1.7 Human Albinism at the Molecular Level

Albinism is a group of inherited disorders that results in little or no production of the protein pigment melanin, which determines the color of the skin, hair and eyes. There are different types of albinism. One common type is called Oculocutaneous albinism (OCA) and involves the eyes, hair and skin. Individuals with OCA type 1A have an inactive tyrosinase protein. Tyrosinase is the enzyme that synthesizes melanin. If tyrosinase is inactive, the Melanocyte cells in the skin, hair, and eyes do not have melanin in their vesicles.

	Adapted from - NIH https://rarediseases.info.nih.gov/diseases/5768/albinism											
	Organisms		Humans with normal skin pigmentation.					Humans with Albino trait (OCA1 type albinism)				
	Cells o cells influence the able trait?											
Molecul What do is happe the mole level?	you think ening on	Protein	Are these two proteins the likely to be the same shape? Why or why not?									
	A A A A A A A A A A A A A A A A A A A	Is the amino acid Hydrophilic?										
		Amino Acids										
		RNA										
		DNA	TAC	TAA	TGC	CGG	ACT	TAC	ΤΑΑ	TTC	CGG	ACT

Hydrophobic (water fearing) Amino Acids	Hydrophilic (water loving) Amino Acids			
Gather on the inside of the protein away from water.	Gather on the outside of the protein forming hydrogen bonds with water			
Alanine (Ala) Isoleucine (Ile) Leucine (Leu) Methionine (Met) Phenylalanine (Phe) Valine (Val) Proline (Pro)	Glutamine (Glu) Asparagine (Asn) Histidine (His) Serine (Ser) Threonine (Thr) Tyrosine (Tyr) Cysteine (Cys) Tryptophan (Trp)			

Second letter

		U	С	А	G		
First letter	υ	UUU UUC UUA UUG Leu	UCU UCC UCA UCG	UAU UAC UAA Stop UAG Stop	UGU UGC UGA Stop UGG Trp	UCAG	
	с	CUU CUC CUA CUG	CCU CCC CCA CCG	CAU CAC CAA CAG GIn	CGU CGC CGA CGG	UCAG	letter
	A	AUU AUC AUA AUG Met	ACU ACC ACA ACG	AAU AAC AAA AAG Lys	AGU AGC AGA AGG AGG	UCAG	Third
	G	GUU GUC GUA GUG	GCU GCC GCA GCG	GAU GAC GAA GAG Glu	GGU GGC GGA GGG	UCAG	