The role of the UK National Screening Committee in the development and implementation of newborn genetic screening programmes

David Elliman

Newborn Genetic Screening – What do we mean?

- Analysing DNA for clinically significant mutations
- May be prime method of screening or part of diagnostic process
- NB – much of newborn screening has a 'genetic' element even though testing the phenotype – PKU and, in part, Congenital Hypothyroidism ...

Cette présentation a été effectuée le 17 novembre 2005, au cours de la journée « Dépistage populationnel en génétique : développement, implantation et évaluation » dans le cadre des Journées annuelles de santé publique (JASP) 2005. L'ensemble des présentations est disponible sur le site des JASP, à l'adresse http://www.inspq.qc.ca/jasp/archives/.

Newborn Genetic Screening – Why is it different?

- Not testing phenotype, so less predictable
- Will pick up carriers
- May throw up non-paternity
- Could disclose conditions for which no management available

Relevant UK Government Advisory Committees

- National Screening Committee
  - Advises the Ministers of Health on whether a screening programme should be started, continued (perhaps with modifications) or stopped.

National Screening Committee

- Previously drift, lack of control, variable quality assurance and audit, and lack of equity ('postcode screening')
- NSC set up in 1996
- Wide representation, including 'patients' and members from all 4 nations
- Child Health and Antenatal Subgroups set up in 1998
- Fetal, Maternal and Child Health Group brought together in 2004

Relevant UK Government Advisory Committees

- National Screening Committee
  - Advises the Ministers of Health on whether a screening programme should be started, continued (perhaps with modifications) or stopped.
- Human Genetics Commission
  - Advises on the ethical, legal, social and economic aspects of developments in human genetics as well as their effects on health and healthcare.

Approval of Screening Programmes - 1

- Raised as an issue
- Is there a possible case for screening?

Approval of Screening Programmes: the Criteria

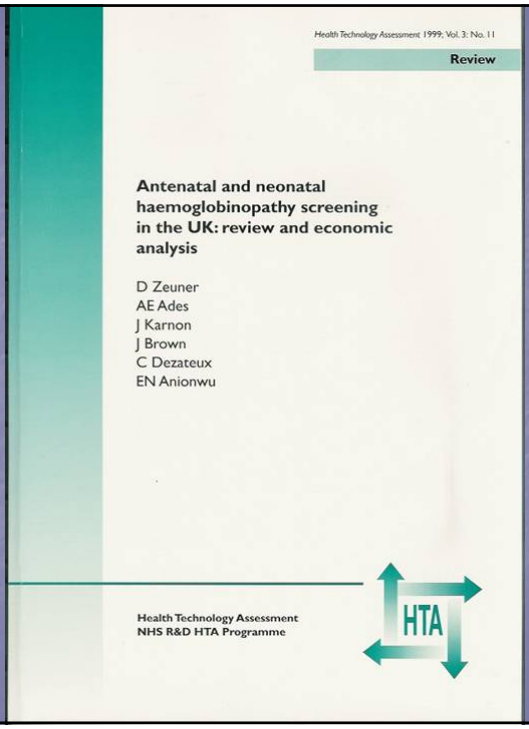
- The condition
- The test
- The treatment/management
- The programme

Additional criteria for *genetic* screening programmes

- 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.
- 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.
- If screening is for a mutation, the programme should be acceptable to people identified as carriers and to other family members.

Approval of Screening Programmes -2

- Raised as an issue
- Is there a possible case for screening?
- All the available data are gathered, ideally including a systematic review which may be commissioned especially for the purpose by HTA.



Antenatal and neonatal haemoglobinopathy screening in UK: review and economic analysis.

HTA 1999

Approval of Screening Programmes - 3

- Raised as an issue
- Is there a possible case for screening?
- All the available data are gathered, ideally including a systematic review - may be commissioned especially for the purpose by HTA.
- Specially convened meeting(s) to discuss evidence

Composition of meeting to consider a screening programme

Groups represented:

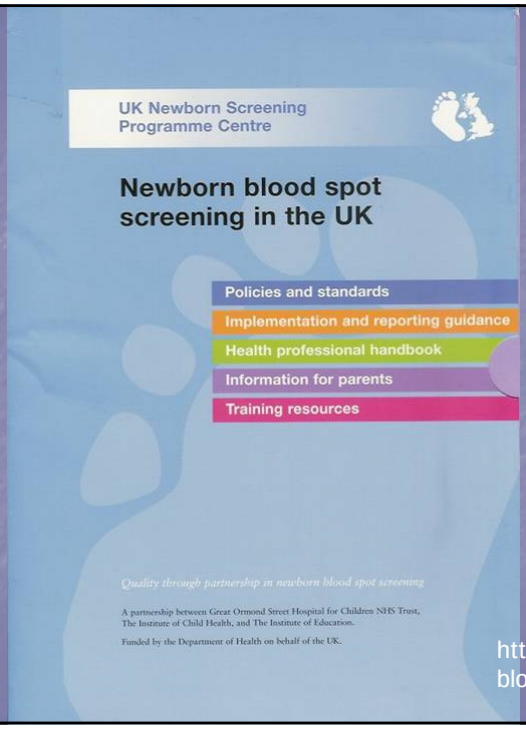
- Clinicians involved in treatment
- Clinicians involved in the screening test
- Public health/commissioners
- Epidemiology
- Health economics
- Lay groups
- Four Departments of Health

Approval of Screening Programmes - 4

- Raised as an issue
- Is there a possible case for screening?
- All the available data are gathered, ideally including a systematic review - may be commissioned especially for the purpose by HTA.
- Specially convened meeting to discuss evidence
- CHSG, now FMCHG, considers the evidence and the practicalities
- NSC recommends to …….. Minister
- Minister makes a decision

Development and Implementation of Screening Programmes

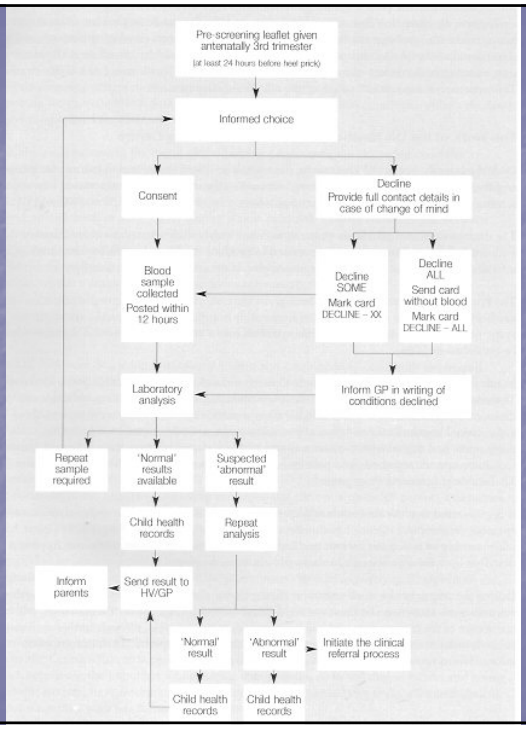
- Detailed review of the resources needed, including diagnostic and treatment facilities.
- Provision of funding
- Decision re timing of introduction – 'big bang' v gradual roll-out or pilots
- Provision of materials for
  - Professionals
  - Parents



Resource Pack for Healthcare Professionals involved in Newborn Blood Spot Screening

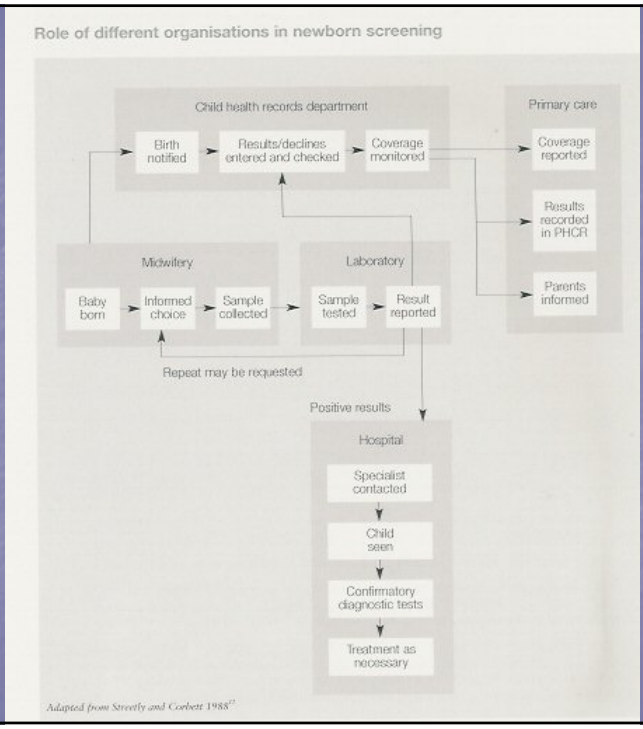
April 2005

http://www.newbornscreening-bloodspot.org.uk/



From 'Policies and Standards'

April 2005



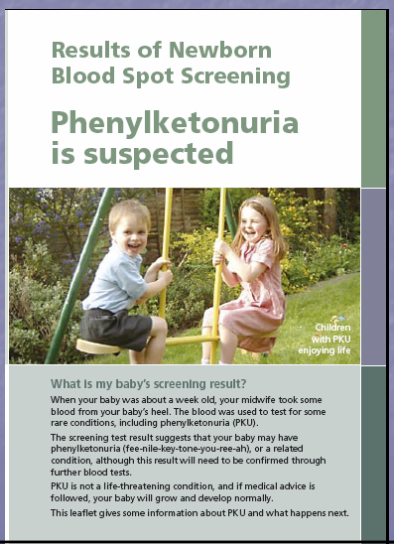
From 'Policies and Standards'

April 2005

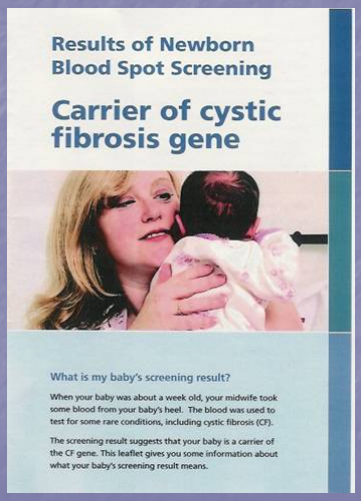
Screening leaflets for parents - 1

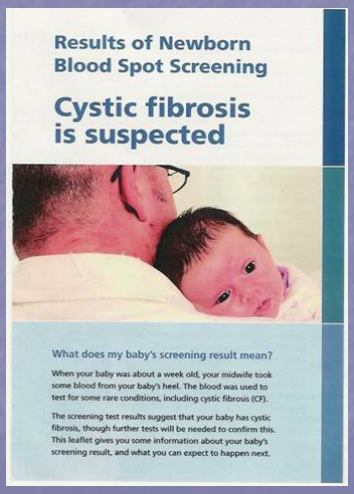
"New born blood spot screening for your baby" Also available in: French, Portugese, Somali, Turkish, Bengali, Arabic, Urdu, Greek, Gujarati and Romoanian.

Screening leaflets for parents - 2



Screening leaflets for parents - 3



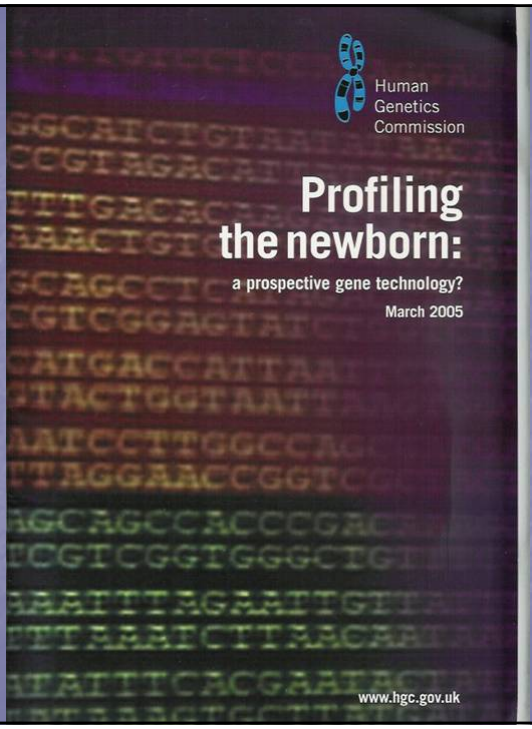


Neonatal Blood Spot Screening – Process Standards

- Timely sample collection
- Timely sample despatch
- Completeness of coverage
- Enhanced tracking abilities
- Timely identification of babies for whom the laboratory has not received a decline notification or a blood sample
- Timely processing of positive screening samples

Neonatal Blood Spot Screening – Other Policies/Guidance

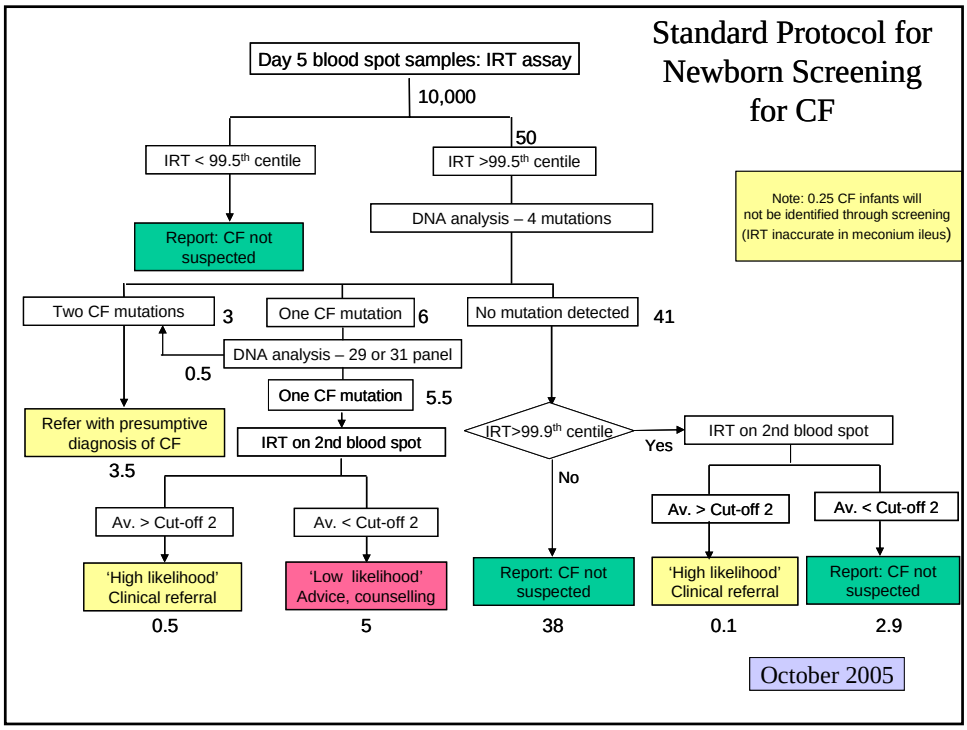
- Consent and communication
- Blood sampling guidelines
- Initial clinical referral standards
- Code of practice for the retention and storage of residual spots



Profiling the newborn: a prospective gene technology?

A report from a Joint Working Group of the Human Genetics Commission and the UK National Screening Committee

March 2005


Standard Protocol for Newborn Screening for CF
Day 5 blood spot samples: IRT assay
10,000
50
IRT < 99.5th centile
IRT >99.5th centile
Report: CF not suspected
DNA analysis – 4 mutations
Note: 0.25 CF infants will not be identified through screening (IRT inaccurate in meconium ileus)
Two CF mutations
3
One CF mutation
6
No mutation detected
41
DNA analysis – 29 or 31 panel
0.5
One CF mutation
5.5
Refer with presumptive diagnosis of CF
3.5
IRT on 2nd blood spot
IRT>99.9th centile
Yes
No
IRT on 2nd blood spot
Av. > Cut-off 2
Av. < Cut-off 2
Av. > Cut-off 2
Av. < Cut-off 2
'High likelihood' Clinical referral
0.5
'Low likelihood' Advice, counselling
5
Report: CF not suspected
38
'High likelihood' Clinical referral
0.1
Report: CF not suspected
2.9
October 2005