the children at the time of the 1967 follow-up are summarised in table 1.

In the 1980 follow-up of the 179 women born in 1933 to 1949, 27 were not traced (25 were not traced through the National Health Service and two had emigrated). The information on the further children of the 152 who were traced is summarised in table 2.

Adding these new data, the totals for children are 21 sons affected in 173 (12.1%) and nine daughters affected in 182 (4.9%). These are now close to the figures for sibs for the same series of female patients, 10.0% and 6.3% for brothers and sisters respectively.

However, it will be seen from tables 1 and 2 that there is an indication of a secular change with a falling proportion of children affected among the more recently born, and there was also some suggestion of this in the figures for sibs. There are also indications that the incidence of pyloric stenosis in the general population may be falling. There is then a case for comparing contemporary births of sibs and children, rather than sibs and children of the same patients.

If we compare the children of patients born in 1933 to 1942 (mostly born between 1945 and 1975) and the sibs of patients born in 1950 to 1965 (who were also mostly born between 1945 and 1975) the figures are 7/51 for sons and 2/61 for daughters, and 8/103 for brothers and 0/105 for sisters. This suggests a higher risk for children. However, the numbers are small and a somewhat larger series from Belfast for patients born in 1957 to 1969 gave 20/160 brothers and 6/159 sisters, close to the figures for the children in the London series.

It is therefore probable that the proportion of sibs and children affected is similar and there is no need to postulate a direct maternal effect.

### Correspondence

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### References


### Delineation of trisomy 9 syndrome

**Sir,**

In response to Frohlich in the August 1982 issue of *Journal of Medical Genetics* (19:316-7) concerning delineation of trisomy 9 syndrome, we describe an infant who lived for 54 days. Skin fibroblast and blood lymphocytes showed trisomy 9, with one of the 9 homologues exhibiting an extra band in the 9qh+ region.1-3 (The 9qh+ variant was not in duplicate.) Variants in association with trisomy 9+ have been previously observed.4 5 In the report in the October 1981 issue of *Journal of Medical Genetics* (18: 390–2) by Frydman *et al*, the partial inversion of 9 described might well be reassessed in the light of recent studies,6 although such variants remain excellent markers, as was shown in the aforementioned report.

This baby was born to a mother who already had one normal child. The pregnancy had been unremarkable and she had only taken Amoxil and Debendox. At delivery there was a suggestion of polyhydramnios which had not been appreciated before. Birth weight was 2170 g. At birth the baby was mildly depressed, with Apgars of 4 at 1 minute, 6 at 5 minutes, and 9 at 10 minutes. Because of orofacial abnormalities the baby was unable to be intubated; however, he was easily resuscitated with bag and mask. There were a number of physical abnormalities. The head was very small with a circumference of 20 cm, well below the 10th centile for 38 weeks. There was a bilateral cleft lip and palate and the philtrum was on the tip of the nose, which was broad. The centre of the forehead protruded. Hypertelorism was present and palpebral fissures were narrow. The anterior and posterior fontanelles were both very large, but tension

### TABLE 1 1967 follow-up.

<table>
<thead>
<tr>
<th>Female patients</th>
<th>Children born up to 1966</th>
</tr>
</thead>
<tbody>
<tr>
<td>Year of birth</td>
<td>Number</td>
</tr>
<tr>
<td>1920–1932</td>
<td>46</td>
</tr>
<tr>
<td>1933–1942</td>
<td>64</td>
</tr>
<tr>
<td>1943–1949</td>
<td>115</td>
</tr>
<tr>
<td>Total</td>
<td>106 (20A)</td>
</tr>
</tbody>
</table>

R = Rammstedt, T = tumour felt and medical treatment, A = affected

### TABLE 2 1980 follow-up of further births.

<table>
<thead>
<tr>
<th>Female patients</th>
<th>Children born 1967–1980</th>
</tr>
</thead>
<tbody>
<tr>
<td>Year of birth</td>
<td>Number</td>
</tr>
<tr>
<td>1933–1942</td>
<td>55</td>
</tr>
<tr>
<td>1943–1949</td>
<td>97</td>
</tr>
<tr>
<td>Total</td>
<td>67 (1R)</td>
</tr>
</tbody>
</table>
Correspondence

appeared normal. Transillumination of the skull was normal. The ears were protruding and low set and there was redundant skin over the back of the neck. The occiput was flat. There was proximal shortness of the arms and the hands were short and stubby with a single crease on the left, but not on the right. The fingers were also short, many had rudimentary nails, and the fifth digit was incurved with only two phalanges. The baby's length of 46 cm was well below the 10th centile.

The legs were proximally shortened and the knees were easily subluxed when extended. The feet were unusual in that they were very foreshortened with retracted toes and there was some incurring of the sole. Again, many of the nails were of a rudimentary nature. There also appeared to be some mild oedema of the lower legs. Both kidneys were palpable on initial examination and appeared to be irregularly enlarged. The penis appeared to be of a rudimentary nature, but was probably of normal size and buried in the scrotum. The left testis could be moved down into the scrotum, but the right was not palpable. A single umbilical artery and a fairly deep sacral sinus were also noted. The baby had a relative bradycardia with poor peripheral pulses; blood pressure was not recorded. No cardiac murmurs were detected. There was a very flattened chest with widely spaced nipples and a short sternum. Chest x-ray showed an abnormally shaped rib cage with horizontally aligned ribs which were normal in number. A diffuse opacity over the left hemithorax overlying the cardiac shadow made interpretation difficult. ECG was unremarkable, apart from low voltages on the left ventricular leads.

A provisional diagnosis of a trisomy was made and the baby received routine nursing care and tolerated tube feeds. On the second day the baby's general condition remained stable, although peripheral pulses were still poor. His general condition deteriorated at around 30 hours of age, with increased cyanosis in room air and a high output cardiac state. The first heart sound was widely split and he developed intermittent tachypnoea. He became jaundiced on day 3 with a bilirubin of 224 and was started on phototherapy. At almost exactly 4 days of age he became acutely and profoundly cyanosed with bradycardia. Milk was subsequently aspirated from the upper airways, and there were widespread crepitations in both lung fields. The general condition remained stable but rather poor on day 5, and he subsequently died at the age of 5½ days.

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References