CASE OF ALBERS-SCHÖNBERG'S DISEASE REVIEWED AFTER 20 YEARS

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In 1936 a case of Albers-Schönberg's disease[1] was described by one of us (J.H.K.) and R. A. Roberts,[2] with photograph and X-ray plates. In January 1956, about 20 years later, the patient presented herself again and the skeletal system could again by X-rayed to assess the present state of her bones.

In the intervening years she had suffered no serious illnesses, had had one infant, and was now wearing glasses and using a hearing aid. Her only complaint now was a neuralgic pain in the right half of the face. She had had some teeth extracted without any ill effect to the mandible. In all the years she had never had any fracture of any of her bones.

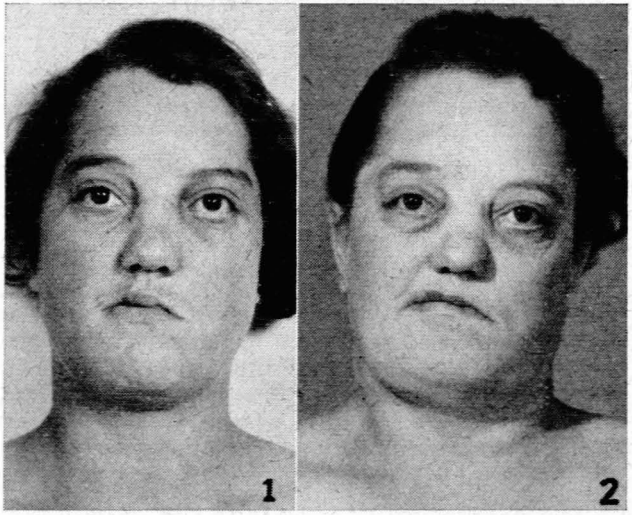

Fig. 1. (Published in Brit. Med. J., 25 April 1936, **1**, 837.) The patient in 1936, when 21 years old. Note the wide separation of the eyes, indicating some degree of ocular hypertelorism, the prominence of the nasal bone, the left-sided facial paralysis, and the massive lower jaw.
Fig. 2. The patient in 1956, for comparison. Note the great massiveness of the lower jaw.

Clinically no untoward pathological departure from the normal could be detected other than features seen in 1936 and still readily visible on inspection—exophthalmos, facial paralysis and the massive lower jaw (Figs. 1 and 2). The great increase in bulk of the lower jaw was particularly striking, the heavy broad bone being palpated only by a wide separation of the fingers. X-ray films reveal the gross appearance.

As in 1935, X-raying of the skull called for a heavy exposure before satisfactory films could be procured, and the features are very much the same as reported then. The rest of the skeletal system, thoracic cage, humerus, hands, spine, pelvis, femur and feet were all X-rayed again and also reveal no appreciable changes from the skiagrams described in 1936, other than those associated with growth. No evidence of fractures was detected. There was no appreciable additional narrowing of the medullary cavities of the long bones. The only really striking change from the appearance of 20 years ago, was the heavy massiveness of the broad lower jaw, while the X-ray films of the skull once again reveal the thickening of the occipital region and the homogeneous thickening of the whole base of the skull. Biochemical examination of blood and urine revealed nothing abnormal.

In 1937 Falconer[3] reported a case of generalized osteosclerosis of familial type, with facial and other deformities. The patient (male) was sent into hospital from Ladismith, Cape, by Blyth, and his sister was reported as suffering from even greater facial deformitity due to marked thickening of the upper and lower jaws. Falconer compared his case with our present case (as reported in 1936[2]) and stated: 'There can be no doubt that Kretzmar and Roberts' case and the present case are identical. Neither Dr. Roberts nor Dr. Blyth are able to trace any relationship between the two families, but as they both come from the Cape Province, it is not altogether excluded'.

Further questioning of the patient in 1956 on this same point elicited the fact that she knew of no branches of the family tree in the neighbourhood of Ladismith, Cape.

'The present case', Falconer stated in conclusion, 'Dzierzynsky's first family, and Lauterburg's case,[4-5] all have in common a generalized osteosclerosis affecting several members of the family with involvement of both the base and

vertex of the skull, the bones of the face and a characteristic alteration of the shape of the metacarpals. There is no tendency to increased fragility of the bones. There are no associated blood changes and the condition apparently runs a benign course. They would appear to present a definite entity. Although there is no evidence of family involvement in Kretzmar and Roberts' case the clinical and radiological features are so absolutely identical that it also must be included in this group'.

From a recent extensive review of the literature on 'osteopetrosis in adults' by Hinkel and Beiler[6] it would appear that the case reported by us falls in the category of the benign type of Albers-Schönberg's disease, where the patient may continue enjoying good health, without suffering from anaemia and without the repeated occurrence of pathological fractures.

SUMMARY

A case of Albers-Schönberg's disease examined in 1935 and reported in 1936 is reviewed in January 1956. No significant changes are revealed in the X-ray films of the skeletal system compared with the descriptions of 20 years ago. There is no history of any fracture in the 20 years and the general health of the patient has been good.

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