Survey of scope of neonatal screening in the United Kingdom

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The national neonatal screening programme for phenylketonuria was established in 1969, with screening for congenital hypothyroidism being added in 1981. The aim of the programme is to reduce morbidity by complete and timely detection and treatment of affected cases. Scientific developments mean that many other disorders can be screened for.[1] The extent of screening in the United Kingdom is uncertain and not all proposed screening programmes meet, or have been formally assessed against, the criteria of suitability.[2] Also, the loss of regional responsibility for overview of programmes may result in district based decisions about the purchase of screening programmes in the future.

The main aim of this survey was to identify the nature and extent of screening for disorders other than phenylketonuria and congenital hypothyroidism by the national programme.

Methods and results

In May 1993 an anonymous questionnaire was sent to all 28 of the NHS neonatal laboratory directors in the United Kingdom identified by the National Committee of Directors of Neonatal Screening Laboratories. The questionnaire asked about tests performed, reporting of positive and negative results, participation in long term follow up of positive cases of phenylketonuria and congenital hypothyroidism, and knowledge of organisational arrangements for monitoring the screening programme. Estimates of coverage of non-national screening programmes identified by the survey were made by dividing the total number of infants reported tested by laboratories in a year by the reported number of live births in 1991 (793 000). For phenylketonuria and congenital hypothyroidism it was assumed that coverage was almost complete (99%).[3]

Twenty six laboratories responded. Of these, 22 screened for both phenylketonuria and congenital hypothyroidism, two for congenital hypothyroidism only, one for phenylketonuria only, and one for neither of these disorders. These laboratories cover over 90% of specimens tested for both phenylketonuria and congenital hypothyroidism. The number of infants tested annually by laboratories ranged from 2500 to 107 000. The number of districts, boards, or trust areas covered by a laboratory ranged from one to 29 with a median of nine. Eighteen laboratories processed specimen cards from outside the NHS, including those from independent midwives attending home births (16 laboratories), private institutions (12), prisons (three), and other sources (four).

The table shows the range of tests performed on named infants, and estimated numbers and proportions of infants tested annually for each disorder. A range of disorders in addition to phenylketonuria and congenital hypothyroidism is screened for in parts of the United Kingdom. Several laboratories detect disorders, such as galactosaemia and tyrosinaemia, as an incidental finding though they are not specifically screening for these disorders.[4] Several pilot or research programmes were identified. Anonymous surveillance of HIV infection was also performed on neonatal specimens by seven laboratories, usually by the microbiological laboratories of the Public Health Laboratory Service covering nine (old) regions.

Disorders tested for in neonatal blood specimens in United Kingdom

Disorder	Estimated No of infants screened annually	Estimated proportion of infants screened (%)	No of laboratories testing specimens	Type of scheme
Congenital hypothyroidism	793 000	99	25	All established
Phenylketonuria	793 000	99	25	All established
Amino acid disorders:				All established
Specific testing:				
Homocystinuria	95 000	12	2	
Tyrosinaemia	25 000	3	1	
On chromatography*	268 000	34	9	All established
Galactosaemia:				
Specific testing	70 000	9	1	Established
Incidental finding	67 000	8	1	Established
Haemoglobinopathies	71 000	9	3	Two established, one pilot
Cystic fibrosis	130 000	16	4	All established
Duchene's muscular dystrophy	20 000	3	1	Pilot
Familial hyperlipidaemia	4000	1	1	Pilot

*Incidential finding; includes tyrosinaemia, maple syrup disease, and histidinaemia.

Comment

This survey has shown that several disorders in addition to phenylketonuria and hypothyroidism are screened for using neonatal blood specimens, and our list may be incomplete. Only the laboratories identified by the National Committee of Directors of Neonatal Screening Laboratories were included in the survey, but those surveyed reported additional screening—for example, pilot projects—by other laboratories. Selective screening programmes for sickle cell disease based on cord blood specimens and undertaken by haematology laboratories were not identified. The survey identified a wider range of conditions screened for than reported elsewhere, though this is fewer than in the United States. Local skills or interests have probably influenced the range of conditions screened for.

In conclusion, the neonatal screening programme has been expanded locally in several places to test for disorders other than those on the national scheme. Not all these programmes meet or have been formally assessed against criteria of suitability of a screening programme.[2] In view of potential expansion of the programme, decisions on implementing new screening programmes must be based on reviews of effectiveness and cost effectiveness.[5] The impact that screening for additional disorders may have on the existing screening programme should be included in this process to ensure that the overall programme is both effective and efficient.

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