Australian Paediatric Surveillance Unit

PROTOCOL SHEET – THE CHARGE ASSOCIATION (January 2000)

Background to study
Children with the CHARGE association have variable combinations of abnormalities including coloboma, choanal atresia and cardiac, renal, genital and gastrointestinal abnormalities. The true incidence of CHARGE association is not known as no systematic study has been performed. Incidence estimates range from 0.1-1.2/100,000 live births. Diagnosis of this condition has been hindered by inconsistencies in diagnostic criteria. Diagnosis may also be delayed, as major features of the disorder may not be readily identifiable, particularly in young children. Adding to the confusion, the CHARGE association shares many of its features with other genetic and birth defect syndromes.

Objectives:
1. What is the incidence of the CHARGE association in Australia?
2. What is the age of diagnosis and the clinical features at presentation?
3. Are children with CHARGE developmentally normal for age?
4. What is the morbidity and mortality of CHARGE?

CASE DEFINITION AND REPORTING INSTRUCTIONS
Any child less than 15 years with newly diagnosed CHARGE association
Children should be reported if they have:
- ***three or more major criteria alone*** or
- ***one or more major criteria with at least two minor criteria (see Table).***

Major criterion	Includes
Coloboma	Iris, retina or choroid
Choanal atresia	Unilateral/bilateral membranous/bony stenosis/atresia
Characteristic ear abnormalities	External ear (lop or cup shaped); middle ear (ossicular malformations, chronic serous otitis); mixed deafness; cochlear defects
Cranial nerve dysfunction	Anosmia; Facial palsy (unilateral or bilateral); sensorineural deafness and vestibular problems; swallowing problems
Minor criterion	**Includes**
Genital hypoplasia	Males: Micropenis, cryptorchidism; Females: Hypoplastic labia Both: Delayed, incomplete pubertal development
Developmental delay	Delayed motor milestones, hypotonia, mental retardation
Cardiovascular malformations	All types: especially conotruncal defects (eg. Tetraology of Fallot); Atrio-ventricular canal defects and aortic arch anomalies
Orofacial clefts	Cleft lip and/or palate
Tracheo-oesophageal fistula	Tracheo-oesophageal defects of all types
Distinctive face	Unilateral facial weakness/asymmetry, broad forehead, flat malar region, thin lips

Follow-up of positive returns
A questionnaire requesting further details and an information pack will be forwarded to clinicians who report a case of CHARGE association. **A copy of the questionnaire is enclosed for your information.**

If you have any questions please contact:
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