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.1 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.2 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 in large kindreds is not well understood. Levels of the 3243 mutant mtDNA in blood are usually lower than in muscle3 and cross sectional data suggest that the level of mutant mtDNA may fall with increasing age.4 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 now carried out a European collaborative study using Guthrie cards to compare levels of the 3243 mutant mtDNA in blood at birth and diagnosis, under the auspices of the European Neuromuscular Centre.

We selected 25 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. A European collaborative study using Guthrie cards to compare levels of the 3243 mutant mtDNA in blood at birth and diagnosis, under the auspices of the European Neuromuscular Centre.

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