

Comparison of Four Genetic Disorders

Feature	Cutis Laxa	Supravalvular Aortic Stenosis (SVAS)	Williams Syndrome	Marfan Syndrome
Main Characteristics	Loose, sagging skin; increased risk of aortic aneurysm; emphysema	Narrowing of the aorta	Intellectual disability; characteristic facial features; cardiovascular defects	Unusually tall stature; long limbs and fingers; heart and aortic defects
Genetic Cause	Mutations in various genes (ATP6V0A2, ATP7A, EFEMP2, ELN, FBLN5)	Mutations in the ELN gene	Deletion of a region on chromosome 7, including the ELN gene	Mutation in the FBN1 gene
Inheritance Pattern	Autosomal dominant, autosomal recessive, X-linked	Autosomal dominant	Autosomal dominant	Autosomal dominant
Gene Function	Mostly involved in elastic fiber formation and	Elastin production	Influences various protein production	Produces fibrillin-1, essential for connective tissue

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	function		and function	formation
Additional Notes	X-linked form known as occipital horn syndrome	Often mild compared to other forms of cutis laxa	Associated with characteristic personality traits	Affects various parts of the body, including lungs, eyes, and skeleton

Please note: This table is for informational purposes only and should not be taken as medical advice. Always consult with a qualified healthcare professional for diagnosis and treatment of any medical condition.