Australian Paediatric Surveillance Unit

STUDY PROTOCOL

COMMENCING January 2004

Haemoglobinopathies (excluding carrier states)

BACKGROUND

Haemoglobinopathies are recessively inherited blood disorders for which there is no cure except, under certain circumstances, bone marrow transplantation. While haemoglobinopathies are rare, gene carriage occurs with high frequency in certain populations. For example, the frequency of the carrier state for ß-thalassaemia is as high as 1 in 5 in Greece and Italy, the frequency of the carrier state for sickle cell anaemia is as high as 1 in 5 in equatorial Africa and alpha thalassaemia and haemoglobin E carrier status are common amongst Asian populations.

The number of children in Australia affected with a haemoglobinopathy is unknown. However the number of cases of haemoglobinopathies in Australia may be increasing due to changes in the composition of the population[1]. In contrast, it has been suggested that the incidence of these disorders may be decreasing overseas because of widespread screening programs including carrier screening and counselling of couples at marriage, pre-conception or in early pregnancy[2].

In all states of Australia, selective screening is the current policy to identify carriers of haemoglobinopathies[3]. Thalassaemia carrier testing is recommended on an ad hoc basis to individuals from high-risk ethnic groups in teenage and early adult years. Carriers can also be detected on routine blood films carried out in early pregnancy. Additionally, screening with a full blood examination alone will not detect carriers of sickle cell disease, and haemoglobin electrophoresis is necessary. Also, there are many individuals in Australia who may not be recognised as being from high-risk ethnic groups (such as second or third generation Southern Europeans) and they may, consequently be unaware of their carrier status or the potential risks of these conditions for their children.

STUDY OBJECTIVES

This study seeks to estimate the incidence and types of haemoglobinopathies in Australian children and their distribution amongst ethnic groups and collect information on the timing and method of diagnosis of haemoglobinopathies in Australia. This information will contribute to efforts to improve the detection and outcome of affected children.

CASE DEFINITION

Report all children under 15 years of age seen in the previous month with a newly diagnosed haemoglobinopathy including:

- structural haemoglobin abnormalities resulting from changes in the amino acid sequence of the globin chains
- thalassaemias, in which the synthesis of one or more of the globin chains is decreased or totally suppressed.

Conditions to be reported include:

- ✓ *Hb SS disease (sickle cell anaemia)*
- ✓ *Hb CC disease*
- ✓ *Hb EE disease*
- ✓ *ß-thalassaemia major*
- ✓ *Hb E/ ß-thalassaemia*
- ✓ *Hb S/ ß-thalassaemia*
- ✓ *Hb SC disease*
- ✓ *Hb H disease*
- ✓ *Hb Barts disease*
- ✓ *Other rarer, severe haemoglobin variants*

Please <u>do not report carrier states</u> such as alpha-thalassaemia minor, beta-thalassaemia minor or heterozygous forms of haemoglobin variants that are generally asymptomatic

FOLLOW-UP OF REPORTED CASES

A brief questionnaire requesting further details will be forwarded to clinicians that report a case of haemoglobinopathy to the APSU.

If you have any comments or questions please contact:

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INVESTIGATOR CONTACT DETAILS (*Principal Investigator)

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REFERENCES

1. Guidelines for diagnosis and management of haemoglobinopathies. NSW Health Department Circular August 1999; 98/4807-5.

2. Cao A, Galanello R, Rosatelli MC. Prenatal diagnosis and screening of the haemoglobinopathies. Baillieres Clinical Haematology 1998;11(1):215-38.

3. National Public Health Partnership. An overview of public health surveillance of genetic disorders and mapping of current genetic screening services in Australia. National Public Health Partnership Report October 2002.