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

SIR.

Baraitser and Patton recently described 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 interpapillary distances between L1 and L5 (case 3). Many of these features are similar to those present in a syndrome described by our group, including short stature, delayed psychomotor development, scanty hair, coarse features, and 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.

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Reference