variable expression in AR disorders is perhaps less rare than assumed.

(6) Interestingly, de novo AR gene mutations are not mentioned as a possible cause for reduced segregation ratios in AR disorders.\(^2\) As new AR mutations usually have no phenotypic effect, we therefore know very little about mutation rates in this category, but it is of no reason to assume that they rarer than de novo autosomal dominant or X linked mutations. The evolving plethora of mutations in the CFTR gene would appear to go against this idea. However, as no proven new mutations have been found among more than 700 families analysed in the Netherlands (H Hailey, Rotterdam, and H Scheffer, Groningen, personal communications). From the Finnish CHH families, only two out of 83 couples were consanguineous\(^2\) and here the mutated alleles are very likely to be identical by descent, although DNA analysis sometimes proves there are exceptions to this rule.\(^3\) Twenty families could be linked genealogically and here the unlinked mutation could be recurrent, or even perhaps new, in the other 61 CHH families. Only when the CHH gene, recently assigned to chromosome 3,\(^4\) is analysed for mutations will the point be clarified, either by now closing in on the gene of the mutation in the father (with proven paternity) or absence of the mutation in the mother (apparent 'non-maternity'), when isodosomy has been excluded.

(7) Gonadal mosaicism for an AR mutation in one of the parents could theoretically be yet another cause of low segregation ratios in AR phenotypes. However, this will be difficult to distinguish from the above mentioned new AR mutation.

(8) Finally, unstable mutations, as in fragile X syndrome, myotonic dystrophy, Huntington's disease,\(^5\) could play a role in AR disorders. Since damage of gene function only occurs in a certain percentage of gametes in which the gene is segregating, the number of expected affected offspring will be reduced.

In conclusion there are a number of possible explanations for a segregation ratio lower than 0.25, which endorses the conclusion of Bundey and Young.\(^6\) Until we know more about the relative importance of all these factors in several AR disorders, we will have to counsel with caution.

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Del(18p) syndrome with a single central maxillary incisor

In 1991 Aughton et al\(^7\) reported a case of del(18p) syndrome and a single central maxillary incisor (SMCI), stating that it was the second report of a patient with this association. They considered that the only former report was that of Dolan et al\(^8\); however Bouden et al\(^9\) had already described a similar case in 1983, which deserves attention.

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Prevention of Mediterranean anaemia in Latium, Italy, today

In four previous papers published in this Journal\(^1\) we reported the continuing programme for the prevention of Mediterranean anaemia (thalassaemia major) in Latium, a region of central Italy. This work, supported by the Regional Health Authorities of Latium, has been carried out by the Rome Microcythemia Institute since October 1975. It consists of the following.

(1) An educational programme among school children in Latium that consists of two steps. The first is detailed information provided class by class with brief lectures and printed and audiovisual material. The second is screening of informed students who have obtained consent to be examined from their parents.

(2) Examination of thalassaemias' students' families and identification of carriers of the thalassaemia trait.

(3) The provision of information and screening campaigns to young adult school leavers. In this last stage the information is imparted at meetings at the Family Health Services, in explanatory pamphlets at the marital Registry Offices in the towns and villages of the region, by information from the Public Health Offices and family doctors or gynaecologists, and recurrent use of the media.

Screening of school leavers evolved in 1978 through the effect of the school screening and has progressively increased. It is carried out in the outpatients department of the Institute and in the Family Health Services of the region.

The results of this continuing work are shown in the table. Young carriers of non-\(\alpha\) thalassaemia comprise about 80% of all carriers in Latium (44,000 in a region with 2,000,000 inhabitants of childbearing age and an incidence of non-\(\alpha\) thalassaemia of 0.02%).\(^2\)

The couples of childbearing age at risk identified (369) comprise 74% of the total (about 5000) in Latium. All these couples are kept under surveillance by our genetic counselling service. In the last 11 years 69 homozygous fetuses have been aborted after prenatal diagnosis. The incidence of newborns affected by Mediterranean anaemia has decreased from 16-04 of 100,000 live births in 1975-76 to 0.71 in 1989-90. In 1991 and 1992 no affected children have been born in Latium.

This programme offers to young thalassae- mic couples the advantage of choosing either postconceptual or preconceptual means of prevention.

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