STUDIES OF FAMILIAL INCIDENCE OF SPONDYLOLYSIS

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It is well known that spondylolysis and spondylolisthesis play an important role in the development of low back pain. At the Department of Orthopaedic Surgery of Kurume University Hospital, 534 cases of spondylolysis and spondylolisthesis were examined during the last 10 years. We have been especially interested in the etiology of spondylolysis and have conducted clinical, radiological and histological studies of both living cases and those that have come to autopsy, reaching the conclusion that spondylolysis is acquired by the physical stress of daily life upon lumbar vertebrae which have some undetermined congenital predisposition for the disease.

This report is taken from a series of studies on the familial incidence of spondylolysis.

A family survey was carried out in 100 cases of spondylolysis and spondylolisthesis comprised of 63 males and 37 females. Ages ranged from 14 to 72 years, and were predominantly over 20. Fig. 1.

The number of parents, siblings and children of the patients examined reached 234, 111 males and 123 females. Among these, 24 were below the age of 9, 61 were from 10 to 19, and 149 were adults above the age of 20. Fig. 2.

Among 243 family members, a defect in the pars interarticularis of the lumbar vertebrae was found on anteroposterior, lateral and oblique X-ray in 67 persons (29%), 36 males (32%) and 31 females (25%). Only 3 persons (12·5%) were below the age of 9. Seven persons (11·5%) were in the second decade, and 57 (38%) were adults. With few exceptions these persons in whom the defect was discovered were free of clinical symptoms. Fig. 3.

In order to assess accurately the frequency of the defect in the Japanese population at large, the lumbar vertebrae in 218 cadavers, 130 males and 88 females, ranging in age from 12 to 74 years, were studied by macroscopic and radiologic observation. The defects were found in 16 (7%) of the 218 cases, 12 males (9%) and 4 females (4·5%). This frequency is in accord with figures previously reported for Japanese (5-10%). With these control results, the familial incidence of 29% is definitely considered significant.

Corresponding values of familial incidence are also high in other countries, as reported by Baker (39%), Freiberg (24·5%), Wiltse (26%) and others. Fig. 4.

Several examples of interesting family trees encountered in our survey are shown in the slide. In families I, II and III, the defects are seen in two generations. In family IV and V the defect extended over three generations. In family IV the defect was seen in the mother and son of the patient, while a similar defect was seen in the father and one of the daughters in family V. In addition, other lumbosacral anomalies, such as spina bidida occulta and transitional vertebrae, were noted in these families. Several X-ray pictures will be shown. Fig. 5.

The left X-ray represents the oblique roentenogram of a patient in family I, a 41 year old female with spondylolysis of the fifth lumbar vertebra. The right X-ray shows a similar picture of her 21 year old daughter, with the defect in the fifth lumbar vertebra. Fig. 6.

This is an anteroposterior roentgenogram of one of the daughters of the patient, a 14 year old girl. Spina bifida occulta of the first sacral and sacralization of the fifth lumbar vertebra are seen. Fig. 7.

This is a lateral roentgenogram of the 11 year old son of a patient in family IV. The defect and slight forward slipping of the fifth lumbar vertebra are seen. Fig. 8.

The left X-ray shows a lateral view of a patient in family V, a 38 year old female with the defect in the fourth lumbar vertebra and marked slipping of the fifth lumbar vertebra. The right X-ray shows a lateral roentgenogram of her 67 year old father with sacral displacement of the fifth lumbar vertebra. Fig. 9.

From the data obtained by survey of the above-mentioned 100 families, the manner of occurrence of the defect in families over a ten year period was analysed. Fig. 10.

Of 42 siblings of patients whose parents were also studied, the defect was found in 9 (21)%.

Number of Patients

Age	Male	Female	Total
14 ~ 19	6	3	9
20 ~ 29	18	5	23
30 ~ 39	20	8	28
40 ~ 49	10	14	24
50 ~ 59	3	6	9
60 ~	6	1	7
Total	63	37	100

Fig. 1.

Number of Patients' Families Examined

Age	Male	Female	Total
1 ~ 9	13	11	24
10 ~ 19	27	34	61
20 ~ 29	14	11	25
30 ~ 39	10	13	23
40 ~ 49	17	14	31
50 ~ 59	9	19	28
60 ~	21	21	42
Total	111	123	234

Fig. 2.

Arch Defect among Family Members

Age	Male	Female	Total
7 ~ 9	2(15.4)	1 (9.1)	3(12.5)
10 ~ 19	5(18.5)	2 (5.9)	7(11.5)
20 ~ 29	6(42.9)	4(36.4)	10(40.0)
30 ~ 39	2(20.0)	1 (7.7)	3(13.0)
40 ~ 49	5(29.4)	6(42.9)	11(35.5)
50 ~ 59	4(44.4)	12(63.2)	16(57.1)
60 ~	12(57.1)	5(23.8)	17(40.5)
Total	36(32.4)	31(25.2)	67(28.6)

( ) : Per cent

Fig. 3.

Spondylolysis in Autopsy Materials

Age	Male	Female	Total
12 ~ 19	2		2
20 ~ 39	17 (3)	4	21 (3)
40 ~ 59	40 (4)	14 (2)	54 (6)
60 ~	71 (5)	70 (2)	141 (7)
Total	130 (12)	88 (4)	218 (16)
Per cent	9.2%	4.5%	7.3%

( ) : Spondylolysis

Fig. 4.

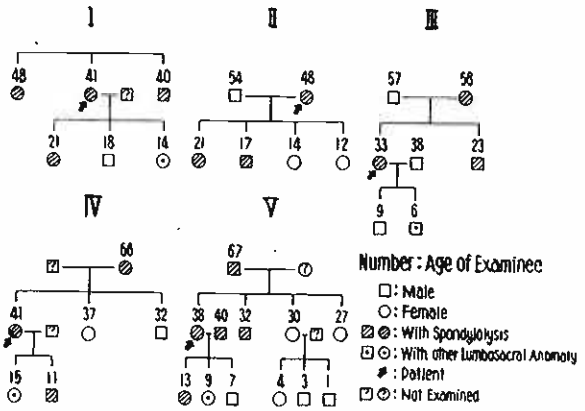

Fig. 5.

Siblings were classified according to groups depending upon the incidence in the parents, as follows:

GROUP A: The defect was present in neither parent.

GROUP B: The defect was present in only one of the parents.

GROUP C: The defect was present in both parents.

In group A, the defect was found in none of the 22 siblings and in group B and C, it was found in 7 of 17 (41%) and in 2 of 3 (67%), respectively. Statistical analysis showed these figures to be significant.

Figure 11: Of 48 children in 14 families in which the survey was also carried out on patients' spouses, the defect was found in 24 (50%). The children were classified into two groups:

GROUP A: The defect was absent in one parent.

GROUP B: The defect was present in both parents.

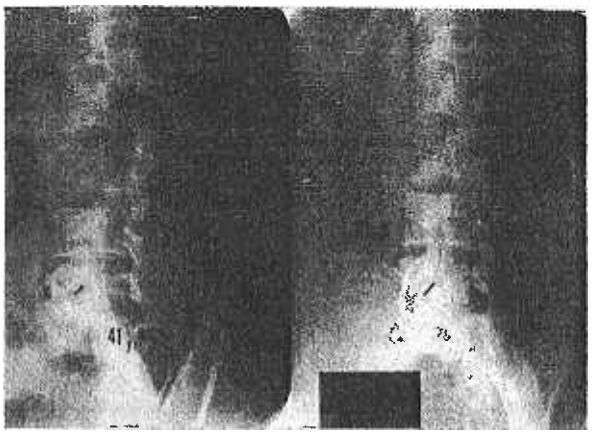

Fig. 6.

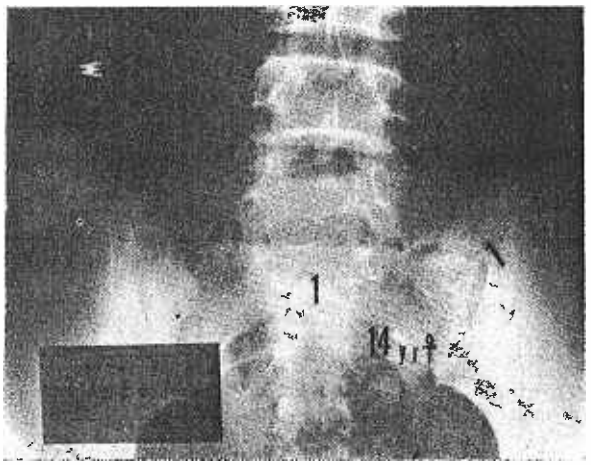

Fig. 7.

Fig. 8.

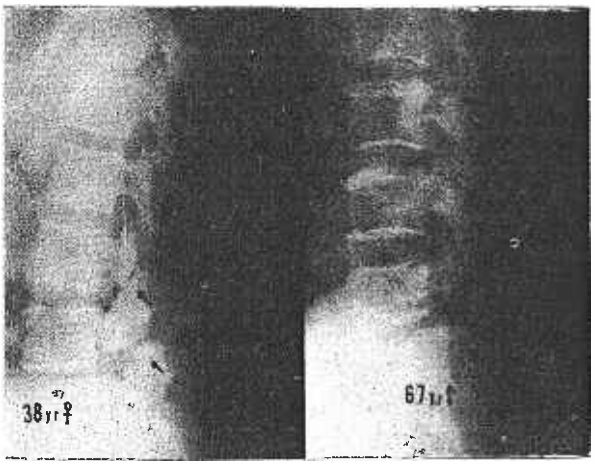

Fig. 9.

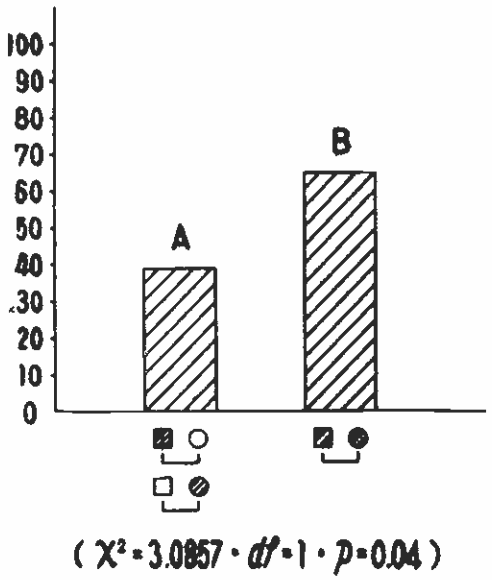

Fig. 10.

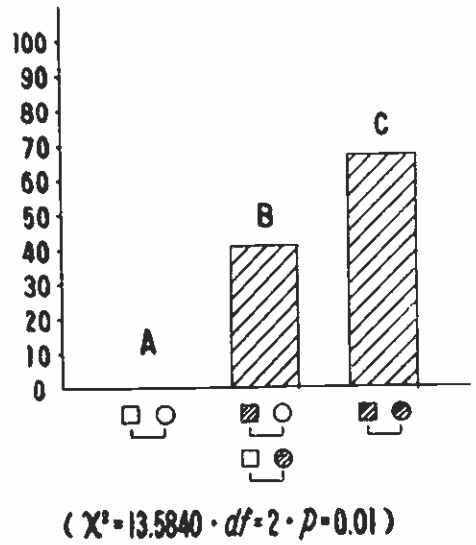

Fig. 11.

No cases were found where neither parent had the defect.

The defect was found in 11 of 28 cases (39%) tn group A and in 13 of 20 (65%) in group B, indicating a significant difference.

CONCLUSION

Upon examination of these findings from the viewpoint of genetics, it is suggested that the predisposition for spondylolysis is inherited by autosomal simple dominant inheritance with a rather high penetrance.