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Hereditary gingival fibromatosis with hemophilia B

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Summary

This work presents a case report of a generalized form of hereditary gingival fibromatosis with hemophilia B as an accompanying disease. In the family of proband, consisting of 28 members, fibromatosis was present in 9 (4 males and 5 females). The pedigree analysis confirmed that gingival fibromatosis was transmited through three generations as an autosomal dominant trait. Neither proband, nor any other family member, showed other abnormalities. Blood coagulation tests reveald hemophilia B (Christmas disease) in the proband. The coagulogram showed prolonged kaolin cephalin time (50 seconds) and low concentration of factor IX (F IX 18%). The case report suggests that hemophilia B should be included in the list of diseases associated with gingival fibromatosis.

Key words: gingival fibromatosis, hemophilia B

Hereditary gingival fibromatosis manifests as an isolated trait, accompanied by other abnormalities or disease, or as a symptom of a specific syndrome. The most common clinical abnormalities associated with gingival fibromatosis are hypertrichosis, epilepsy, mental retardation, and defects of the eye, ear, nose, skeleton and nails (Fletcher[1], Gorlin et al.[2], Jorgenson and Cocker[3]). Isolated gingival fibromatosis without other abnormalities is considered a special entity which differs from the fibromatosis accompanied by hypertrichosis, epilepsy or mental retardation (Cohen[4]). When only gingival fibromatosis develops, it usually means that the disease was inherited as an autosomal dominant trait (Fletcher[1], Savara[5], Zackin et al.[6], Becker et al.[7]), even though the autosomal recessive transmission of the disease has also been described (Gorlin et al.[2], Jorgenson and Cocker[3]). Raeste et al.[8] have reported on the autosomal dominant transmission of fibromatosis with variable penetration.

According to the localization, Jorgenson and Cocker[3] differentiate two types of isolated gingival fibromatosis: the generalized form involving all gingiva of both jaws, and the localized or focal form involving only part of the gingiva

(usually the palatine surface of the maxillar tubera or lingual surface of the mandibular ridge).

In an article on gingival fibromatosis Fletcher[1] mentions disturbed blood coagulation – among other symptoms which might accompany fibromatosis – but does not precisely define the type of disease. Hemophilia B with gingival fibromatosis has not been previously reported in literature.

This article presents a case report of a generalized from of hereditary gingival fibromatosis with hemophilia B as an accompanying disease.

FAMILY REPORT

In the family of proband III-3, consisting of 28 members, fibromatosis was present in 9 (4 males and 5 females). The pedigree analysis confirmed that gingival fibromatosis was transmitted through three generations as an autosomal dominant trait (Figure 1). The father of the proband (II-3) showed a marked generalized form of fibromatosis (Figure 2). Hyperplastic gingiva in the maxilla and mandible almost entirely covered the occlusal surfaces and incisor edges. Neither examinee II-3, nor any other family member, showed other abnormalities.

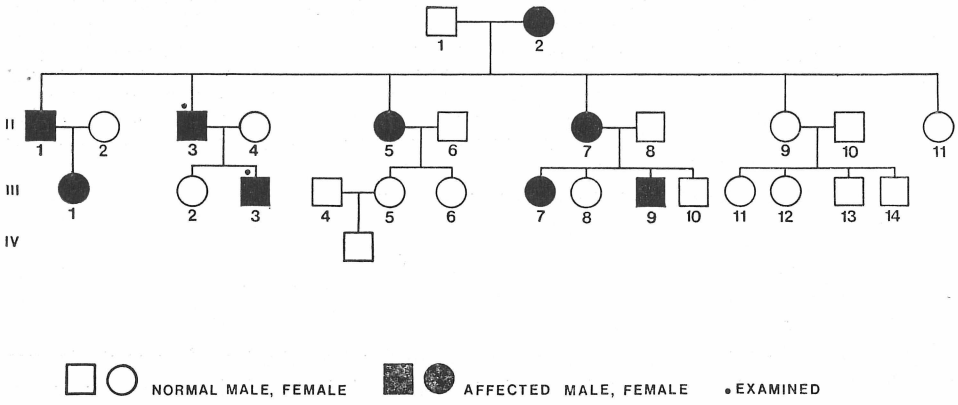

Figure 1. The family pedigree shows autosomal dominant mode of inheritance

CASE REPORT

Proband III-3 was 16 qears old. Aside from generalized hyperplastic gingiva no other abnormalities were present. Gingiva covered half of the dental crown in the lateral teth area and was less pronounced in the frontal region of the mandible and maxilla (Figure 3). Malocclusion Class II, division two, with a markedly deep overlap of frontal teeth was diagnosed on the basis of the clinical examination and analysis of the LL X-ray. The maxilliary central incisors were typically rotated and protruding, mandibular incisors retroverted, and the lower canine teeth typically rotated. The bite was deep and there was contact between the mandibular and maxillary incisors and the gingiva.

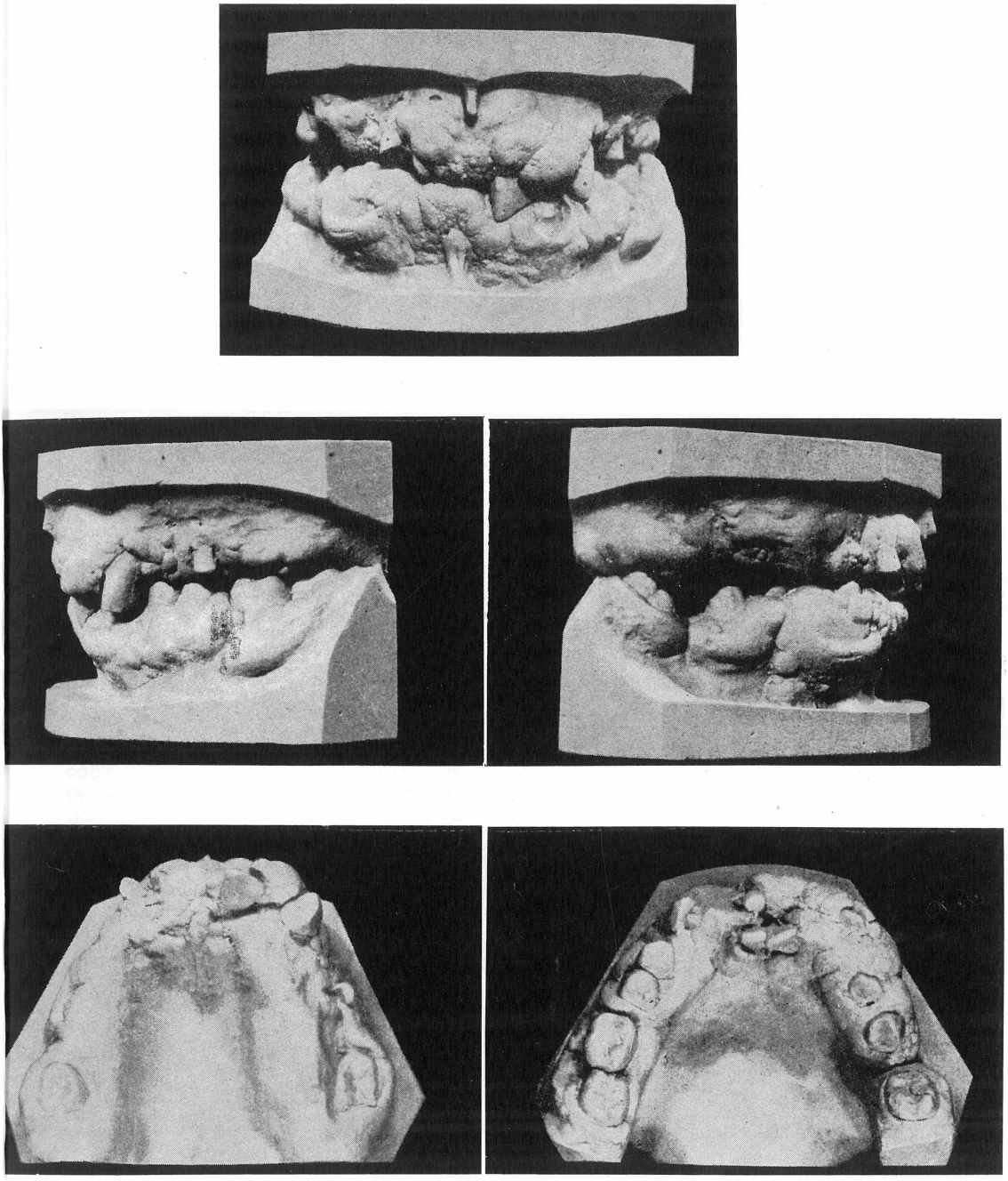

Figure 2. Extensive gingival fibromatosis in proband's father (II-3)

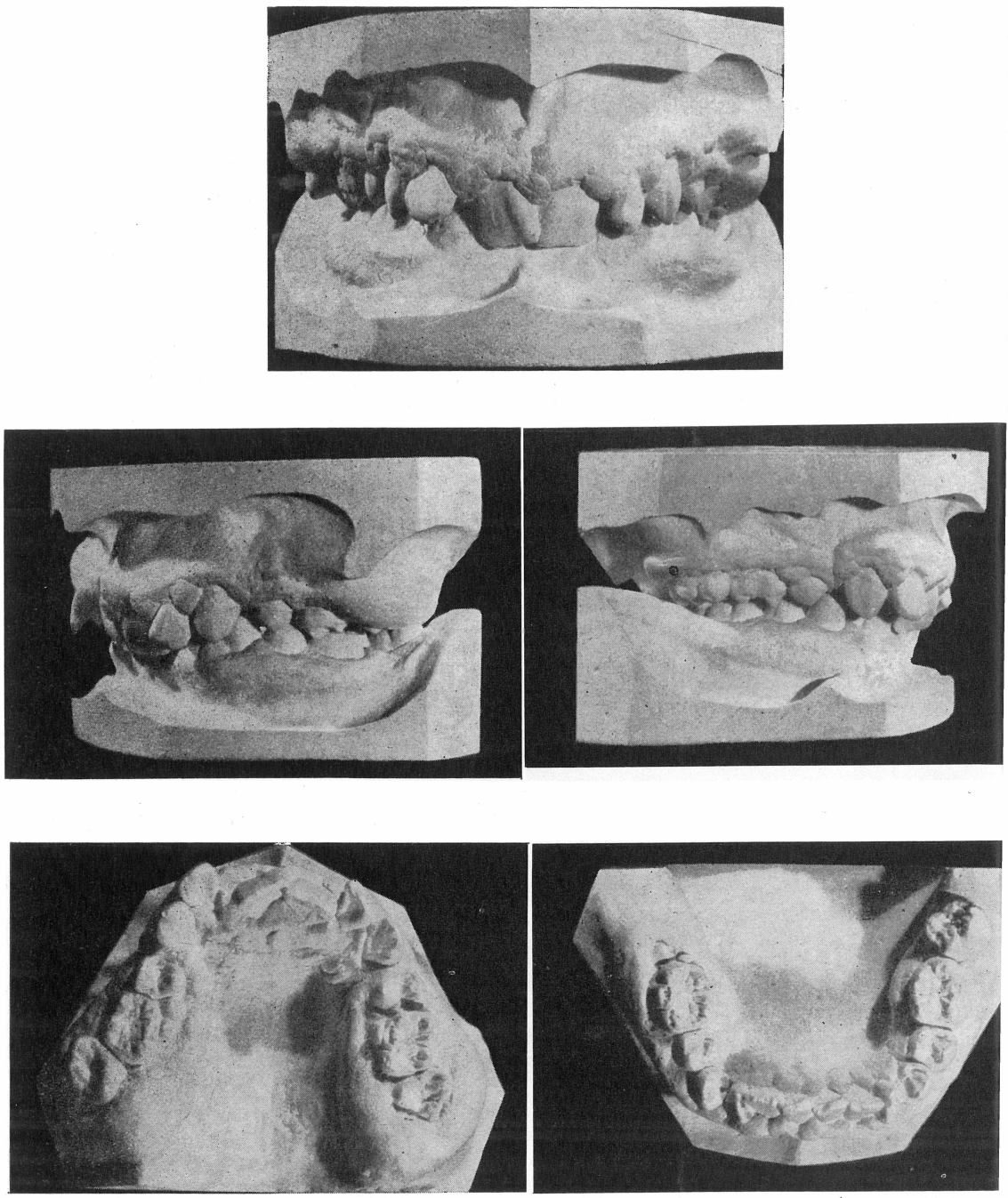

Figure 3. Gingival fibromatosis in the proband (III-3) two years after gingivectomy

Table
VALUES OF THE BLOOD COAGULATION TESTS

Platelet count	$117 \times 10^9/1$
Bleding time	0.15 ks
Clotting time	0.55 ks
Prothrombin time	11.5 sec. > 1.0
Recalcified plasma clotting time (Howell)	0.179 ks
Prothrombin consumption	0.070 ks
Thromboplastin generation time (TGT)	2 min. 14 sec. 4 min. 10 sec. 6 min. 9 sec.
Partial thromboplastin time (PTT) (Kaolin-cephalin time)	50 sec.
Thrombin time	34 sec.
Factor VIII	0.36
Factor IX	0.18
Factor XIII	> 1800 sec.

Anamnestic data indicated that the disease began with second dentition. The proband took neither hydantoin preparations nor any other kind of drug which could cause gingival enlargement. Figure 3 presents the condition of gingiva two years after gingivectomy, that is recurrence of the previous stage. Blood coagulation tests performed before gingivectomy, revealed hemophilia B (Christmas disease) in the proband. The coagulogram (Table 1) showed prolonged kaolin cephalin time (50 seconds) and low concentration of factor IX (F IX 18%).

DISCUSSION

Hereditary gingival fibromatosis usually develops at the beginning of permanent dentition (Fletcher[1], Gorlin et al.[2], Jorgenson and Cocker[3]), although it can occur in the primary dentition (Fletcher[1], Savara[5], Henefer and Kay[9]). Histologically fibromatosis is characterized by hyperplasia of the connective tissue and thickened epithelium (Henefer and Kay[9], Aurer et al.[10]). Henefer and Kay[9] report that fibromatosis histologically resemble keloid.

Gingival fibromatosis is a leading symptom in the following syndromes: Murray-Puretić-Drescher, Rutherford, Laband, Cross, Jones and Byars-Jurkiewitz (Cohen[4]). The Murray-Puretić-Drescher syndrome shows gingival fibromatosis and multiple hyaline fibromas. Rutherford syndrome manifests as gingival fibromatosis with corneal dystrophy. The Laband syndrome shows gingival fibromatosis with hepatosplenomegaly, defects of the ear, nose, bone and nails. The Cross syndrome includes gingival fibromatosis with microphathalmia, mental retardation, athetosis and hypopigmentation. Gingival fibromatosis and sensorineural deafness characterize the Jones syndrome. The Byars-Jurkiewicz syndrome consists of gingival fibromatosis, hypertrichosis and giant fibroadenomas of breast.

Literature contains relatively few articles on pedigree patients with fibromatosis (Fletcher[1]) and the genetic mechanism responsible for the isolated gingival fibromatosis is not completely understood (Jorgenson and Cocker[3]). This indicates the need for analyzing the hereditary mechanism in a larger number of examinees with fibromatosis to shed light on its genetic transmission.

The pedigree analysis in the present study revealed an autosomal dominant inheritance of fibromatosis which was transmitted through three generations. Examinee III-3 was the only family member who had hemophilia B together with fibromatosis. Since hemophilia B is transmitted as an X-linked recessive trait, the proband could have only inherited the gene from his mother. The disease is never transmitted from the father to the son, and develops exclusively in males. Males with hemophilia B transmit the gene for the disease to half of their daughters who are then carriers. Female carriers transmit the disease to half of their sons, and half of their daughters will be latent carriers of the gene without the disease manifesting. These theoretical genetic viewpoints have practical implications in genetic counselling which should be an integral part of patient menagement (Lucas and Prescott[11]). Not one of our proband's first generation offspring can have fibromatosis with hemophilia. As an autosomal dominant trait, fibromatosis wil be transmitted to about half of the offspring regardless of the sex. The proband cannot transmit hemophilia to his sons, but can transmit the gene for the disease to half of the daughters who then become carriers. In the second generation it is possible that a woman with fibromatosis and a carrier of the gene for hemophilia will transmit both of these genes to her son, so that both diseases manifests together. If the proband does not have a daughter with fibromatosis who is the carrier of the gene for hemophilia, these two diseases will not manifest together in future generations.

This case report of hereditary gingival fibromatosis with hemophilia B sugests that hemophilia B should be included in the list of diseases associated with fibromatosis.

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Sažetak

NASLJEDNA FIBROMATOZA GINGIVE S HEMOFILIJOM B

Rad predstavlja prikaz slučaja generalizirane forme nasljedne fibromatoze gingive s hemofilijom B kao pratećom bolešću. U obitelji probanda, koja se sastojala od 28 članova, fibromatoza je bila prisutna u 9 (4 muška i 5 ženskih). Genealoškom analizom utvrđeno je da se fibromatoza gingive prenosila kroz tri generacije kao autosomno dominantno svojstvo. Ni proband, niti ostali članovi obitelji nisu pokazivali bilo kakvih drugih abnormalnosti. Testovima koagulacije krvi u probanda je utvrđena hemofilija B. Koagulogram je pokazivao produženo kaolin cefalinsko vrijeme (50 sekundi) i nisku koncentraciju faktora IX (F IX 18%). Prikaz slučaja ukazuje da među bolesti koje mogu doći s fibromatozom gingive treba uključiti i hemofiliju B.

Ključne riječi: fibromatoza gingive, hemofilija B