Iris coloboma, ptosis, hypertelorism, and mental retardation

SIR,

Drs Baraitser and Winter reported three patients with a new syndrome consisting of iris coloboma, ptosis, hypertelorism, and mental retardation (*J Med Genet* 1988;25:41-3). They correctly state that in Noonan's syndrome no report of iris colobomas has been published. They partially used this fact to exclude the diagnosis of Noonan's syndrome in their patients, although they felt the facial gestalt also differed.

I have seen a sporadic case of a patient with Noonan's syndrome (figure) who had a left iris coloboma without retinal abnormalities. He was short and had mild psychomotor delay. He also had a dysplastic pulmonary valve, pectus excavatum, low set, thick ears, and malar flattening. I saw this

patient in consultation with Dr Jacqueline Noonan, who also agreed with the diagnosis.

The iris coloboma may well be unrelated to the Noonan's syndrome in the above patient: however, it can no longer be stated that colobomas have not been reported in a patient with Noonan's syndrome.

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Teratogenicity of ergotamine

SIR,

Hughes and Goldstein¹ hypothesised on the basis of one case that ergotamine, acting either alone or in synergy with propranolol and caffeine, produced fetal vasoconstriction resulting in tissue ischaemia and subsequent malformation. This case report prompted us to check the database of the Hungarian Case-Control Surveillance of Congenital Anomalies, 1980-1986.² This system has both prospective (prenatal care logbook) and retrospective (questionnaire) sources of data.

Within the group of mothers who delivered matched healthy newborns (the so called negative controls), 0.11% were treated with ergotamine during pregnancy (table 1). Additionally, 0.32% of mothers who delivered babies with Down's syndrome (the so called positive controls) used this drug. (However, this figure relates to only one pregnant woman.) The rate of ergotamine use was 0.14% in the total sample of malformed index patients ($p > 0.10$). Of these 13 malformed babies, four had neural tube defects. Three mothers in the latter group were treated with ergotamine during the first trimester of pregnancy. Other groups of congenital anomalies did not show any significant increase of ergotamine treatment during pregnancy. These cases are available for further investigation concerning the occurrence of minor brain abnormalities including arrested cerebral maturation and paraplegia. After matching, the numbers of index patients and negative matched controls were 10:9 in the total samples but 4:0 in the neural tube defects cases (McNemar test: $\chi^2 = 9.00$, $p < 0.01$). The latter finding needs further study.

FIGURE Patient with Noonan's syndrome. Note left iris coloboma.