

[17] Alkaptonuria (Alcaptonuria): It is a genetic disease caused due to a recessive gene known as Alkapton.

In normal person homogentisate oxidase is formed in liver which causes break down of homogentisic acid ^{into} compound which enter into Krebs cycle.

In homozygous (aa) condition these gene obstacle to formation of homogentisate oxidase, so that homogentisic acid accumulates and excreted in urine. This acid is also known as Alcapton or Alkapton. When it combines with oxygen form a black pigment, causing the urine to black. This pigment slowly in the cartilage of ears and nose. It also cause mild arthritis.

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[18] Albinism: → It is a genetic disease caused due to a recessive gene.

It The affected person (homozygous recessive aa) fails to form tyrosinase which is necessary to form Melanin from Dihydroxyphenyl - alanine.

Lack of melanin in skin, hair, and iris, there is no colour and the person is known as albino.

Albinism is itself not a physical problem, but it leads to disorder of eye and skin due to bright light.

SEX - CHROMOSOMAL

ABNORMALITIES

Due to non-disjunction of sex-chromosomes, abnormal gametes (x) or (y) or (0) are formed causing abnormalities in the individual product.

□ TURNER'S SYNDROME :- It is a genetic disease caused due to aneuploid condition due to lack of one of the sex chromosome. The individual has $(2x0) + 44$ condition.

It is formed due to non-disjunction during gametogenesis for sex-chromosome. The union takes place between $(22 + 0)$ & $(22 + x)$ gametes.

The individual is phenotypically female short stature, underdeveloped breasts, undifferentiated gonads, webbed neck, narrow hip, subnormal intelligence, sparse pubic hair, small uterus, reduced ovaries, broad chest with widely spaced nipples, they may not ovulate or menstruate.

It occurs one in five thousand birth (0.1 in 5000).

[2] Noonan's Syndrome:- It is also known as Male Turner's Syndrome
(i.e. $44+0X$) The chromosome no. is 45.

It is caused due to non-disjunction of sex chromosome during gametogenesis causing $(A+0)$ gametes.

They are phenotypically male, short statured, webbed neck, drooping upper eyelid, etc and less developed gonads.

They are not viable long.

[3] Super Females:- They are due to trisomy of extra X chromosome. It is caused due to non-disjunction of X chromosome during oogenesis. $(A+XX)$
The total chromosome number in the individual may be 47 or 48. $(2A+XXX)$
or $(2A+XXXX)$

The individual is a female with abnormal sexual development and mental retardation, low fertility

[4] Super Male:- They are due to trisomy of extra-Y chromosome caused due to non-disjunction of sex chromosomes during spermatogenesis
The chromosome set up is $(2A+XXY)$

The individuals are phenotypically male with unusual height.

They overproduce testosterone (male hormone), hence more aggressive than normal males. They are somewhat mentally retarded, but with a criminal bent of mind. Though the last view i.e. criminality is objected by some recent workers.

[5] Klinefelter's Syndrome: It is a genetic disease caused due to extra-chromosome formed due to non-disjunction during gameto-genesis.

The individual have $2A + XXXY$ or $2A + XXXY$.

They are phenotypically male, tall with long legs, have small testes, obesity, sparse body hair. There may be many female characteristics i.e. enlarged breasts.

Testes is small, prostate is small azospermia, with reduced mental intelligence.

Nuclei show barr body.

Patients suffers with incidence of osteoporosis, leg ulcers and breast cancer.

It occurs about 1 in 2000 of live births.