UK centres are not following the Royal College of Pathologists’ recommendations for storage of Guthrie cards: a national policy is needed

Stored neonatal blood spots are a valuable source of DNA for retrospective diagnosis. A recent working party of the Royal College of Pathologists recommended storage of neonatal screening test (Guthrie) cards for at least 20 years provided that no deterioration of the sample has occurred. Our recent attempts to trace such cards convince us that a UK national policy and central funding for storage of such cards is necessary.

Mitochondrial encephalopathy with stroke-like episodes (MELAS) is frequently associated with a mitochondrial DNA point mutation A3243G. Segregation and proliferation of this mutation within different tissues is not well understood. Levels of the 3243 mutant in blood are usually lower than in muscle and cross sectional data suggest that the level of mutant in blood may fall with increasing age. There is concern that cases of MELAS may be missed if diagnosed using the polymerase chain reaction (PCR) on blood. Longitudinal studies are necessary to clarify this issue. We have attempted to evaluate this aspect of the test using Guthrie cards to compare levels of the 3243 mutant mtDNA in blood at birth and at diagnosis, under the auspices of the European Neuro muscular Centre.

We have collected Guthrie cards from patients with MELAS born in the UK after 1970 and, with the informed consent of the patients or parents or both as appropriate, sought their Guthrie cards for comparison with the stored birth load at birth. It has been possible to locate only four cards, and one of these has been autolysed (this destroys DNA). Hence, the failure rate was 87%, despite cooperation from clinicians and patients.

A telephonic survey of the 25 UK neonatal screening laboratories showed marked variability in practice between regions. Two centres use serum for screening rather than blood spots, and one stores blood spots for five months only. Only 12 centres store cards for longer than 10 years, and the trend is towards shorter periods because of financial pressures.

We suggest that a national policy for storage of neonatal blood spots is needed, and this may require central funding. Centralisation of cards and records would be an advantage as families now move between regions frequently and do not always recall their previous addresses. Cards should be stored in such a way as to prevent cross contamination, particularly if PCR is to be used in analysis of the blood spot.