**L5- Dna Repair**

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**Fanconi anaemia (FA)**

* It is a rare [genetic disease](https://en.wikipedia.org/wiki/Genetic_disorder). Among those affected the majority develop [cancer](https://en.wikipedia.org/wiki/Cancer), most often [acute myelogenous leukemia](https://en.wikipedia.org/wiki/Acute_myelogenous_leukemia), and 90% develop [bone marrow failure](https://en.wikipedia.org/wiki/Aplastic_anemia) (the inability to produce blood cells) by age 40. About 60–75% of people have [congenital defects](https://en.wikipedia.org/wiki/Congenital_defects), commonly [short stature](https://en.wikipedia.org/wiki/Short_stature), abnormalities of the skin, arms, head, eyes, kidneys, and ears, and developmental disabilities.

**Lynch syndrome (HNPCC or hereditary nonpolyposis colorectal cancer)**

It is an [autosomal dominant](https://en.wikipedia.org/wiki/Autosomal_dominant) genetic condition that has a high risk of [colon cancer](https://en.wikipedia.org/wiki/Colon_cancer) as well as other cancers including [endometrial cancer](https://en.wikipedia.org/wiki/Endometrial_cancer) (second most common), [ovary](https://en.wikipedia.org/wiki/Ovarian_cancer), [stomach](https://en.wikipedia.org/wiki/Stomach_cancer), [small intestine](https://en.wikipedia.org/wiki/Gastrointestinal_cancer),

[hepatobiliary tract](https://en.wikipedia.org/wiki/Gallbladder_cancer), upper [urinary tract](https://en.wikipedia.org/wiki/Urinary_tract), [brain](https://en.wikipedia.org/wiki/Brain_tumor), and [skin](https://en.wikipedia.org/wiki/Skin_cancer). The increased risk for these cancers is due to inherited mutations that impair [DNA mismatch repair](https://en.wikipedia.org/wiki/DNA_mismatch_repair). It is a type of [cancer syndrome](https://en.wikipedia.org/wiki/Cancer_syndrome).

**Werner syndrome (WS)**

It is also known as "adult [progeria](https://en.wikipedia.org/wiki/Progeria)", is a rare, [autosomal recessive](https://en.wikipedia.org/wiki/Autosomal_recessive) [progeroid syndrome](https://en.wikipedia.org/wiki/Progeroid_syndrome) (PS), which is characterized by the appearance of premature [aging](https://en.wikipedia.org/wiki/Senescence). The median and mean ages of death are 47–48 and 54 years, respectively. The main cause of death is [cardiovascular disease](https://en.wikipedia.org/wiki/Cardiovascular_disease) or cancer.

**Xeroderma pigmentosum**
 It is a rare autosomal recessive [genetic disorder](https://en.wikipedia.org/wiki/Genetic_disorder) of [DNA repair](https://en.wikipedia.org/wiki/DNA_repair) in which the ability to repair damage caused by [ultraviolet](https://en.wikipedia.org/wiki/Ultraviolet) (UV) light is deficient.

**L7 Mutations**

Known disorders in humans include:

**Deletions**
1) **Wolf-Hirschhorn syndrome**, which is caused by **partial deletion of the short arm of chromosome 4**
2) **Jacobsen syndrome**, also called the **terminal 11q deletion disorder**.

**Duplications**

Known human disorders include **Charcot-Marie-Tooth disease type 1A**, which may be caused by **duplication** of the gene encoding **peripheral myelin protein 22 (PMP22) on chromosome 17**.

**L10 Transcription Part 2**

SLE : Systemic lupus erythematosus is an auto immune disease

Production of a.b. to one of the snRNPs,U1-snRNP

Rash on the forehead & cheek bones, giving the wolf like appearance.

Severe kidney damage may follow with arthritis, accumulation of fluid around the heart, & inflammation of the lungs.