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family. First, it allowed the diagnosis of SMA to be confirmed, since the affected child carried a homozygous deletion of SMN exons 7 and 8, as previously reported in more than 90% of SMA patients.[4 5 7 8 10 11] Second, the direct analysis of the SMN gene and its flanking markers C212–C272 provided strong evidence for a de novo deletion of maternal origin and rejected the hypothesis of genetic heterogeneity since the affected and unaffected children carried different genotypes at the SMN locus. The risk for the mother of bearing another affected child should therefore be considered to be very low. Indeed, analysis of 41 families with two affected children did not provide any evidence for germinal mosaicism using microsatellite DNA markers, suggesting that this situation is very rare (data not shown, available on request). For this reason, the combined analysis of SMN and its flanking markers C212 and C272 are still of interest for detecting de novo deletions, estimating their actual frequency, and delivering accurate prenatal prediction in SMA.

In conclusion, the use of SMN gene testing has important implications for genetic counselling in SMA families. First, it allows the diagnosis of SMA to be confirmed when a proband harbours a homozygous SMN gene deletion. In contrast, the absence of a homozygous deletion may suggest either a misdiagnosis or the presence of intragenic mutations in the SMN gene, as previously reported.[7 8] A clinical re-evaluation of the proband and the search for intragenic mutations by screening of each SMN exon should be undertaken before making prenatal diagnosis feasible.[12] Second, the use of SMN and the closest genetic markers as probes enables detection of de novo deletions in the probands. Establishing the status of the parents at the SMN locus should be helpful in estimating the actual frequency of de novo deletions at this locus. Finally, several reports have estimated the frequency of homozygous SMN deletions in asymptomatic carriers to be below 1% (0.8%).[5 13 14] Moreover, these observations have been reported in the mildest form of the disease only. Consequently, fetuses harbouring homozygous SMN gene deletions should be predicted to be affected in families with severely affected probands.

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NOTICES

Standing Committee on Human Cytogenetic Nomenclature 1996–2001

Elections for the Standing Committee on Human Cytogenetic Nomenclature were held at the 9th International Congress of Human Genetics in Rio de Janeiro, Brazil, on 21 August 1996. The following members were elected for the period 1996–2001: Patricia A Jacobs, (UK) (Chairman), Uta Francke (USA), David H Ledbetter (USA), Norio Niikawa (Japan), Avirachan T Tharapel (USA), Niels Tommerup (Denmark), Angela M Vianna-Morgante (Brazil). Issues regarding human cytogenetic nomenclature can be addressed to any member of the committee.

Second European Forum on Quality Improvement in Health Care

The Second European Forum on Quality Improvement in Health Care will be held in Paris, France, on 24–26 April 1997. The forum will consist of one day teaching courses, invited presentations, posters, and presentations selected from submissions, and a scientific session. For more information contact: BMA, Conference Unit, PO Box 295, London WC1H 9TE, UK. Tel: +44 (0) 171 383 6478. Fax: +44 (0) 171 383 6869.

Molecular Advances in Cancer Epidemiology and Prevention

This postgraduate course will be held at the Sheraton Palace Hotel, San Francisco, California, USA on 20–22 February 1997. Category 1 CME credit. For more information, please contact: University of California, Office of Continuing Medical Education, 1855 Folsom Street, MCB–630, Box 0742, San Francisco, CA 94143–0742, USA. Tel: (415) 476–4251. Fax: (415) 476–0318. WWW: http://cme.ucsf.edu.

Correction

In the November 1996 issue of the journal, on page 940, the current affiliation of Dr F M Pope as MRC Connective Tissue Genetics Group, Strangeways Research Laboratory, Worts Causeway, Cambridge CB1 4RN, was inadvertently omitted.

Notice to Contributors

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Chromosomes: *ISCN 1995. An International System for Human Cytogenetic Nomenclature*, Mitelman F (ed); S Karger, Basel, 1995
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Enzymes: Enzyme nomenclature: recommendations of the nomenclature committee of the International Union of Biochemistry. New York: Academic Press, 1992. Information also available on URL http://expasy.hcuge.ch/sprot/enzyme.html

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