

PSEUDOXANTHOMA ELASTICUM

An Ultrastructural Study of Oral Lesions

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Abstract. Oral lesions from one patient with pseudoxanthoma elasticum showed large amounts of thick and twisted collagen fibrils embedded in and presenting continuity with large masses of threads, while normal collagen fibrils were few. Calcification was observed inside and around elastic fibres. Some fibroblasts appeared normal, some completely destroyed.

Morphological alterations of both elastic and collagen fibres have previously been reported in pseudoxanthoma elasticum (PXE) (2, 3, 4). The present report further supports the impression of a severe collagen malformation in this disease.

MATERIAL

Biopsy specimens were taken from typical PXE lesions in the oral mucosa of one patient previously reported (4).

METHODS

The specimens were fixed in a 4% glutaraldehyde solution, buffered at pH 7.4 with phosphate salts for 1 hour at 4°C. For post-fixation, 1% osmic acid solution in 0.1 M phosphate buffer (pH 7.4) was used. After stepwise dehydration in increasing concentrations of ethanol the specimens were embedded in epoxy resin, and ultrathin sections were cut with an LKB ultramicrotome. The sections were stained by a combined technique using uranyl acetate and lead citrate and studied in a Siemens electron microscope (Elmiskop 1A) operated at 80 kV with double condensers.

RESULTS

The lesions contained large amounts of thick and twisted collagen fibrils, while the amount of normal collagen fibrils was found remarkably reduced (Fig. 1). Regularly, the twisted collagen fibrils appeared

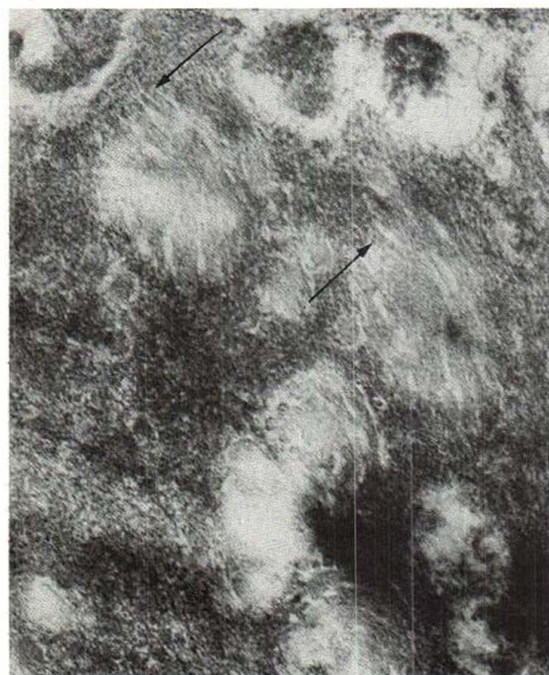
embedded in large masses of threads, and frequently there was a direct continuity between these two structures (Figs. 2 and 3). Circular holes were seen inside longitudinally and transversely sectioned thick collagen fibrils (Figs. 4 and 5). Often the threads were arranged in bundles simulating twisted or normal collagen fibrils (Fig. 5). Calcification was observed inside elastic fibres and without any relation to elastic tissue. Fibroblasts containing normal cell organelles were seen, as well as cell remnants free in the extracellular spaces (Fig. 6).

DISCUSSION

Thick and twisted collagen fibrils have previously been observed in PXE (2, 3, 4). However, in the oral mucosa of the present study they were observed in much larger quantities than in the skin lesions and artery walls previously reported (2, 3). The presumption that the large masses of unbanded threads are of collagen nature (4) is further supported by the demonstration in this study of continuity between twisted collagen fibrils and these masses. Furthermore, both show a tendency to form circular structures, appearing as round holes inside the thready masses (2), as round or oval bulgings formed by banded or unbanded threads (4), and as circular holes within twisted collagen fibrils. Furthermore, the thready material was seen to occur in bundles simulating collagen fibrils. Thus, the irregularity of collagen, originally observed as irregular reticulin fibres in the light microscope (2), appears to occur at the ultrastructural level as irregularly arranged normal or twisted collagen fibrils (2) and bundles and masses of threads. Biochemical findings of increased lysine and hydroxylysine contents in



Fig. 1. Thick and twisted collagen fibrils transversely and obliquely sectioned (black and white arrows, respectively). $\times 60\,000$.



newly synthesized collagen in PXE skin lesions might indicate an abnormality in the cross-link formation (1). The calcification inside elastic fibres and in areas without any relation to elastic tissue corresponds to the findings in PXE skin lesions (2) and in artery walls of PXE patients (3). Cystic endoplasmic reticulum seen in the fibroblasts of the skin lesions (4) was not observed in the oral mucosa. The reason might be that some of the fibroblasts appeared completely destroyed.

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Fig. 2. Twisted collagen fibrils embedded in masses of threads. Direct continuity between the thready masses and the twisted collagen fibrils is seen (arrows). $\times 60\,000$.

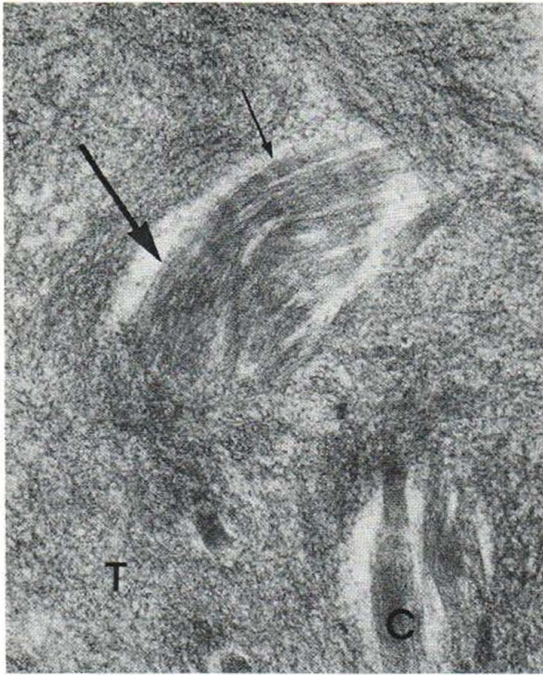


Fig. 3. Twisted collagen fibril showing periodicity (*thin arrow*) or appearing as a thready material (*thick arrow*). *T*, Thready material; *C*, collagen fibril. $\times 60\ 000$.

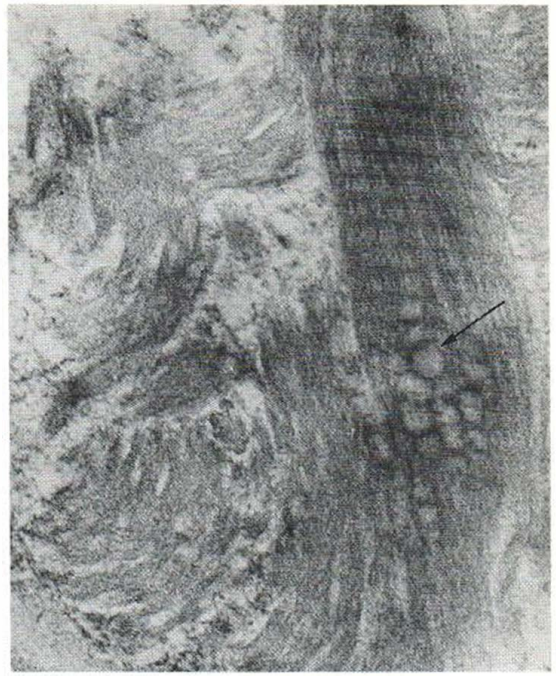


Fig. 4. Circular holes (*arrow*) within a collagen fibril. $\times 60\ 000$.

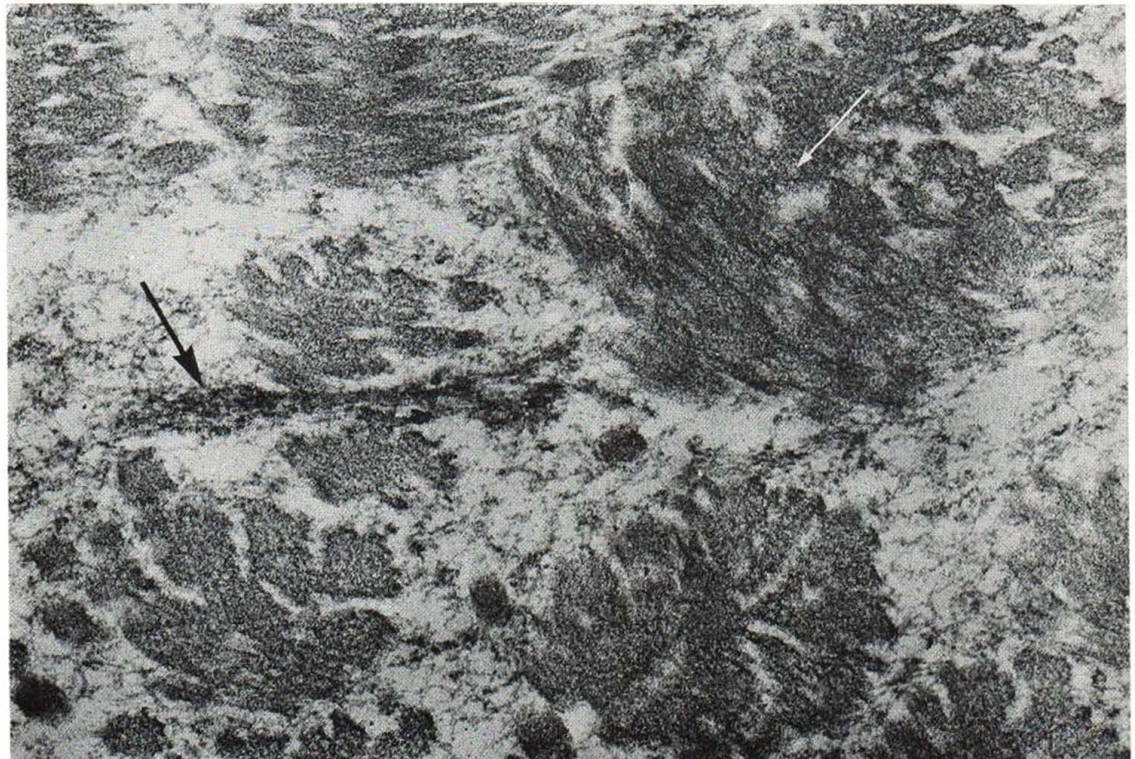


Fig. 5. Bundles of threads (*black arrow*) simulating a collagen fibril are surrounded by twisted collagen fibrils. Circular

holes (*white arrow*) are seen within the collagen fibrils. $\times 60\ 000$.

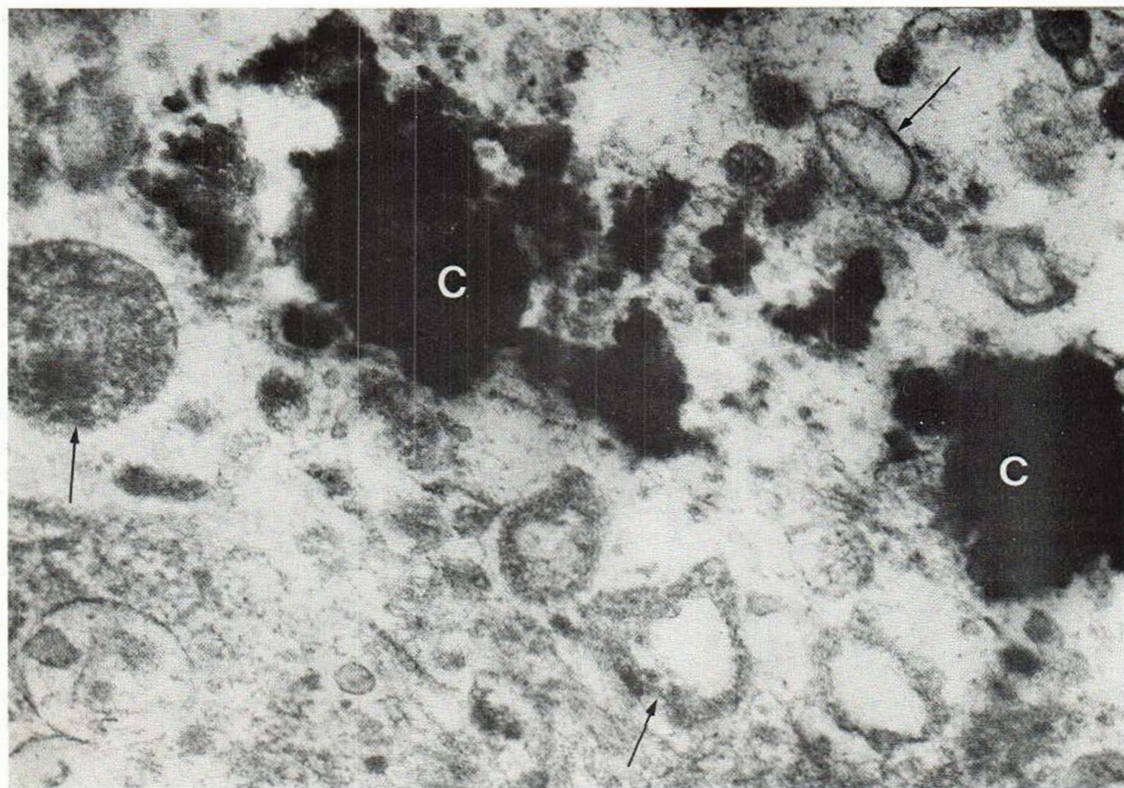


Fig. 6. Cell-remnants (arrows) are seen in the extracellular spaces. C. Calcification. $\times 60\ 000$.

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