NEWBORN SCREENING BC

Information sheet for health care providers

Newborn Screening: Hemoglobin Traits and Benign Variants

Newborn screening in BC includes a screening test to identify babies with sickle cell disease and related hemoglobin disorders. As a result of this screening, some newborns are identified who have an atypical hemoglobin pattern that is not associated with a clinic disease state. This may be a hemoglobin trait, meaning the newborn has one copy of a hemoglobin variant and one copy of normal hemoglobin (a carrier), or it may be a combination of hemoglobin variants that is considered clinically benign. These hemoglobin patterns are not associated with clinical disease state, but could have implications for future reproductive decision-making.

Traits:

- Sickle cell trait
- Hemoglobin C trait
- Hemoglobin D trait
- Hemoglobin E trait
- Hemoglobin variant carrier

Combinations:

- Homozygous hemoglobin C
- Homozygous hemoglobin D
- Homozygous hemoglobin E

Points to discuss with the family (see the accompanying pamphlet)

- Newborn screening has **not** identified a disorder that requires treatment or monitoring.
- Carriers of hemoglobin variants are common among people of African, Asian, Southeast Asian, Middle Eastern, Latin American, Caribbean and Mediterranean descent.
- Babies carrying one of these variants are **not** more likely to get sick than other babies.
- No special treatment is required. This will not change into a disease later on.
- In almost all cases, at least one parent will also be a carrier for this variant.
- There is a small chance that both parents are carriers of a hemoglobin variant which could pose a risk of having a child with a blood disorder in their **next** pregnancy.
- Testing is available to find out if either or both parents carry this hemoglobin variant or a different variant.

Parental and Family Testing

Parents and adult family members of these newborns may want to know if they carry a similar trait. **This testing can be arranged by their primary care provider** by requesting both a **complete blood count** and **"Hemoglobinopathy Investigation"** through any community laboratory. This blood test will provide a profile of the hemoglobin forms present, and will identify carriers for the hemoglobin variants listed above. Carrier testing for these hemoglobin variants does **not** require a molecular (DNA) test.

Genetic counselling is available if both reproductive partners have a hemoglobin variant/thalassemia trait and they are at risk of having a child with a clinically significant hemoglobin disorder. The genetic counsellor will review inheritance, reproductive implications and testing options for future pregnancies. A referral can be made to the Provincial Medical Genetics Program in Vancouver (604-875-2157) or Vancouver Island Medical Genetics in Victoria (250-727-4461). A genetic counselling referral is not recommended if only one partner has a hemoglobin variant.

If you have questions regarding a couple's hemoglobinopathy screen result and their potential reproductive risk, please contact the Medical Genetics clinic and speak to a genetic counsellor.

Provincial Health Services Authority

Newborn Screening BC is a collaboration of Provincial Lab Medicine Services, BC Children's Hospital and BC Women's Hospital and Health Centre, and Perinatal Services BC, all part of the Provincial Health Services Authority.