gene and one gene for thalassemia. Thus, the baby's father should also be tested. Your baby has a chance of having thalassemia only if *both* of you carry the thalassemia gene.
- A negative test means you don't carry the gene for thalassemia, and your baby will not have this blood disorder.

## *What happens after a positive result?*

Talk with your doctor. He/she will discuss which further tests you may want to consider. Depending on your situation, you may also be offered a referral to a genetics program. Again, the choice of further testing or counselling is entirely up to you.