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Hyaline fibromatosis syndrome

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Hyaline fibromatosis is a rare genetic disorder inherited in an autosomal recessive pattern. It is a disorder of the connective tissue observed due to mutations in the anthrax toxin receptor-2 (ANTXR2) gene, also known as capillary morphogenesis gene-2 (CMG2). The ANTXR2 gene is known to be a transmembrane protein majorly involved in tissue remodelling and adhesion of the extracellular matrix (ECM). The disorder manifests by the accumulation of amorphous hyalinised fibrous tissue mainly in the papillary dermis along with other tissues of the body. Some of the symptoms of this disease include decreased joint mobility, gingival hypertrophy, skin lesions, perianal masses etc. Hyaline fibromatosis syndrome is a collective term for infantile systemic hyalinosis (ISH) and juvenile hyaline fibromatosis (JHF). ISH was the term used for the severe form of the disease manifesting early in childhood with significant risks of morbidity and mortality. It is known to be caused due to insertion or deletions of the ANTXRs gene leading to the translational frameshift mutations. JHF is a milder form characterised by in-frame and missense mutations in the cytoplasmic domain of the ANTXR2 protein manifesting much later in childhood with a lesser risk of mortality. Since both JHF and ISH are caused by mutations in the ANTXR2 gene, recently the nomenclature has been unified. A permanent cure is not available currently for this disorder and only palliative treatment in the form of surgery and painkillers is available. The occurrence of this disorder is more common in Turkey, the Middle East, India and Morocco due to the prevalence of consanguineous marriages as a part of the tradition. Molecular genetic testing and prenatal testing should be advised for couples with increased risks to prevent further complications and contamination of the gene pool.

Keywords: Hyaline Fibromatosis Syndrome, Infantile Systemic Hyalinosis, Juvenile Hyaline Fibromatosis, ANTXR2, Molecular Genetic Testing

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