Gardner’s Syndrome Without Polyposis?

W. Fuhrmann
Institut für Humangenetik der Universität Heidelberg (Direktor: Prof. Dr. F. Vogel)

U. W. Schnyder
Universitäts-Hautklinik Heidelberg (Direktor: Prof. Dr. U. W. Schnyder)

K.-H. Kärcher
Universitäts-Strahlenklinik (Czerny-Krankenhaus) Heidelberg (Direktor: Prof. Dr. J. Becker)

H. Pfeifer
Universitätsklinik und Poliklinik für Mund-, Zahn- und Kieferkrankh Heidelberg
(Direktor: Prof. Dr. Dr. R. Reiter)

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A family is described in which five persons in three generations were affected with the typical osseous and cutaneous lesions of Gardner’s syndrome, but did not show any signs of polyposis. Four individuals could be studied clinically. In an 18-year-old boy and his 45-year-old mother special X-ray studies of the large bowel were performed and revealed normal appearance of the mucosa. It is suggested, that this represents an example of a bisymptomatic osseo-cutaneous type of the Gardner syndrome. The existence of other bisymptomatic types could be expected. A cutaneo-intestinal form may be represented by an earlier description by Oldfield. No cases of a familial osseo-intestinal type are known to the authors. Gardner’s syndrome is caused by a single pleiotropic gene and inherited as an autosomal dominant. The bisymptomatic forms may be due to other alleles at the same locus. Familial polyposis of the colon without extra-intestinal symptoms represents a different entity due to another possibly nonallelic gene.

In 1950 and 1953 Gardner and coworker defined a syndrome, the characteristic features of which were multiple or diffuse intestinal polyposis, osteomas, fibromas, and epidermal cysts. In these families the polyposis very frequently gives rise to carcinoma of the rectum or colon. The pedigree of Gardner’s original family strongly suggested dominant inheritance due to a single autosomal gene with pleiotropic effect. Furthermore, from this family and several subsequent publications of other authors the impression was gained that the intestinal polyposis was a regular feature of Gardner’s syndrome. Its manifestation was found with high constancy after completion of the 3rd or 4th decade of life. Other authors suggested that two closely linked genes were causative in Gardner’s syndrome.

We have seen a family with all the typical features of Gardner’s syndrome except for intestinal polyposis. This seems to raise the question whether a bisymptomatic form of Gardner’s syndrome exists. A detailed description of our observation will be given elsewhere, as we believe that this is one of the first families with Gardner’s syndrome described in the German language literature.
Case report

The pedigree of the family No. is given in Fig. 1.

The first patient who came to the attention of the Heidelberg University hospitals was the boy N. R., III,8, born 1953. A small firm and indolent swelling was known on the top of his head since birth. At the age of 4 years a tumorous swelling measuring about $8 \times 4$ cm was noted over his left mandibular angle. X-ray studies showed slight erosion of the cortical bone. The swelling over his skull had grown to a size of $5 \times 4 \times 6$ cm. Surgical biopsy from the tumor over the mandible revealed firm, dense connective tissue with few cells but rich in hypervascular capillary vessels. Bundles of connective tissue were grown in different directions. The diagnosis of a fibroma or fibrosarcoma was considered. Since, how-