Australian Paediatric Surveillance Unit

PROTOCOL - CONGENITAL ADRENAL HYPERPLASIA

OBJECTIVES:

i) to determine the incidence of classical and non-classical CAH in Australian children
ii) to compare case ascertainment from a trial period of newborn screening in NSW with that of APSU reporting
iii) to characterise selected clinical aspects of patients with CAH

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SUMMARY PROTOCOL:

Congenital adrenal hyperplasia (CAH) is a rare, potentially life-threatening condition with significant morbidity. Most CAH presents in the newborn period, but non-classical cases present later. The Australian Paediatric Surveillance Unit (APSU) provides a framework by which information on the incidence and clinical characteristics of new cases of CAH can be studied. For CAH presenting in the newborn period, a unique opportunity has arisen to survey CAH by the APSU simultaneously with a pilot study of newborn screening which will be performed by the Oliver Latham Laboratories in NSW. The APSU survey aims to collect information on all new cases of CAH diagnosed under the age of 16 years.

Australia does not currently have newborn screening for CAH, although this has been practised in Sweden, New Zealand and Scotland and some regions of Italy, France, Japan and the USA for some time. Screening involves measurement of 17- hydroxyprogesterone on newborn dried blood spots used for the screening of other metabolic disorders. Comparisons of newborn screening data with case search studies suggest that a significant number of cases of early-onset CAH are missed on clinical grounds and argue for screening. Delayed diagnosis has a risk of neurological morbidity. Missed diagnosis may also occur, death possibly being attributed to some other cause eg. sepsis. The APSU survey of CAH will occur simultaneously with a proposed pilot CAH screening program in NSW, commencing in late 1995 for a period of 2 years. This will screen all newborn infants in NSW for 21-hydroxylase deficiency. Comparison of cases detected by screening in NSW with those reported to the APSU in NSW and other states during this period will give an indication of the efficacy of newborn screening.

While the major focus of this study is on the neonatal presentation of CAH, information which is lacking in the literature on non-classical CAH will also be obtained. Thus, potential benefits of the proposed survey include obtaining information on:

1. the number of cases of classical CAH being missed in the absence of newborn screening, and hence an indication of the possible efficacy of a newborn screening program.
2. the number of cases of non-classical CAH detectable by newborn screening. These would otherwise only be detected later in childhood, by which time significant morbidity may have ensued.
3. the incidence of new cases of CAH (classical and non-classical) in Australia
4. an Australian overview of clinical and biochemical characteristics of CAH at presentation

CASE DEFINITION:

Any new case of CAH in a child less than 16 years, confirmed biochemically by elevations in adrenal steroid precursors[*]
with or without
clinical evidence of salt-losing or simple-virilizing CAH (clinical features may include: ambiguous genitalia, salt-wasting, adrenal crisis, virilization, hypertension).

* elevated basal levels or synacthen test stimulated levels of 17-hydroxyprogesterone and/or other steroid precursors according to individual laboratory reference standards.

Note: Where there is borderline elevation of 17-OHP in sick or preterm neonates, a measurement repeated at a later time to confirm the diagnosis should be sought. Cases identified by newborn screening will always require confirmation of diagnosis by standard biochemical mehtods.

REPORTING INSTRUCTIONS:

First appearance on APSU card: Please report all suspected or newly diagnosed cases of CAH of any type seen since May 1 1995.

Subsequent months: Please report all suspected or newly diagnosed cases of CAH of any type seen during the last month.

REFERENCES:

1. White PC, New MI, Dupont B. Congenital adrenal hyperplasia (2). *N Engl J Med* 1987; 316:1580-1586.
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3. Pang SY, Wallace MA, Hofman L, Thuline HC, Dorche C, Lyon IC *et al*. Worldwide experience in newborn screeening for classical congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *Pediatrics* 1988; 81:866-874.
4. Pang S, Clark A. Congenital adrenal hyperplasia due to 21-hydroxylase deficiency: newborn screening and its relationship to the diagnosis and treatment of the disorder. *Screening* 1993; 2:105-139.