FAMILIAL MULTIPLE CYSTIC DISEASE OF THE JAWS

WILLIAM A. JONES, M.D., F.R.C.P.(C)
Queen's University, Kingston, Ontario

Familial multilocular cystic disease of the jaws, the title of this paper, is a provisional name, given to describe an unusual condition occurring in three children, members of the same family, of the Hebrew race, aged six, five, and four years (Fig. 1). It is characterized by marked fullness of the cheeks and jaws and a slight upward turning of the eyes, revealing white lines of sclera beneath. There is also noticeable swelling of the submaxillary regions. The full round cheeks and the upward cast of the eyes give the children a peculiarly grotesque, cherubic appearance. As a matter of fact, they are active, intelligent children, showing a child's normal interest and curiosity as to their surroundings.

Both jaws are felt as hard, protuberant masses bulging outward to form a bilateral painless swelling of the face. The teeth are irregularly placed and many are missing. The oldest boy pos-

1 Read before the Radiological Society of North America at the Seventeenth Annual Meeting, at St. Louis, Nov. 30–Dec. 4, 1931.
sesses only nine teeth at present, while the little girl has ten. The alveolar ridges are extremely wide, and in the upper jaw this gives rise to a narrow V-shaped palate. There is no evidence of local infection in the mouth or nose, and the tonsils are not enlarged. The submaxillary lymphatic glands are chronically enlarged in all three children. At the present time the greatest degree of enlargement is seen in the youngest. Five months ago that distinction belonged to the little girl.

Fig. 2 brings out clearly the enlargement of the superior maxilla. The irregularities of dentition and the multiple cystic formation of the jaws are well seen in the accompanying roentgenograms. In this connection it may be said that radiological examination of the chest and of the other bones of the body reveals nothing of an unusual nature. Fig. 3 shows the left side of the jaws of the six-year-old boy. The jaws are remarkably symmetrical, and the disturbance is widespread throughout the bony parts. The arrangement of circular translucent areas and bony septa suggests cystic adamantinoma, but the involvement of practically all of both jaws by that condition would be most unusual.

This boy was apparently normal at birth, and the deciduous teeth were present at one and a half years. He had some thymic attacks early in his second year. A radiograph failed to show any definite enlargement of the thymus but symptoms cleared up under x-ray therapy. The lower jaw happens to show in films made at
that time, and no abnormal condition can be observed in the bone. About the end of the second year the cheeks and jaws began to enlarge and the submaxillary lymphatic glands were swollen. This condition became more noticeable during the next two years, but for the last two years there has been no apparent progress of the lesion as far as external appearances are concerned, and the rest of the face and head are growing proportionately larger. The lymphatic enlargement is not as marked as formerly. It would appear from these signs that the lesion is self-limiting, and this view is partially supported by the appearance of the jaws in the film. It will be seen presently that the bony septa are greater in number and much more heavily marked than in the films of the other children.

Fig. 4 shows the condition of the jaws in the girl, aged five. The cystic areas are larger on the average and the bony walls are fewer in number and more finely marked than in her older brother. This child was apparently quite normal as a baby. She cut her first tooth at eleven months, but the lower jaw never received its full complement of deciduous teeth. She now has only eight baby teeth and two permanent ones. Her cheeks and jaws began to enlarge about the beginning of the third year. A few months ago this child showed a rather marked secondary anemia, felt poorly, and was listless. At this time there was an increase in the size of the submaxillary glands. There was no rise in temperature and no white blood cell changes. A syrup of iodine and iron was given. The child is now healthy and active. The lymph glands have receded to half their former size.

Fig. 5 shows the youngest of the children, aged four. The changes are similar to those seen in his sister. The cystic areas are large, and the amount of bony network is comparatively the same.

This boy was also normal as a baby. He cut his first teeth at six months and had all his deciduous teeth. He now has seventeen baby teeth. His cheeks and jaws began to enlarge about the end of the second year. He is healthy and active, but lately there has been a marked increase in the size of the lymphatic glands under the chin. An enlarged submaxillary lymph node was removed from this child for examination (Figs. 6 and 7). The report of the pathologist, Prof. James Miller, is as follows: "Submaxillary lymph node from right side of neck. Oval mass measuring 2 x 1.5 cm., pinkish grey in color. Microscopic examination shows diffuse fibrous overgrowth. The capsule is thickened, as are also the trabeculae. There is well marked proliferation of endothelial cells in the sinuses. This appears to be the precursor of the fibrosis. The lymph follicles are well preserved but show in their
Figs. 3-5. Roentgenograms of the Three Children, Showing Disturbances of Dentition and Cystic Areas in the Jaws

Note the small size of the cystic areas and the relatively heavy bony septa in the oldest child (Fig. 3) as compared with his sister (Fig. 4) and younger brother (Fig. 5).
centers a tendency to fibrosis. The fibrous tissue here, as elsewhere, tends to show hyaline change.

There is a fourth child in the family, about one year of age, who up to the present is normal. There is no evidence of any similar condition existing in other relatives.

![Image of lymph node with fibrosis and endothelial cells](image)

**Figs. 6 and 7. Low-power and High-power Views of a Section of a Lymph Node from the Four-year-old Boy, Showing Fibrosis and (below) Collection of Endothelial Cells.**

These cases are presented for diagnosis. I believe that they represent an anomalous development of the dental structure with widespread tumor formation, and that the lymphatic glands are secondarily the seat of chronic inflammation. In view of the history of the eldest boy it would appear that the condition may be self-limiting.