Correspondence

Journal of Medical Genetics, 1982, **19**, 390-394

Interrelationship of different dysraphic malformations and consequences for genetic counselling

SIR,

A girl in our paediatric care suffers from meningo-myelocele (lumbosacral) with peculiar additional features and an interesting family history. The pedigree suggested to us the possibility of a genetic relationship between different types of dysraphism (figure).

FIGURE Family pedigree.

Our patient, apart from her spina bifida, has a very narrow arched palate impeding proper speech. Her articulation is almost as bad as if she had full cleft palate. Her otherwise healthy mother has the same anatomical and functional abnormality. The mother's brother had a defect of the abdominal wall and died in the neonatal period.

We wondered whether abdominal defects (omphalocele, gastroschisis) are relevant to familial disposition towards other closure defects (old term: status dysraphicus)?

We have not found clear indications of a causal relationship between failed closure of the neural tube and that of the ventral body wall.¹ In one interesting family, however, three such defects were present together in one patient, while his three sibs had exomphalos only (one of them also had an arched palate and mandibular hypoplasia).²

The distribution of dysraphic features in our family, too, raises the possibility of a common pathogenesis (be it genetic or environmental or both) of these malformations. If so the appearance of one of them would warrant measures to prevent, or to diagnose antenatally, the others. This conclusion is analogous to that drawn by Cohen and colleagues³

regarding preceding cases of hydrocephalus. Their argument was, of course, supported by a large number of relevant observations. I should be grateful for information on similar occurrences to the one I present in this letter.

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'Microcytogenetics' and Langer-Giedion syndrome

SIR,

The identification of minute cytogenetic aberrations in a few disorders of proved or suspected Mendelian origin is causing growing interest, and the term 'microcytogenetics' has been proposed by de Grouchy¹ to denote this new field of research. Evidence is now available of associations such as aniridia-Wilms' tumour and 11p13 deletion, retinoblastoma and 13q14 deletion, Prader-Willi syndrome and chromosome 15, and X linked mental retardation and the Xq27→28 fragile site.

Recently Pfeiffer² and Bühler *et al*³ have suggested a new association between Langer-Giedion (LG) syndrome and small deletions of the long arm of chromosome 8. They have reported two patients, with del(8)(q13→q22) and del(8)(q24) respectively, showing the characteristic features of LG syndrome, including mental retardation, short stature, microcephaly, peculiar facies, cone-shaped epiphyses of the phalangeal and metacarpal bones, and multiple cartilaginous exostoses (MCE). Further evidence for a similar association has been provided by Fryns *et al*⁴ in a subject with a deletion of band 8q21. The patient displayed dysmorphic features reminiscent of LG syndrome and MCE, which were detected at 4 years, but were apparently absent at 15 months, when first examined.⁵

According to Bühler *et al*³ an association between