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RESEARCH ARTICLE

Oral findings in Zinsser-Engman-Cole syndrome: a case report.

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Abstract

Zinsser Cole Engmansyndrome also known as dyskératosiscongenita (DC) is a very rare genetic affection characterized by the association ofreticular hyper pigmentation, nail dystrophy and leucoplakia of the mucous membranes. We report the case of a 14 years old child already diagnosed with DC and which was complaining about extensive caries, and teeth loss. Clinical examination revealed a poor oral hygiene, extensive caries and periodontal disease. DC is a inherited bone marrow failure characterized by excessively short telomeres in highly proliferative tissues. Its prevalence is estimated at one in a million.DC has many clinical features and oral manifastations. It have a poor prognosis and its current treatment remain non satisfactory. DC needs a multidisciplinary approach and a regular follow-up.

Introduction:-

Zinsser Cole Engmansyndrome also known as dyskératosiscongenita (DC) is a very rare type of ectodermal dysplasia. It's a multi-system disorder whose symptoms appear in childhood andleads to death between 16 and 50 years.

DC occurs mainly in young male with a male to female ratio of 13/1 and mostly appears between 5 and 12 years[1–2].

This genetic affection is typically characterized by the association of a traditional dermatological triad: Reticular hyper pigmentation, nail dystrophy andleucoplakia of the mucous membranes. The medullary insufficiency associated to this disease in 85, 5% is responsible for opportunistic infections[3].

Case report:-

A14 years old male teenager was admitted to both departments of pediatric of Casablanca University following extensive caries, teeth loss and toothache.

Medical history revealed that he's diagnosed with dyskeratosis congenital after genetic and telomere length testing providedthe identification of telomere disorder.

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He was lately admitted for chronic mucocutaneous candidiasis and an abscess of the right buttock. The patient suffers also from growth retardation. His skin was thin, dry and wrinkled in some areas and he showed nail dystrophy of the hands and feet. (Fig 1, 2 and 3).

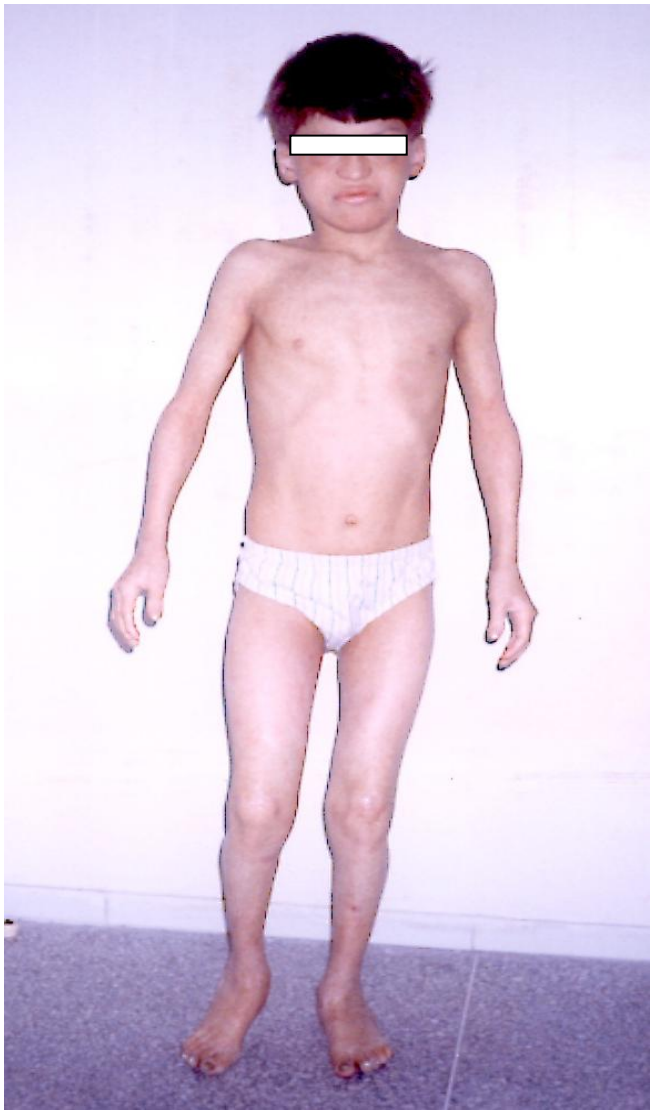

Figure 1: Growth retardation and thin skin

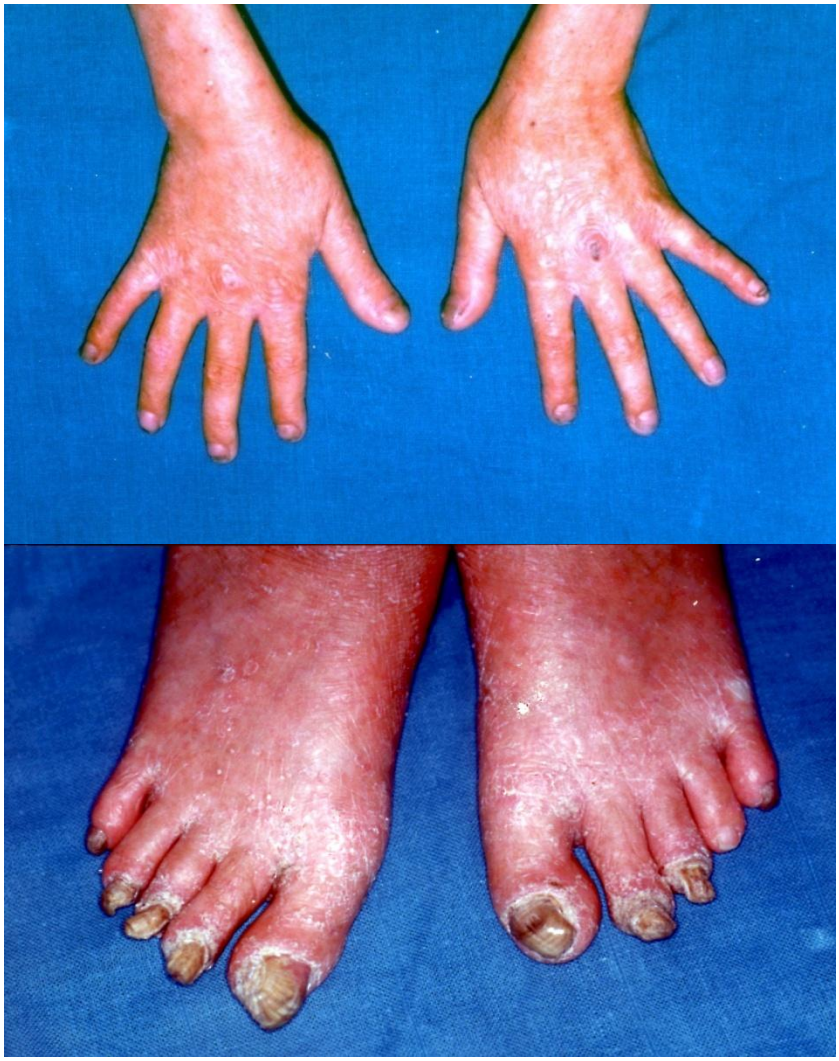

Figure 2 and 3 : Nail dystrophy of the hand and feet

His obstetrical antecedents were without particularities and his birth weight was 4 kg without signs of suffering of any neonatal infection. His vaccination was carried out correctly according to the national immunization program, without any complication.

Family history revealed that he was born from a consanguineous marriage of second degree and that his brother which was also diagnosed with DC died 3 years ago from a lethal infection.

Extra oral examination showed facial deformity with reticular hyperpigmentation of his face and neck. The patient was pale but vital signs were within normal limits (Fig 4).

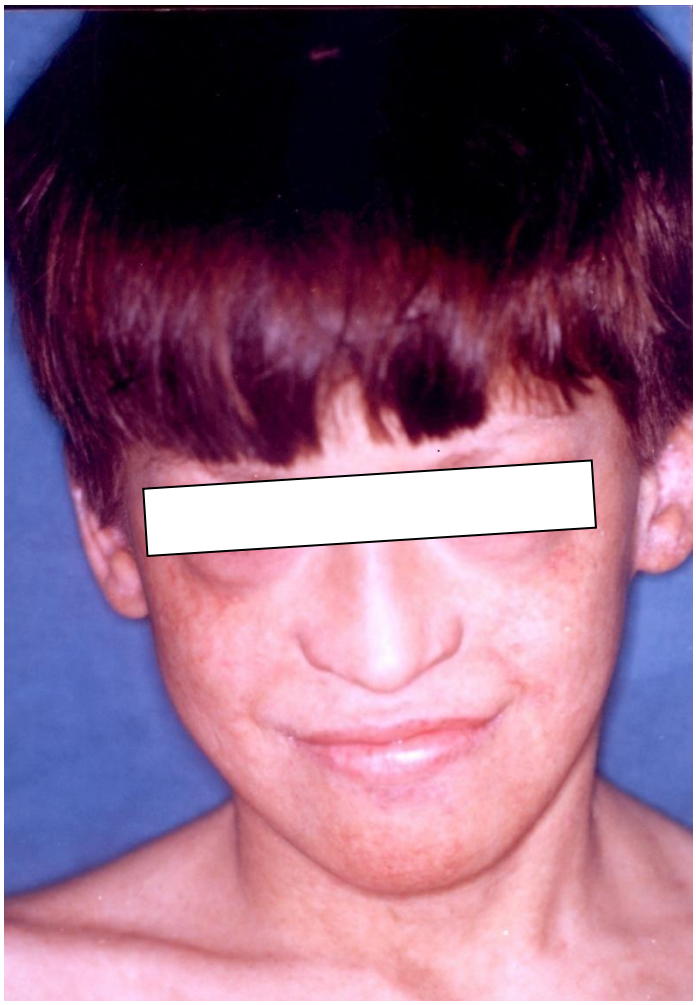

Figure 4 : Facial deformity with reticular skin hyperpigmentation

Intra oral examination revealed poor oral hygiene, leukoplakia on the tongue and the mandible gingival mucosa, extensive caries of all his first permanent molars. The patient suffered from periodontal disease which led to many early teeth loss (Fig 5).

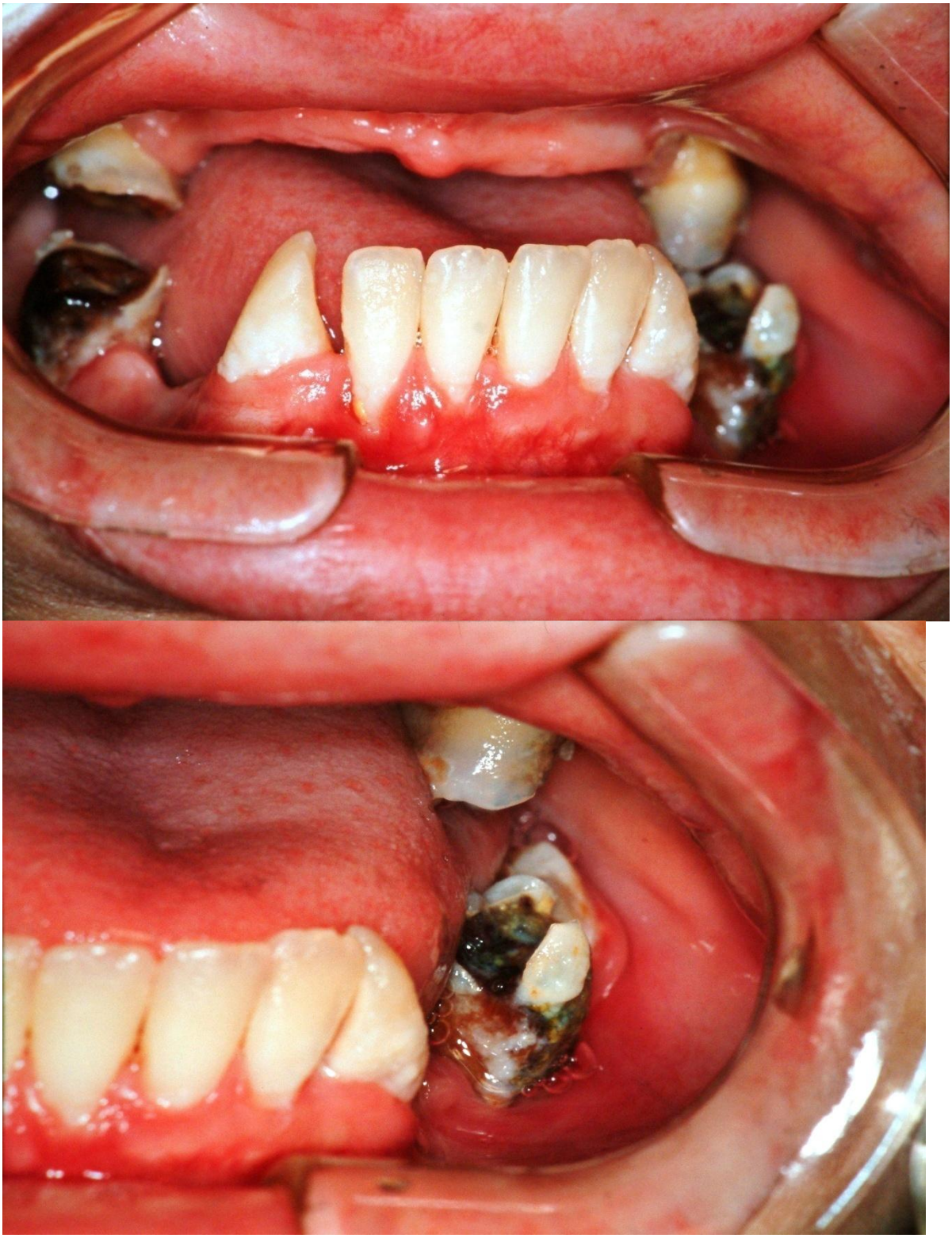

Figure 5: Extensive caries of the first permanent and leukoplakia in the mandible gingiva

Radiographic examination, showed a bone radiolucency image around the permanent molars and the two central incisors (Fig 6).

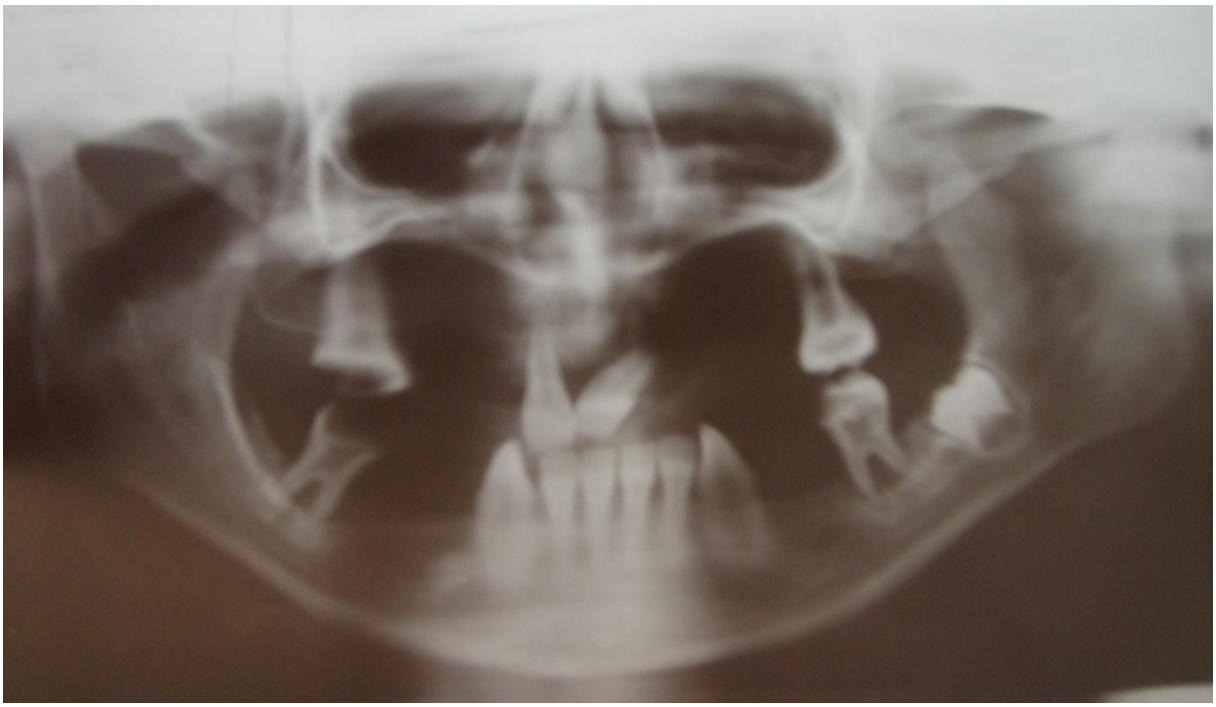

Figure 6: Bone radiolucency image around the permanent molars and the two central incisors

Periodontal treatment was performed and the first molars with the two incisors were removed. A dental prosthesis has been made and the child was very happy to find his lost smile.

Discussion:-

DC is a rare inherited bone marrow failure characterized by excessively short telomeres in highly proliferative tissues. Its prevalence is estimated at one in a million [2-3].

Five genes have been identified, all encoding for components of telomerase andinvolved in maintaining telomere length: DKC1 (locus Xq28, sex-linkedtransmission), TERC (locus 3q21-q28, autosomal dominant), TERT (locus 5p15.33, autosomal dominant) ,NOP10 (locus 15q14- q15, autosomal recessive) and TINF2 (locus 14q12, autosomaldominant)[4].

DC is a very polymorphous disease at the clinical level, with several modes of inheritance. Several clinical symptoms of the disease can appear after a latency period. These features make DC particularly difficult to diagnose.

The main clinical features of this multi-system bone marrow failure disease are mucocutaneous abnormalities (cutaneous hyperpigmentation, dystrophy of the nails, leukoplakia of the oral mucosa), haematopathies (pancytopenia 50%) and an increase predisposition to cancer.

Other abnormalities in other systems may also be associated: pulmonary disease fibrosis, abnormalities of pulmonary vasculature, eye defects like conjunctivitis, blepharitides, pterygium andepiphora, mental retardation, short stature, hair loss/grey hair/sparse eyelashes, eye defects and deafness [5].

The oral manifestations described in the literature are: Hypodontia, Short blunted roots, Extensive caries lesions, Delayed eruption, Taurodontism, Gingival inflammation with oedema, Alveolar bone loss, Periodontitis, Thin enamel gingival recession,Smooth atrophic tongue mucosa, Dry mouth, Angular cheilitis and Leukoplakia[3–6–7].
General hygiene recommendations include brushing teethtwo to three times a day with fluoridated toothpaste and flossingonce a day at a minimum to help prevent tooth decay. Somedentists recommend using prescription strength fluoridetoothpaste or antibacterial mouth rinse to aid in reducing oraldisease.

Biannual dental checkups and cleanings arerecommended to monitor for the presence of oral pathology andprevent the development of significant dental decay and gumdisease.
Precautions during routine dental treatment may benecessary in the presence of low platelet counts and white bloodcell levels.

Access to quality dental care for medically compromisedpatients may be challenging outside of large urban centers.

The risk of squamous cell carcinoma is high in individual with DC estimated approximately 35%, with a peak in the third decade. Many other malignant tumors and hemopathiesl have also been reported like bronchial, colon, larynx, esophagus, stomach and pancreas carcinomas, Hodgkin's disease and leukemia[3].

Malignant degeneration of these leucoplakia is not uncommon and it’s expected after 10 to 15 years of evolution, hence the need for periodic reviews to detect signs of transformation.
DC has a poor prognosis, the current treatments remain non satisfactory.
Mortality is often associated to severe pancytopenia, development of malignancy and other multisystem complications of the disease [2].

Conclusion:-

DC needs a multidisciplinary approach: genetic, pediatric, hematologicand a special dental care. Regular follow-up of the patient is essential especially with the high possibility of malignant changes within oral and other mucosal sites.

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