Hereditary Progressive Arthro-ophthalmopathy (Stickler Syndrome): A Clinical Analysis and Search for Linkage

by

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This research project characterizes the clinical stigmata of Stickler syndrome and attempts to identify a chromosomal linkage relationship between the gene responsible for Stickler syndrome and several predetermined marker loci. The procedure was to interview and examine a very large family of 96 people ascertained from an affected proband, and to collect blood, saliva and urine samples on selected individuals. These samples were genotyped and the marker loci were evaluated. The LIPED computer program was employed to analyse the data to establish a positive or negate a chromosomal relationship. The data obtained from the examination and linkage analysis were organized in tables and a detailed family pedigree was constructed. Although a positive linkage relationship could not be established, linkage was ruled out at four marker loci. A great deal of clinical information was obtained from the evaluation of this family. A large amount of intrafamilial variation was observed and trends of specific symptom complexes were noted in different sibships. It is evident that more data need be collected on additional families to elucidate not only the linkage data, but also to further delineate the clinical features of Stickler syndrome.