Brief Report
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A Report on Papillon Lefevre Syndrome

Wang Weiguang*

University of Wolverhampton, Research Institute in Healthcare Science, UK

Brief Report

Papillon-Lefevre syndrome (PALS) is a rare genetic illness that causes skin thickening (keratoderma) on the palms of the hands and soles of the feet (palmoplantar), periodontitis (inflammation of the tissues around the teeth), and early (premature) tooth loss. The skin of the palms and soles is frequently dry and scaly in patches (patches). Infections are a possibility in certain regions. Gum shrinkage is a side effect of periodontitis. Symptoms commonly appear between the ages of one and five. By the age of four, most children have lost their primary teeth, and by the age of fourteen, they have lost their secondary teeth. PALS are caused by mutations in the CTSC gene and is inherited in an autosomal recessive manner, meaning that a kid must have a mutation in both copies of their CTSC gene to be affected. PALS are suspected in children who have thick skin patches, gum inflammation, and tooth loss at an early age. To validate the diagnosis, genetic testing is used. Medications to assist lower the risk of infection in the skin and dentures to replace missing teeth are two treatment possibilities. A bone transplant is another therapy option that can help save teeth before they are gone. If your kid has been diagnosed with PALS, discuss all treatment options with their doctor. Additional help may be available through support groups.

Periodontitis and palmoplantar keratoderma are two symptoms of PLS. Most primary teeth are lost by the age of four, and most permanent teeth by the age of fourteen, due to significant periodontal deterioration. In the first few years of life, hyperkeratosis of the palms and soles of the feet appears. Periodontal destruction occurs nearly immediately following the eruption of the last molar tooth. Teeth are engaged essentially in the same sequence as they erupt. The autosomal recessive genetic condition Papillon-Lefèvre syndrome has an incidence of one to four instances per million. Palmoplantar hyperkeratosis can be widespread or localised in this condition. It has neither a gender nor a racial preference. Papillon-Lefèvre syndrome's aetiology and pathogenesis are currently unknown.

Papillon-Lefèvre syndrome, on the other hand, is thought to be caused by a genetic defect on chromosome 11 that codes for cathepsin C, a lysosomal protease found primarily in epithelial regions such as the palms, soles of feet, knees, and keratinized oral mucosa, which is the most commonly, affected areas in Papillon-Lefèvre syndrome. PLS is caused by mutations in the cathepsin C gene (CTSC), which is found on human chromosome 11q14.1-q14.3. The condition is inherited as an autosomal recessive trait. This signifies that the disorder-causing gene is found on an autosome and that two copies of the defective gene are necessary to be born with the disorder. Both parents of a person with an autosomal recessive disorder possess one copy of the defective gene, although they normally have no signs or symptoms of the disorder.

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***Address for Correspondence:** Weiguang Wang, University of Wolverhampton, Research Institute in Healthcare Science, UK, Email: w.wang2@wlv.ac.uk*

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