Correspondence

the 9p trisomy, 9q trisomy, and 9p monosomy conditions. However, few cases have been described to delineate the trisomy 9 syndrome. In fact, non-mosaic trisomy 9 has rarely been reported in term infants\(^{1-3}\) and only once by prenatal diagnosis.\(^{4}\) I would like to elucidate further the anatomical dysmorphology and variability of this syndrome by reporting the prenatal diagnosis and subsequent anatomopathological examination of a trisomy 9 fetus.

Although advanced parental age has not been a hallmark of this syndrome, the ages of the mother and father of our case were 45 years and 41 years, respectively. There was no history of consanguinity and no family history of birth defects, mental retardation, or multiple miscarriages. Elective termination of pregnancy was accomplished at 19 weeks’ gestation.

The male fetus delivered was grossly abnormal, weighing 176 g, with a crown/heel length of 19.5 cm, a crown/rump length of 13.9 cm, and a head circumference of 14 cm.

The facial appearance was characterised by a high forehead with flat supraorbital ridges, hypertelorism, enophthalmus, and bilateral corneal opacities. The nasal bridge was broad, the tip bulbous, and the nares anteverted. There was a poorly demarcated philtrum and cupid’s bow, and marked micrognathia. The pinnae were small, simple, and low set. The neck was short and broad with mild webbing.

There was complete absence of the left hemidiaphragm. The heart had a double outlet right ventricle with a small but normally formed aortic valve, normal aortic and pulmonary outflow tracts, and a membranous ventricular septal defect. The mediastinum was shifted to the right and the hypoplastic left lung was approximately one-third the size of the right lung. The left hemithorax was occupied by the liver, spleen, stomach, and segments of the intestine. Apart from the retroperitoneal viscera only the descending colon and the greater portion of the liver remained in the abdomen. There was a small closed lumbar meningocele of 1 × 1 × 0.2 cm (amniotic fluid α-fetoprotein was reported to be within normal limits). There was complete lack of gyration of the cerebral hemispheres and absence of the optic tracts.

Although the facial appearance of the fetus reported here bears striking resemblance to those previously reported, corneal opacities, meningocele, and diaphragmatic hernia have not been previously noted. Though the eyes were not fully examined, the absence of optic tracts coupled with the appearance of enophthalmus suggest microphthalmia. Cardiac anomalies have been a frequent finding, including another case of double outlet right ventricle. Cystic kidneys, a finding in other cases, were not seen in this case.

I wish to stress the importance of reporting cases of rare autosomal aneuploidy, for they help to facilitate meaningful genetic counselling.

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References


Wishes of patients with retinitis pigmentosa concerning genetic counselling

Sir,

It was first established by Emery and Smith\(^{1}\) that only a minority of patients at high risk for transmitting a serious disease actually receive genetic counselling. At that time it was not known whether the remainder wished to have genetic counselling or not, but since then experience with registers for Duchenne muscular dystrophy and Huntington’s chorea,\(^{2,3}\) and experience with a genetic service for handicapped school leavers,\(^{4}\) has shown that the majority of such patients or their families welcome the offer of genetic counselling.

For the last four years, patients with retinitis pigmentosa have been visited as part of a study to determine its incidence and heterogeneity within the City of Birmingham.\(^{5}\) The opportunity was taken to ask every index patient his or her views on genetic counselling. Some patients were severely affected with loss of vision in their thirties and forties, but the majority of patients (those with dominant or non-genetic retinitis pigmentosa) had a mild disease with preservation of useful sight in their fifties and sixties.

There were 142 index patients who were visited, although more patients are in the incidence study in which secondary cases are included. Twelve patients did not give their views on genetic counselling as they considered them to be irrelevant: four because they were unmarried and eight because they had
decided not to have children anyway, five because of their severe visual handicap. Eight index patients were not asked for their views on genetic counselling because of difficulty in communication: three were deaf from Usher's syndrome, one was a recent Indian immigrant, three were mentally retarded (two with the Lawrence-Moon-Beidl-Bardet syndrome), and one was aged five. This left 122 index patients aged 14 to 83 whose views on genetic counselling are presented in the table. Ninety-one patients (0.75) either had genetic counselling or would have liked to have had it, in the sense that, had the risk been high, they would have thought seriously before having children. Of the 46 patients who had already been counselled, 11 had either been given incorrect advice or had misunderstood it. The proportion of patients who wished for genetic counselling was lower among the first generation immigrants than among patients born in the UK (0.41 compared to 0.82).

The patients have been categorised in the table according to their awareness of the disease before planning a family, and also according to their risks assessed by the authors. Thus, 'high risk' patients (those with a risk of about 1 in 10 or worse for a child or grandchild being affected) consisted of those with observed autosomal dominant inheritance; those with an X linked pedigree; and those isolated male patients who either had a female relative identified as a carrier on fundal examination or retinal function tests or both, or, if no female relative was available to be tested, had a high empirical risk of suffering from X linked retinitis pigmentosa. (Based on the population study in Birmingham males who were blind by the age of 40, or likely to be so, were more likely to have X linked retinitis pigmentosa than autosomal recessive retinitis pigmentosa). Three Muslim patients with probable autosomal recessive disease were also given high risks for having an affected child because they had married a first cousin.

Patients considered to have a low risk for having an affected child or grandchild were those with autosomal recessive disease (with the three exceptions mentioned above), isolated female cases, and isolated male patients who either had a mild disorder, or whose mother or daughter or both had normal retinal function tests and fundal examination.

The results show that the majority of patients with retinitis pigmentosa do in fact wish to have genetic counselling, in spite of the variable severity of the condition. Nine patients refused to be visited for the study without giving a reason and it is possible that none of the nine wished to discuss genetics. Even if this were so, there would still be 91/131 (0.69) who would like to have genetic counselling and who would take high risks seriously. The provision of genetic counselling should therefore be part of the management of patients with poor sight.

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**References**


**Table**: Numbers and proportions of patients with retinitis pigmentosa who had, or would have liked to have had, genetic counselling

<table>
<thead>
<tr>
<th>Patients born in UK</th>
<th>Patients born outside UK*</th>
<th>Total patients</th>
</tr>
</thead>
<tbody>
<tr>
<td>Patients who had symptoms or knew of an affected relative before starting a family</td>
<td>60/68 (0.88)</td>
<td>6/14 (0.43)</td>
</tr>
<tr>
<td>Patients with no symptoms and no knowledge of an affected relative before starting a family</td>
<td>221/32 (0.69)</td>
<td>31/8 (0.38)</td>
</tr>
<tr>
<td>Patients known by doctor to have a high risk (see text) for having an affected child or grandchild</td>
<td>47/54 (0.87)</td>
<td>2/8 (0.25)</td>
</tr>
<tr>
<td>Patients considered by doctor to have a low risk for disease in a child or grandchild (see text)</td>
<td>35/46 (0.76)</td>
<td>7/14 (0.50)</td>
</tr>
<tr>
<td>All patients</td>
<td>82/100 (0.82)</td>
<td>9/22 (0.41)</td>
</tr>
</tbody>
</table>

*9 West Indians, 2 Greek Cypriots, 11 Asians (7 Muslims, 2 Hindus, 2 Sikhs)
†These patients wished to know the genetic risks on behalf of their children
Wishes of patients with retinitis pigmentosa concerning genetic counselling.
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