LOBSTEIN'S SYNDROME

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The purpose of this brief communication is to add a footnote to a paper published in this journal 19 years ago (Ottley, 1932) describing a family of which many members showed the hereditary condition named by Lobstein, who in 1833 gave the first account of it, osteopsathyrosis. The cardinal

hair, and deficient formation of dental enamel.

The hypothesis that the condition originates in an inherited developmental defect of the mesoderm appears to have won general acceptance. The association with defects in tissues of other origin, e.g. in the enamel of the teeth, would, on this

TABLE I
Genealogical Tree of Family

manifestations of Lobstein's syndrome are fragility of bones, blue coloration of the sclerae, and deafness (otosclerosis), occurring singly or in combination in affected members of certain families. Various subsidiary features have been described in some families, including malformation of the skull (*crâne à rebord*), precocious arcus senilis, laxity of ligaments, abnormal electrical reactions of the muscles, white patches in the nails, unusual fineness of the

supposition, be merely fortuitous.

In the family under consideration the most striking feature is the tendency for the females to be more severely affected than the males. No male member is known to have exhibited the whole triad of cardinal signs. Since the date of my first paper, two members, numbers 9 and 21 (mother and daughter), have had operations for cystic mastitis, the mother on both breasts, the daughter on one,

at the age of only 21. This daughter, now aged 23, shows a particularly well marked arcus senilis, most striking in so young a woman, and sclerae of a slatey blue. She has never had a fracture, but fractures are a less conspicuous feature in this family than in some of the families which have been reported. She has as yet no sign of otosclerosis. The mother, number 9 in Table 1, the genealogical tree brought up to date, had an operation for

hallux valgus in 1937. The excised bone showed no histological abnormality. She is now stone deaf, able to hear nothing without a hearing apparatus and very little with it.

REFERENCES

- Lobstein, J. (1833). 'Traité d'Anatomie pathologique.'
vol. 2, p. 204. Paris.
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