A patient with multiple fibromatosis occurring at the sites of multiple cartilagenous dysplasia was described. Collagen types solubilized with pepsin from the fibromatous tissue were fractionated by a different salt concentration and analyzed by SDS-polyacrylamide gel electrophoresis which indicated that the tissue produces predominantly "short-chain" collagen. Western blotting of the subunits indicated a cross reaction with antisera of the type VI collagen. The results of rotatory shadowing electron microscopy confirmed the characteristic short chain structure.

We examined a patient with multiple fibromatosis, who has a hereditary disease with regions of multiple articular dysplasia surrounded by numerous protuberant tumors. He has a short stature (102 cm in height, 21.5 kg in body weight) and a gargoyle-like face. His articular joints were deformed by various sized tumors (2–4 cm diameter) which started as a gel-like form and grew with various elasticities, from soft to hard, or in occasion became calcified. Such abnormally excessive tumor formation at articular regions prevents his joint functioning. Tumorous gingiva prevents mastication. His mentality developed normally. Almost all laboratory data were within normal limits, except for those indicating anemia and low proteinemia due to malnutrition. X-ray and computed tomography showed that his long bones grew relatively normally but many articular bone deformed with articular dysplasia. Simply put, he has regions of articular dysplasia surrounded by multiple fibrous tumors over his whole body.

Elastic tumors were removed from his cervical region at operation weighed about 100 g. Histological examination showed that the tumor consisted of various fibrillar formation. The tissues were homogenized and suspended in a 0.4 M NaCl solution, pH 7.4, containing protein inhibitors. Collagen was extracted with limited pepsin digestion with stirring at 4°C for 24 h. The extracted collagen was then fractionated with different salt concentrations. The components were the helical form of type VI short-chain collagen and the subunits (Fig. 1). On SDS-polyacrylamide gel electrophoresis of type VI collagen, the non-reduced forms were found to corresponding to approximate Mr 140 KD, whereas the reduced subunits gave three main bands, pepsinized α1[VI], α2[VI]− and α3[VI]− chains.

The pepsinized VI collagen was predominantly found in the acidic 0.7 M NaCl supernatant in which it accounted 86% of the total collagen. The proportions of the pepsin extractable-collagen types I, III, V and VI separated from the tumorous materials were 32%, 3%, 3% and 62% to the total collagen, respectively. The antibodies raised against pepsin-extractable type VI collagen were then used to characterize the forms of antigen of the type VI collagen produced in the tumor. The antibody against the pepsinized α1[VI] subunit recognized the antigens not only for the α1[VI]−-chain but also for the pepsinized α2[VI]− and α3[VI]−-chains, moderately the monomeric domains and lower Mr of approximately 45 KD (Fig. 1). Rotatory shadowing electron microscopy of type VI collagen revealed abundant amounts of unique structures (Fig. 1).
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Fig. 1. Immunoblotting with the antibodies for the pepsinized-type [VI] antigens on SDS-polyacrylamide gel electrophoresis.

Pepsin-extracted type VI collagen prepared from the multiple fibromatosis case (c) and unreduced (a) and reduced forms (b) of human placenta.

Fig. 2. Rotatory shadowing electron microscopic features of type VI short chain collagen extracted with pepsin from the multiple fibromatosis case.

The VI short chain collagen forms rod-like structure as (a) an end to end type and (b) opened dimers connected with globular domains. The bar indicates 100 nm length.