Elastin Metabolism

- One tropoelastin gene (ELN)
- The longest lasting protein about 74 years
- Can't be synthesized after 12 or 13 years
- a soluble tropoelastin +lysyl oxidase ➡️insoluble durable cross linked
- lysyl oxidase:

1•participate in Aldol condensation and Schiff base

2•it is extracellular copper enzyme that catalyze the formation of aldehydes

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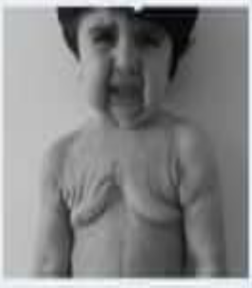
Elastin metabolic daises

Diseases	Caused by	characterized by	inherited in
1. Cutis laxa 	ELN , EFEMP2 ATP6V0A2, ATP7A, ,, FBLN5 gene medications that remove Coppere اي في ٣تنتين ATPفيلن	1. Skin Saggin 2.emphysema aorta 3. aneurysm	1. autosomal dominant 2. autosomal recessive 3. X-linke (occipital horn syndrome→Menkes syndrom)
2. Supravalvular تضيق aortic stenosis (SVAS)	60 mutations in the *ELN* gene 	shortness of heart defect breath, chest pain, and ultimately heart failure	an autosomal dominant
3. Williams syndrome 	*ELN(in 7 chromosome)* , *GTF2I, GTF2IRD1, CLIP2,* and *LIMK1)* ليمك جاب ل كليب 2 gift	connective tissue abnormalities and cardiovascular disease	autosomal dominant
4. Marfan syndrome 	*FBN1*/encodes the connective protein fibrillin-1)	tall, with long limbs and long, thin defects of the heart valves and aorta. It may also affect the lungs, the eyes, the skeleton and the hard palatefingers.	autosomal dominant

We have at least five different pigment:

1.Melanin :brown in color

2.Melanoid: diffuse through the epidermis

3.Keratin :Yellow to orange

4.Hemoglobin &5.oxyhemoglobin :in blood

Melanin

- Derivatives of amino acid =|| Tyrosin||
- A brown ,non refractile ,finely granular with diameter less than 800 nm

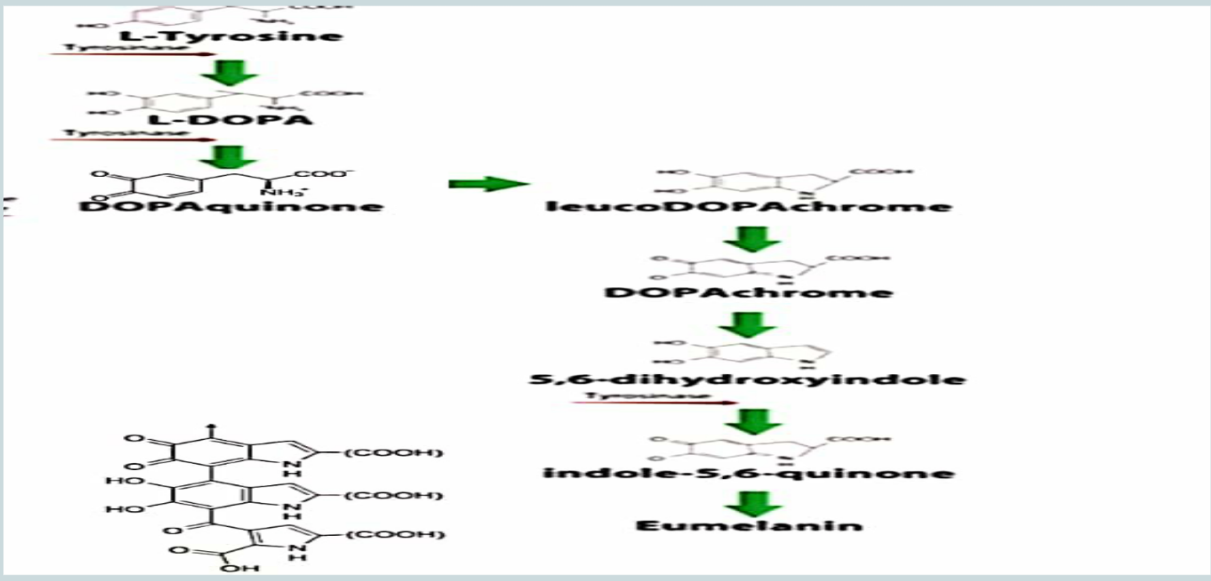
- The process of melanin synthesis::

L-Tyrosine by tyrosine hydroxylase➡️L-DOPA and by DOPA oxidase ➡️DOPA quinone and then it's either A. Aumelanin or

B. Pheomelanin

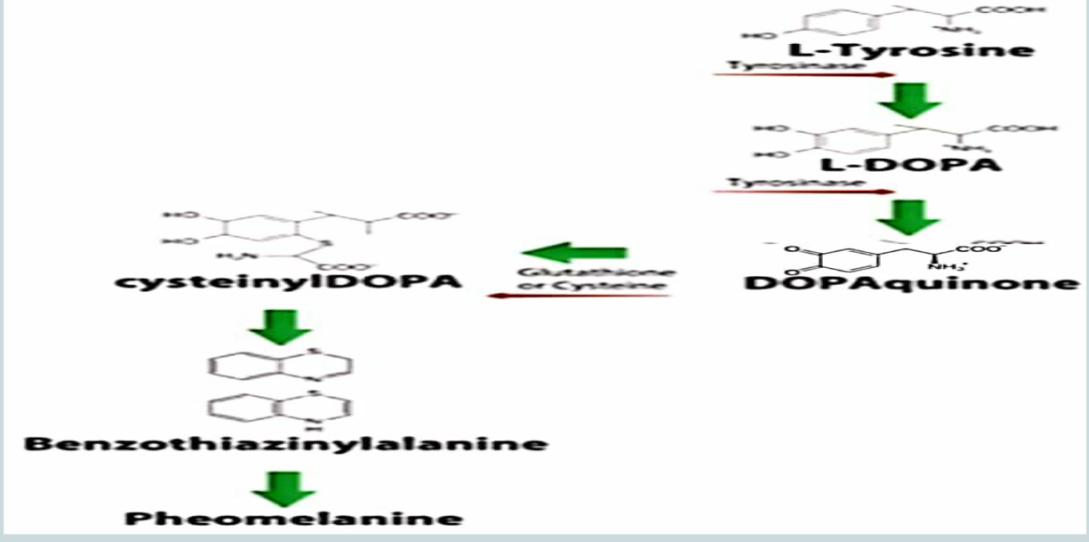
A.Aumelanin:

- brown black in color
  - of dihydroxyindole carboxylic acid



B.Pheomelanin

- cysteine containing
- red brown of benzothiazine
- responsible for red hair and freckl



Mutation

▪ UV rays cause Mutation in DNA and cause two pyrimidine to link together and form a bulge

▪ We have an 1.excision repair system but the chane to have a Mutation is also 1 in 4

2.photoprotection :to convert uv light to heat ,➡️if this not happened a harmful free radical like singlet oxygen and hydroxyl radical

▪ People with dark skin have a lower level of Vitamin.D because melanin block your absorption to vit.D

Albinism ▪ inherited as recessive or dominant

▪ and affect one eye or both and (iris color),skin ,

▪ make a photophobia and vision defect ,cancer ,strabismus ,rapid eye movement

▪ A defect in tyrosine Metabolism

Hyperpigmentation: ▪ darkening in skin and nail

▪ causes: inflammation, sun,acne vulgaris ,injury

▪ with age melanocyte become less diffuse and less controlled