THE FAMILIAL INCIDENCE OF RHEUMATOID SPONDYLITIS*

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The familial incidence of rheumatoid spondylitis has not been adequately appreciated. The medical literature contains a few reports of families with more than one member having this form of spinal arthritis, but no serious effort to determine the familial incidence has been made. Two reports have described the existence of rheumatoid spondylitis in brothers (Stecher and Hauser, 1946; Weil and Allolio, 1930); the occurrence in identical twins has also been noted (Campbell, 1947; Ray, 1932). Also Scott (1942) has mentioned the development of rheumatoid spondylitis in a brother and sister, in two sisters, and in a father and son. Dawson (1935) was convinced that the familial incidence of rheumatoid spondylitis was greater than the incidence in the general population, and referred to a report by Berckhardt of a family in which the patient's mother and four brothers also had rheumatoid spondylitis. On the other hand, in the excellent and comprehensive review of 1,035 cases of rheumatoid spondylitis (the largest recorded single series) reported by Polley and Slocumb (1947), the familial incidence of the disease was not discussed.

The purpose of this paper is to report the familial incidence of rheumatoid spondylitis as it was found in a systematic study of the families of patients with rheumatoid spondylitis in the arthritis clinic of the Hospital for Special Surgery, New York City, and the Rackham Arthritis Research Unit, University of Michigan.

Method of Study

One hundred and fourteen patients with rheumatoid spondylitis were asked whether any person among the parents, siblings, or children had pain, stiffness, or limitation of motion of the back. Thirty affirmative answers were obtained. In many instances the patients were quite positive that other members of their family had a disability similar to theirs. We were able to examine twenty-four of the suspected relatives, belonging to eighteen families. Some submitted to examination voluntarily, while others had to be sought out. Thus,

no sampling bias is introduced as might have been the case had a purely voluntary group been used.

Results

Of the twenty-four suspects investigated, thirteen cases occurring in ten families showed findings characteristic of rheumatoid spondylitis such as: back pain and tenderness, stiffness, limitation of motion of the spine, and radiographic evidence of sacro-iliac arthritis and/or calcification of spinal ligaments. Thus it was found that in ten of the 114 families one or more members of the family in addition to the presenting patient, showed evidence of rheumatoid spondylitis. If there were no other cases of rheumatoid spondylitis in the unexamined suspects, the familial incidence of this disease would be 9 per cent. (see Table). If the incidence of spondylitis in the unexamined group of thirteen families in whom there were suspects were the same as was found in the examined group, the familial incidence would increase to 13 per cent. Certainly the true familial incidence in our group of cases lies between 9 and 13 per cent. Since most of the cases were found in siblings, this high incidence becomes more remarkable when it is noted that in this group of 114 cases eight had no siblings.

Illustrative Cases

A convincing example of the familial incidence of the disease is found in family H. (Fig. 2). H. H., the first individual to be examined, was found to have characteristic clinical and radiographic changes of advanced rheumatoid spondylitis. Not until he was specifically questioned did he state that his father and two brothers had a disability thought to be similar to his. They were subsequently examined and found to be suffering from the same disease. Radiographs of the spine in all four cases showed calcification of spinal ligaments and other ankylosing features of spondylitis (Fig. 1). It is interesting to note in the radiographs of the father (M. H.) that the sacro-iliac joints appear normal while the dorsal and lumbar spine show extensive disease of facets and ligamentous calcification.

Less striking, but showing involvement in two generations is the family F. (Fig. 3), in which a brother and a

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FIG. 1.—Family H. Father and three sons show evidence of rheumatoid spondylitis. Note (in C) that in the father there are extensive pathologic changes in the spinal ligament and intervertebral facets, but that the sacro-iliac joints are normal.

FIG. 4.—Radiographs of the pelvis and lumbar spine of twins, aged 23, showing arthritis of the sacro-iliac joints.

son of the patient are afflicted with rheumatoid spondylitis.

Included among our cases is a pair of identical twins, aged twenty-three, each of whom while in military service began to complain of pain low in the back radiating to the buttocks. Tenderness over the sacro-iliac areas with associated lumbar muscle spasm was present. Radiographs revealed early arthritic changes in the sacro-iliac joints (Fig. 4).

Discussion

In the post-mortem study of 10,000 vertebral columns Schmorl and Junghanns (1932) found eight cases of rheumatoid spondylitis, an incidence of only 0.08 per cent. This is in sharp contrast with the familial incidence of rheumatoid spondylitis, found in our study to be at least 9 per cent. This suggests that the familial incidence of rheumatoid spondylitis is approximately one hundred times greater than the incidence in the general population. However, it should be noted that Schmorl's study was confined to anatomical changes. It is possible that many cases of rheumatoid spondylitis diagnosed on clinical findings show such slight deviations as to be overlooked on post-mortem examination of the spine, so that the discrepancy in the incidence in Schmorl's cases, based on anatomical studies, and our cases, based on clinical studies, must be borne in mind.

The relatively high familial incidence of rheumatoid spondylitis suggests that there may be important hereditary factors. However the similarity of environmental influences within families must also be considered. Further investigations of the hereditary and environmental influences on this disease are necessary.

The practical point to be emphasized is that the managing physician should be alert to the likelihood of a higher incidence of this disease among the close relatives of his patients, and steps should be taken to discover them as soon as possible, for with early diagnosis the chance for more successful therapy is greatly increased.

Summary

Among the close relatives of 114 cases of rheumatoid spondylitis, the familial incidence of this disease clinically was found to be at least 9 per cent.

The physician managing patients with rheumatoid spondylitis should be alert to discover other cases among relatives of these patients.

TABLE
STATISTICS OF FAMILIAL INCIDENCE
RELATING TO 114 CASES OF RHEUMATOID
SPONDYLITIS

	No.	Per cent. based upon 114 families
Families in which spondylitis was suspected among other members from history of patient	31	37
Families examined	18	16
Families in which rheumatoid spondylitis was found by examination, among relatives	10	9
Families which could not be examined	13	—

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La Nature Familiale de la Spondylite Ankylosante

RÉSUMÉ

On a observé parmi les proches parents de 114 malades atteints de spondylite rhumatismale l'existence de la maladie dans au moins 9 pour cent des cas. Le médecin traitant des malades atteints de spondylite rhumatismale devrait chercher à découvrir rapidement d'autres malades parmi les parents de ceux qu'il soigne.