

GreenMCMeds Disease Screening Genes

Disease	Summary	Blood	Breathing	Cancer	Delays	Hearing	Heart	Infection	Kidneys	Liver	Muscles	Seizures	Skeletal	Vision
Alpha Thalassemia	Alpha Thalassemia affects proper formation of alpha-globin, a subunit of hemoglobin, which is a key component to bringing oxygen to body tissue.	•								•			•	
Aortic Dysfunctions	Aortic Dysfunctions disrupt the heart from pumping blood with enough pressure to reach all parts of the body, in order to maintain oxygenated tissue.						•							
Beta Thalassemia	Beta Thalassemia affects proper formation of beta-globin, a subunit of hemoglobin, which is a key component to bringing oxygen to body tissue.	•									•		•	
Bloom Syndrome	Bloom Syndrome disrupts proteins that preserve the integrity of DNA, as well as catalyze key reactions that are crucial for DNA unwinding.			•	•									
Canavan Disease	Canavan Disease causes defects in brain enzymes which interfere with the development of the neuronal myelin sheath.				•						•	•		
Cystic Fibrosis	Cystic Fibrosis disturbs proper electric potential by preventing chloride ion transport in the lungs, thickening mucus and clogging bodily passages.		•					•						
Cystic Fibrosis Related Disorders	Cystic Fibrosis Related Disorders share some, but not all of the characteristics of cystic fibrosis, and are linked to mutations on different genes		•					•						
Familial Dysautonomia	Familial Dysautonomia disrupts protein formation, leading to neuronal complications affecting involuntary actions in the nervous system.				•			•			•		•	•
Fanconi Anemia	Fanconi Anemia disrupts production pathways of a specific protein involved in DNA repair, leading to significant abnormalities in different organs.	•		•			•		•				•	
Galactosemia	Galactosemia affects enzymes that facilitate the degradation of galactose, a sugar molecule, leading to excess buildup, and eventually damage, in tissues.				•			•		•				
Gaucher Disease	Gaucher Disease affects enzymes that facilitate lipid metabolism, leading to excess buildup, and eventually damage, in various tissues and organs.	•					•			•			•	
Glycogen Storage Disease Type II	Glycogen Storage Disease Type II affects enzymes that break down the complex sugar molecule glycogen, leading to excess buildup in tissues.		•		•		•			•	•			
Glycogen Storage Disease Type IV	Glycogen Storage Disease Type IV affects enzymes that break down the complex sugar molecule glycogen, leading to excess buildup in tissues.						•			•	•			
Long Q-T Syndrome	Long Q-T Syndrome affects the potassium channels in the heart which causes elongation of the heartbeat, leading to issues in blood circulation.						•							
Maple Syrup Disease	Maple Syrup disease affects the formation of certain protein building blocks, causing urine to have a distinct scent of maple syrup.				•						•	•		
Mucopolidosis Type IV	Mucopolidosis Type IV disrupts normal intellectual development and causes vision impairment.				•						•			•
Niemann Pick Disease	Niemann Pick Disease disrupts a protein involved in the transportation of lipids.		•		•			•		•	•	•		
Non-Syndromic Hearing Loss	Non-Syndromic Hearing Loss is caused by dysfunction in proteins involved with holding cells together, as well as allowing certain nutrients to pass through.					•								
OTC Deficiency	OTC Deficiency is caused by deformed enzymes that are responsible for converting toxic ammonia into urea, a much more neutral substance.		•		•					•	•	•		
Phenylketonuria	Phenylketonuria is caused by deformed enzymes that are responsible for breaking down amino acids, leading to damage in the brain.				•		•					•		
Sickle Cell Disease (Anemia)	Sickle Cell Disease is caused by red blood cells that are deformed into a sickle-like shape, blocking blood vessels and causes circulation complications.	•	•				•		•	•				
Spinal Muscular Atrophy	Spinal Muscular Atrophy disrupts normal development by degeneration of cells.		•								•			
Tay-Sachs	Tay-Sachs disrupts the degradation of toxic substances in the brain in infants.				•	•				•	•	•		•
Usher Syndrome	Usher Syndrome affects processes involved with cell adhesion, leading to deafness and impaired vision.					•								•

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