



APSU STUDY

HAEMOGLOBINOPATHIES

(Excluding carrier states)

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The study developed from an initial experience in my clinical practice in the south west of Sydney

An infant born to healthy parents of Lebanese origin became anaemic and was found to have sickle cell disease. The parents had no idea they were carriers and had not been tested

The child later developed serious complications including recurrent splenic sequestration and needed splenectomy by age 1

RATIONALE FOR THE STUDY

Although blood screening recommendations exist throughout Australia that address the issues of thalassaemia, sickle cell disease and other inherited haemoglobin disorders these protocols are not preventing some cases of these disorders occurring

Changes in population with increased immigration from Asia and the Middle East have produced increasing numbers of 'at risk' families particularly in urban centres

STUDY OBJECTIVES

To document the incidence of new diagnoses of haemoglobinopathies in Australia

To provide data that can help judge the effectiveness of current screening programs

To increase awareness of these conditions amongst the population and medical professionals

DEMOGRAPHICS

ABS figures: 1991: 25% overseas born
2001: 29% overseas born
Sydney: 35%, rural 5%
Auburn: 56%, Hawkesbury 2%

Languages : Chinese 4.3%
Arabic 3.3% (Sydney figures)
Italian 1.5%

INITIATION OF STUDY

Establishment of a group of investigators consisting of general paediatricians, geneticists and clinical haematologists from NSW and Victorian hospitals and an epidemiologist from Dept of Human Services Victoria

Submission of proposal and protocols to APSU – accepted after modifications

Other sources of ascertainment such as genetic labs suggested – being evaluated

?WHICH DISORDERS

WHO estimates that 6% of the world population carry a haemoglobin disorder and 7% of children born today are carriers. There are over 700 haemoglobin disorders but the majority are of little significance.

Therefore the common carrier states alpha thalassaemia minor, beta thalassaemia minor and common asymptomatic heterozygous forms of haemoglobin variants were excluded. HbEE was included although usually relatively asymptomatic.

PROTOCOL - BACKGROUND

Carrier state beta thal up to 20% in Greece and Italy, sickle nearly 20% in central Africa and alpha thal and HbE carrier status common in Asians

Current screening guidelines recommend selective screening on ad hoc basis in high-risk ethnic groups. Routine FBC showing microcytosis mandate further testing and partner testing. However Hb electrophoresis is required to pick up sickle carriers as FBC usually normal. 2nd generation effect

CASE DEFINITION

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

Structural Hb abnormalities resulting from changes in the amino acid sequence of the globin chain

Thalassaemias, in which the synthesis of one or more of the globin chains is decreased or totally suppressed

CONDITIONS TO BE REPORTED

Hb SS (sickle cell anaemia)

Hb S/beta thalassaemia

HbS/C

Beta thalassaemia major

HbEE

HbE/beta thalassaemia

HbCC

HbH + OTHER SEVERE VARIANTS

HbBarts

QUESTIONNAIRE

34 questions

Ethnicity, consanguinity, family history, parental awareness of the problem, antenatal diagnosis and brief details of treatments requested

PRELIMINARY DATA

**10 notifications received for the months of
January, February, March and part of April 2004**

7 questionnaires returned

Disorders: Hb SS (2)

HbS/beta thal

Hb E/beta thal (2)

beta thal major

Hb Zurich

PRELIMINARY DATA

Ethnicity: African American/African American

Middle East/Middle East

Burmese/Fijian Indian

Greek Cypriot/Italian

Caucasian/Caucasian

Italian/Italian

Thai/Thai

No children tested antenatally. Mothers aware of status: 5/7. Fathers aware : 2/7

INVESTIGATORS



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Financial Assistance from AHEPA gratefully acknowledged