National Screening Committee

Child Health Sub-Group Report on Congenital Cataract

May 2005

Congenital cataract

Testing for congenital cataract, using the red reflex, forms part of the general physical examination at birth and at 6-8 weeks. Observation of the general appearance of the eyes and any abnormal eye movements, although not screening procedures, should form part of the examination.

The Condition

1. **The condition should be an important health problem. YES**
Congenital cataract is rare, but the consequences in terms of visual loss are profound.

2. The epidemiology and natural history of the condition, including development from latent to declared disease, should be adequately understood and there should be a detectable risk factor, disease marker, latent period or early symptomatic stage. YES
About 2-3 cases per 10,000 births of which one third or less are unilateral. About one third are isolated, two thirds have other eye or systemic anomalies. Not all are present at birth and not all are dense or cause major vision loss. Treatment in first few weeks of life highly desirable for unilateral – slightly more margin for bilateral but treatment before 6-8 weeks desirable for optimum outcome. With early surgery – good functional vision. Without – blindness.

3. All the cost-effective primary prevention interventions should have been implemented as far as practicable. YES
There is a national rubella immunisation programme (given as MMR). Genetic advice is offered for familial and syndromic cases in previous pregnancies.

4 If the carriers of a mutation are identified as a result of screening the natural history of people with this status should be understood, including the psychological implications. N/A

The Test

5 **There should be a simple, safe, precise and validated screening test. YES**
Inspection of the eyes with an ophthalmoscope to see the red reflex will detect cataract. It appears easy but may not be so in the first few days of life as the infant's eyelids may be swollen, the eyes may be closed or the infant may be fractious. Examiners are often poorly trained and as the condition is so uncommon, it is difficult to maintain skills.

6 The distribution of test values in the target population should be known and a suitable cut-off level defined and agreed. YES
Cataract is either seen or not seen. It is difficult to validate because the condition is rare and not easily simulated.

7 The test should be acceptable to the population. YES

8 There should be an agreed policy on the further diagnostic investigation of individuals with a positive test result and on the choices available to those individuals. YES
Urgent referral to an ophthalmologist, preferably a paediatric specialist.

9 If the test is for mutations the criteria used to select the subset of mutations to be covered by screening, if all possible mutations are not being tested, should be clearly set out. N/A

The Treatment

10 There should be an effective treatment or intervention for patients identified through early detection, with evidence of early treatment leading to better outcomes than late treatment. YES
Early surgery leads to a good outcome.

11 There should be agreed evidence based policies covering which individuals should be offered treatment and the appropriate treatment to be offered. YES
Surgery should be offered.

12 Clinical management of the condition and patient outcomes should be optimised in all health care providers prior to participation in a screening programme. YES

The Screening programme

13 There should be evidence from high quality Randomised Controlled Trials that the screening programme is effective in reducing mortality or morbidity. NO
No formal trials have taken place, but there is clinical evidence that the age of referral has fallen steadily over recent years, though it is still not earlier enough.

Where screening is aimed solely at providing information to allow the person being screened to make an "informed choice" (eg. Down's syndrome, cystic fibrosis carrier screening), there must be evidence from high quality trials that the test accurately measures risk. The information that is provided about the test and its outcome must be of value and readily understood by the individual being screened. N/A

14 There should be evidence that the complete screening programme (test, diagnostic procedures, treatment/ intervention) is clinically, socially and ethically acceptable to health professionals and the public. YES

15 The benefit from the screening programme should outweigh the physical and psychological harm (caused by the test, diagnostic procedures and treatment). YES

16. The opportunity cost of the screening programme (including testing, diagnosis and treatment, administration, training and quality assurance)) should be economically balanced in relation to expenditure on medical care as a whole (ie. value for money). YES

17. There should be a plan for managing and monitoring the screening programme and an agreed set of quality assurance standards. NO
This is under consideration.

18. Adequate staffing and facilities for testing, diagnosis, treatment and programme management should be available prior to the commencement of the screening programme. YES

19 All other options for managing the condition should have been considered (e.g. improving treatment, providing other services), to ensure that no more cost effective intervention could be introduced or current interventions increased within the resources available. N/A

20 Evidence-based information, explaining the consequences of testing, investigation and treatment, should be made available to potential participants to assist them in making an informed choice. YES.
This will be made available in the form of a leaflet describing the neonatal and six to eight week physical examinations.

21. Public pressure for widening the eligibility criteria for reducing the screening interval, and for increasing the sensitivity of the testing process, should be anticipated. Decisions about these parameters should be scientifically justifiable to the public. N/A

22 If screening is for a mutation the programme should be acceptable to people identified as carriers and to other family members. N/A.

The complete physical examination is under review in terms of quality standards, training and information materials.