

Carpenter syndrome belongs to a group of rare genetic disorders known as “acrocephalopolysyndactyly” (ACPS) disorders. All forms of ACPS are characterized by premature closure of the fibrous joints (cranial sutures) between certain bones of the skull (craniosynostosis), causing the top of the head to appear pointed (acrocephaly); webbing or fusion (syndactyly) of certain fingers or toes (digits); and/or more than the normal number of digits (polydactyly). Carpenter syndrome is also known as ACPS type II. Carpenter syndrome appears to affect males and females in relatively equal numbers. More than 70 cases of the disorder have been recorded. In 10 patients that had sequence analysis for the disease causing gene, homozygosity (two copies) for the same nonsense mutation, (a change in the DNA that causes a change in the protein) was found. This is indicative of a founder effect in patients of northern European descent, which means that a high prevalence of a genetic disorder in an isolated or inbred population is due to the fact that many members of the population are derived from a common ancestor who had the disease causing mutation.