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Pericentric inversion and sterility

SIR,

In January 1986 we published the first familial case of pericentric inversion of chromosome 1 involving the whole of the short arm and associated with sterility in two brothers.¹

The inherited transmission of this chromosomal anomaly was shown by the cytogenetic study of the proband's mother who is a carrier of the inversion. Later, another brother of those reported contacted our department, worried about his possible infertility. The karyotype showed the same pericentric inversion, 46,XY,inv(1)(p36-3q12), as in his brothers and he had severe oligozoospermia (about 200 000 per ml).

Therefore, we have three sterile men with the same maternally transmitted chromosome abnormality. This may be interpreted as further evidence of the susceptibility of spermatogenesis to structural chromosome rearrangements.

ALBERTO BARROS*†, M CARMO TAVARES*,
M PAULA GOMES*, AND M PURIFICAÇÃO TAVARES*

*Department of Medical Genetics, Faculty of
Medicine, University of Oporto;
and †Department of Obstetrics, S João Hospital,
Oporto, Portugal.

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The craniocardioskeletal syndrome and the Noonan-like short stature syndrome are possibly the same entity

SIR,

Baraitser and Patton¹ recently described a Noonan-like short stature syndrome (possibly new) in four children (three female, one male), presenting with sparse hair, mildly slow development, posteriorly rotated ears, short nose, low hair line, and shield shaped chest. Other features included: heart murmur and prominent philtrum (cases 1 and 2), hypertrophic cardiomyopathy (case 4), pectus excavatum (case 1), and increased head circumference with moderate hydrocephalus, low set ears, and narrowing of the interpedicular distances between L1 and L5 (case 3). Many of these features are similar to those present in a syndrome described by our group,^{2,3} including short stature, delayed psychomotor development, scanty hair coarse face (cases 2 and 4 of Baraitser and Patton are remarkably similar to cases C and D of our report), flattened nasal bridge, short nose with antverted nostrils, long philtrum, low set, posteriorly rotated ears, short and wide thorax, cardiac murmur, cubitus valgus, and delayed bone age.

There are, however, some discordant features present in our cases, such as mild exophthalmos, cutis laxa, and wrinkled palms and soles (washed woman's hands).

We think that it is useful to compare the clinical picture as well as the radiographical data in order to obtain the best delineation of the syndrome, which we have assumed to be an autosomal dominant disorder, mainly because of the advanced parental age in our cases. Recently, McKusick⁴ catalogued this syndrome as a separate entity (entry 11462).

JOSÉ SÁNCHEZ CORONA AND JOSÉ MARÍA CANTÚ
División de Genética
Unidad de Investigación Biomédica
Centro Médico de Occidente
Instituto Mexicano del Seguro Social
Apartado Postal 1-3838
Guadalajara, Jalisco, Mexico

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