PAPILLON LEFEVRE SYNDROME

(Case Report with Cytogenetic Study)

by

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The syndrome of hyperkeratosis of palms and soles, with early loss of deciduous and permanent teeth due to premature destruction of the periodontal tissue, was first described by Papillon and Lefevre in 1924. Gorlin et al (1964) published an extensive analysis of 46 well documented cases. A few more cases of the syndrome were reported aftrewards (Velou et al, 1969). We did not come across any cytogenetic studies in the syndrome in the available literature. As such, the report of a case of the syndrome with cytogentic study may be of interest.

CASE REPORT

M., 20 years old male, reported with complaints of scaling of skin of palms and soles and loss of teeth. The patient started having scaling of skin of palms and soles during early childhood and the same was persisting. The severity of the lesions used to become more during winter. Five years prior, his teeth started falling off one after the other and in a period of two years he lost all his teeth. The falling off of each tooth was preceded by swelling of gum and formation of a pocket between it and the gum. As the parents of the patient had expired, the fate of the milk teeth could not be ascertained and the history of the consanguinity of the parents could not be elicited.

Examination of the oral cavity showed absence af all the teeth with healthy gum margins. Palms (Fig. 1) and soles showed marked thickening with yellowish colouration. Dorsum of hands and feet (Fig.2) showed thickening, desquamation and erythema. Front of knees (Fig. 2) and back of elbows showed similar lesions. The finger nails showed transverse grooves. Systemic examination did not reveal any abnormality.

Routine examination of urine, stools and blood showed no significant abnormality. Serum calcium, phosphorus and alkaline phosphatase were within normal limits. X-ray of the skull showed absence af any erupted or developing tooth and no evidence of intracranial calcification (Fig. 3). X-ray of hands showed marked diminution of density of bones. Skin biopsy taken from the front of the knees showed histological picture that is consistent with hyperkeratosis. Cytogenetic study from bone marrow showed a normal karyotype without any numerical or structural abnormality of chromosomes.

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DISCUSSION

Papilon-Lefevre syndrome is constituted by the palmoplantar hyperkeratosis and periodontosis leading to loss of teeth. This syndrme manifests itself usually between the age of one and four years. The syndrome started during early childhood in our patient. The hyperkeratosis is transgradient i. e., the lesions extend over the wrist, ankles and dorsa of the hands and feet. Sometimes, the front of knees and back of elbows show the hyperkeratotic patches. In our patient all the sites were affected.

The dental changes are very predominant, The deciduous teeth erupt normally but they fall off at the age of 4 or 5 years. In our case, no definite history could be obtained about milk teeth. The eruption of permanent teeth is also normal but they are expelled by the age of 16 years. In our case, the permanent teeth were lost by the age of 17 years. Gorlin et al. (1964) stated that the third molars escape exfoliation. Haim and Munk (1965) recorded that all the teeth including the third molars were markedly mobile in their first patient aged 17-years. Velou et al. (1969) reported probable involvement of the third molars in their patient. Our patient showed absence of all teeth clinically and no evidence of any developing tooth radiologically. As such it may be concluded that the third molars were also exfoliated in our patient.

Gorlin et al. (1964) suggested that calcification of the dura may be the third component of the syndrome. There was no radiological evidence of calcification of the dura in our patient.

Absence of a gross numerical or structural abnormality of chromosomes in the present case indicate that the disease is inherited at subchromosomal level of genes. Occurrence of the disease in either sex rules out a sex linked inheritance. Therefore, with the pattern of familial affections of the proband as reported (Gorlin, 1964), the inheritance is apparently autosomal recessive in nature.

SUMMARY

The case of a 20 years old male with Papillon - Lefevre Syndrome is reported. The patient had lost all the teeth at the age of 17 years and there was no evidence of any developing tooth on radiological examination. Cytogenetic study of bone-marrow showed a normal karyotype without any numerical or structural abnomality of chromosomes.

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