It has been shown in an earlier paper (Lorber, 1965) dealing with the family history of 722 infants born with spina bifida cystica or encephalocele, that 6.8% of 1256 sibs were also affected, either by the same condition or by the closely allied lesions of anencephaly or congenital hydrocephalus. This high incidence in sibs is suggestive of hereditary factors playing a part in the causation of these disorders. Several earlier workers have also come to similar conclusions, and details of their contribution have been given in the quoted paper. In no series, however, is the incidence in sibs 1:4, as one would expect in simple recessive transmission. Thus it appears possible either that there is multifactorial inheritance, or that extraneous factors need to be present for spina bifida to become manifest. It is suggested, however, that simple recessive inheritance is not altogether excluded for two reasons (Lorber, 1965). The first is that spina bifida is a potentially lethal condition even before birth, and the gene responsible for it may lead to very early foetal death with non-recognition of the defect, or even of the pregnancy. The second reason is that, as far as we are aware, there is no knowledge about the connexion between spina bifida cystica and spina bifida occulta. Spina bifida occulta is very common in the general population and may conceivably represent the mildest form of this disorder. If this reasoning were correct then the true incidence of these malformations in sibs could reach the proportion of 1:4, as demanded by the laws of recessive inheritance.

In order to get nearer to determining the influence of heredity in spina bifida cystica, we decided to investigate the relation between this condition and spina bifida occulta by determining the incidence of the latter among the parents of patients with spina bifida cystica. The incidence of spina bifida occulta in a control series of adults, drawn from the same population, was also determined.

It was considered unwise and unprofitable to determine the incidence of spina bifida occulta in sibs, while they were still children. First, we did not wish to expose them to unnecessary radiation, and secondly, the radiological incidence of spina bifida occulta in children is so high in the general child population, reaching over 80% (Sutow and Pryde, 1956; Fawcitt, 1959), that finding even very high figures would have been meaningless.

Material and Technique of Investigation

(a) Control group. The control group consisted of a consecutive series of 200 adult patients referred for any radiological investigation which included a satisfactory antero-posterior view of the lower lumbar and upper sacral spine. The majority of patients were referred for excretion urography. Patients being investigated for 'backache' or symptoms suggestive of a localized lumbosacral lesion were excluded.

Spina bifida occulta was only considered to be present when there was a clear defect in one or more neural arches. Several subjects showed irregularities of various types in the neural arches, without evidence of nonfusion. For the purpose of this survey these were considered as normal.

(b) Parents of affected children. We considered the ethical problem of subjecting symptom-free adults to ionizing radiation which might not be to their direct benefit. This hazard was considered in relation to the potential benefit in genetic counselling, should this investigation provide positive information. These facts were discussed with the parents of children with spina bifida cystica before asking them to have a single antero-posterior film of their lumbosacral spine. The average gonad dose received was 10 milliroentgens (mr.) in males, and 80 mr. in females, compared to an annual background radiation of 100 mr. (Ardran and Crooks, 1964).

Several parents refused, but most were anxious to co-operate and obtain, if possible, information for their own potential benefit, and for the benefit of others who are or will be faced with the same difficult problems as they were themselves. Those parents who already had two or more affected infants were particularly anxious to take part.
TABLE I
Spina Bifida Occulta in Parents of Children with Spina Bifida Cystica and in Adult Controls: Result of Spine x-ray Films

<table>
<thead>
<tr>
<th>Subjects</th>
<th>Normal</th>
<th>Spina Bifida Occulta</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>No.</td>
<td>%</td>
</tr>
<tr>
<td>Parents</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Mother</td>
<td>161</td>
<td>85·7</td>
</tr>
<tr>
<td>Father</td>
<td>131</td>
<td>73·2</td>
</tr>
<tr>
<td>Both</td>
<td>292</td>
<td>79·6</td>
</tr>
<tr>
<td>Controls</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Women</td>
<td>80</td>
<td>96·3</td>
</tr>
<tr>
<td>Men</td>
<td>111</td>
<td>94·9</td>
</tr>
<tr>
<td>Both</td>
<td>191</td>
<td>95·5</td>
</tr>
</tbody>
</table>

Results

The incidence of spina bifida occulta in normal controls was 3 out of 83 or 3·7% in women, and 6 out of 117 or 5·1% in men, giving an over-all incidence of 4·5% (Table I). The incidence in parents was 27 out of 188 or 14·8% in mothers, and 48 out of 179 or 26·8% in fathers: the over-all incidence here was 21·4% among 367 parents. The difference in incidence between control women and mothers was significant at the 1% level ($\chi^2 = 6·76$) and between control men and fathers at 0·1% level ($\chi^2 = 22·3$). For the whole group the differences were significant at the 0·1% level ($\chi^2 = 26·05$) (Table I).

We have analysed separately the incidence of spina bifida occulta in those parents who had two or more children affected by spina bifida cystica. As is seen in Table II, the incidence in these parents was almost identical to that of the whole groups. In only two instances was spina bifida occulta present in both parents.

Discussion

The incidence of spina bifida occulta in the fifth lumbar or the first sacral vertebra is so common, that it is usually regarded as a variant of the normal

and of no significance. The first sacral segment is far more often affected than the fifth lumbar vertebra (Walker and Bucy, 1934; Brailsford, 1953; Sutow and Pryde, 1956). Radiologically demonstrable spina bifida occulta is far commoner in children than in adults. For example in Fawcitt’s series (1959), 82% of 500 English children had radiological evidence of spina bifida occulta. The progressive decrease of the incidence up to adult age was well shown by Sutow and Pryde (1956) in a Japanese survey, in which an incidence of 58·7% among 155 children, 7-8 years old, fell to 28% among 182 adults. The incidence of spina bifida occulta in adults of Western communities is usually much lower. In the series of 500 described by Gillespie (1949), it was 4·8%, and in that by Sutherland (1922), a very large series of 12,000, it was just over 5%. His sex incidence of 2 : 1 in favour of males was much the same as in our series of parents of children with spina bifida cystica. The incidence of 5% in our control group agrees well with these, and other larger reported series of cases, though Walker and Bucy (1934) reported an incidence of about 2% in a group of over 7000 subjects in Chicago. (One might anticipate a higher incidence of spinal abnormalities in patients referred as in our control series with urinary tract symptoms. However, the incidence of spina bifida occulta in the control group agrees with that obtained in the other series noted in the text; in addition, any increase in spinal abnormalities in the control group would tend to lessen the difference between it and the parents of affected children.)

In our own series the incidence of spina bifida occulta was some 4 times higher in parents of our patients with spina bifida cystica than in our control group and the differences are highly significant at the 0·1% level. The sex ratio of approximately 2 : 1 in favour of males is of the same order as in the ‘normal’ population. Such a considerable excess of cases among parents suggests that spina bifida occulta is aetologically related to spina bifida cystica, and this finding helps a little towards the evidence that spina bifida cystica may be a recessively inherited condition (Lorber, 1965). Nevertheless, detailed analysis of the individual family patterns does not help in giving any more accurate genetic advice to couples. The incidence of spina bifida occulta was no commoner in parents with more than one affected child, and among 36 such couples in only 2 instances was spina bifida occulta present in both parents. In 21 couples with two or more affected children both parents had a negative spine x-ray film. Altogether there were only 7 couples in the whole series who both had spina bifida occulta.

TABLE II
Spina Bifida Occulta in Parents with Two or More Affected Children: Results of Spine x-ray Films

<table>
<thead>
<tr>
<th>Subjects</th>
<th>Normal</th>
<th>Spina Bifida Occulta</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>No.</td>
<td>%</td>
</tr>
<tr>
<td>Mothers</td>
<td>30</td>
<td>83·3</td>
</tr>
<tr>
<td>Fathers</td>
<td>25</td>
<td>75·7</td>
</tr>
<tr>
<td>Both</td>
<td>55</td>
<td>79·7</td>
</tr>
</tbody>
</table>
Incidence of Spina Bifida Occulta in Parents and in Controls

Summary

Radiological investigation of the lumbo-sacral spine was undertaken to determine the incidence of spina bifida occulta in parents of children affected with spina bifida cystica. The incidence of spina bifida occulta was 14.3% among 188 mothers and 26.8% among 179 fathers: an over-all incidence of 20.4% among 367 parents. The incidence of spina bifida occulta in a control group of 200 adults living in the same district was 5%. Although this difference is highly significant, no help can be derived for the genetic advice of individual couples, because spina bifida occulta was not commoner among parents with more than one affected child, and because in the majority of families neither parent had spina bifida occulta.

References


Spina bifida cystica. Incidence of spina bifida occulta in parents and in controls.
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Arch Dis Child 1967 42: 171-173
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