

Alagille Syndrome (???????)– Figure (4): Pedigree chart showing autosomal dominant inheritance pattern. The hallmark of the syndrome is bile duct paucity, leading to cholestasis (reduction in bile flow), alongside cardiac anomalies and distinct facial features. --- ### Conclusion Alagille Syndrome is a genetically inherited disorder with a complex clinical presentation, primarily due to mutations in the JAG1 or NOTCH2 genes. DNA Investigated Using BLASTn The nucleotide sequence of the JAG1 gene is compared using BLASTn to detect point mutations, deletions, or duplications associated with the syndrome. Protein Analysis Using EXPASY The protein sequence from the JAG1 gene is analyzed using EXPASY to determine molecular weight, isoelectric point, and structural properties. (???????) --- ### Introduction Alagille Syndrome is a rare genetic disorder affecting multiple organ systems, including the liver, heart, kidneys, and eyes. Biopsy and DNA Extraction Procedures Liver biopsy and DNA samples from affected individuals are used to analyze genetic material for mutations in specific genes. Mode of Inheritance Alagille Syndrome is inherited in an autosomal dominant manner. Patient's Clinical Features Photographs of an affected individual show typical physical characteristics, including skeletal abnormalities and jaundice. – Figure (5): Photos of a patient's clinical manifestations (e.g., butterfly vertebrae, facial features). – Primary Symptoms: – Chronic liver disease due to bile duct abnormalities. – Characteristic facial features (broad forehead, pointed chin). – Figure (3): Comparative analysis of normal and mutated JAG1 protein. Early diagnosis and a multidisciplinary approach to treatment can significantly improve patient outcomes. – Heart defects, such as pulmonary artery stenosis. – Skeletal abnormalities, including butterfly vertebrae. ##### 2. ##### 3. ##### 4. ##### 5