

Done By:

Arwa Al-Madani

Sarah Bin-Hussain

Bedoor Al-Qadrah

Reham Al-Henaki

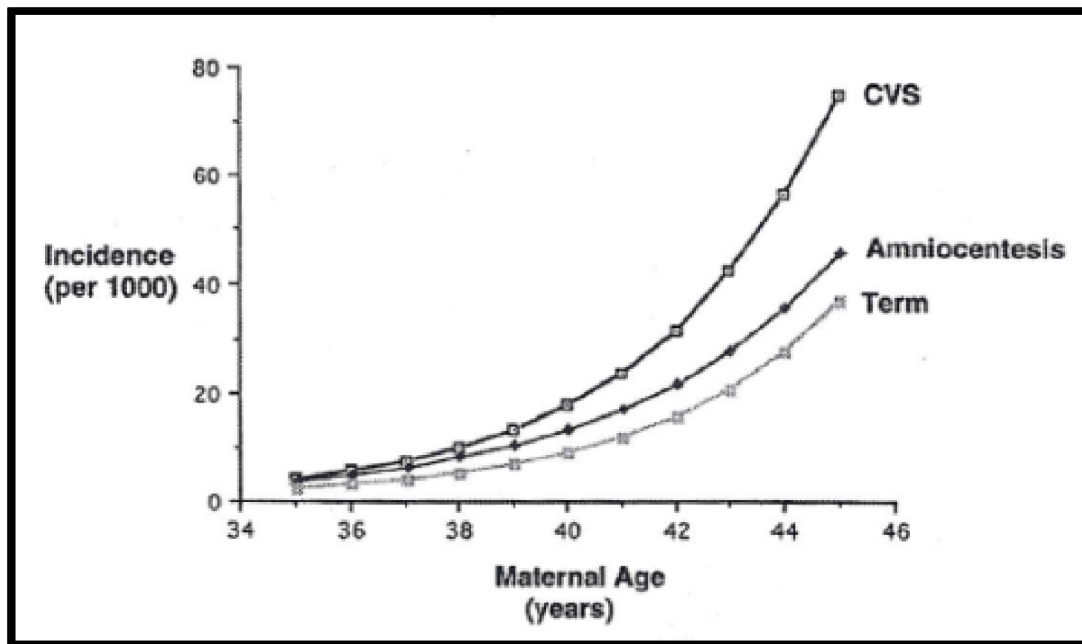
The highlighted words → are translated words in the end

| | DOWN SYNDROME | TURNERSYNDROME | KLINEFELTER SYNDROME |
|----------------------|--|---|---|
| EPIDEMIOLOGY | 1,511 000 births. | 0.4 /1000 female births (but 20 % of chromosome anomalies found in early miscarriages, i.e. about 10% early miscarriages). | <ul style="list-style-type: none"> • 1.5 /1 000 male births. • increased maternal age. |
| Malformations | <ul style="list-style-type: none"> -Heart (40%): -Digestive (10 %): -Ocular | <ul style="list-style-type: none"> • cardiovascular (20-30%). • renal (40-50 %): horseshoe kidney ... • congenitally dislocated hip, scoliosis • sense-organs | _____ |
| DIAGNOSIS | <ul style="list-style-type: none"> -Free trisomy 21 (92,5 % of cases): -Free trisomy 21 in mosaic (2,5 % of cases): -Trisomy 21 due to translocation: -Other | <ul style="list-style-type: none"> • 45, X . : 55 % of cases. • mosaicisms ... --> • [r(x); del (Xp), del (Xq); i(Xp), i(Xq)] | <ul style="list-style-type: none"> • 47 XXY homogeneous: 80 % of cases. • XXXY, XXXXY, XXYX: 10 %. • in mosaic: 5-10 % |

| | DOWN SYNDROME | TURNR SYNDROME | KLINEFELTER SYNDROME |
|--------------------------|---|--|---|
| Clinical features | <p>The most frequent viable chromosome disease. Associates: <i>dysmorphia</i> + <i>psycho-motor delay</i>, and possible <i>visceral malformations</i> (found in more than 1/3 of cases)</p> <ul style="list-style-type: none"> • frequent microcephaly, short neck, • flat occiput and brachycephaly; • moon-shaped face; • flat nasal bridge; • epicanthus (regresse with age); • upward slanting | <p>growth retardation and impuberism</p> <ul style="list-style-type: none"> • prenatal and postnatal growth retardation • single umbilical artery frequently. • excess of skin and webbed skin on the nucha (pterygium colli). • widely spaced nipples. • lymphoedema of hands and feet. • 'small size (adult < 1,45 m). • -normal or subnormal intelligence • -Osteoporosis above | <p>normal intelligence or mild retardation,</p> <ul style="list-style-type: none"> • infertility, • possible behavior or psychiatric problems, • Tall stature • Long legs • Frontal baldness absent • Slightly feminized physique <ul style="list-style-type: none"> ○ Gynecomastia ○ Narrow shoulders ○ Wide hips ○ Feminine fat and hair distribution ○ Reduced body hair |

| | | | |
|--|---|---|---|
| | <p>palpebral fissures;</p> <ul style="list-style-type: none"> • Brushfield spots in the iris • frequently open mouth; • high arched narrow palate; • late appearing/malformed teeth (numerical anomalies, • agenesis of lateral incisors ...) • short and broad; • brachymesophalangia of the 2nd and 5th fingers; • clinodactyly of the 5th finger; • flat feet; • first toe set apart from the others • by a gap, with a crease. • transverse palmar crease • hypotonia +++ • the mental retardation. • children's behaviour: -Seizures . | <p>45 yrs.</p> <ul style="list-style-type: none"> • -short neck. • 'pterygium colli . • -low hair line. • low set ears. • 'shield chest. • 'widely spaced nipples. • 'Coarctation of aorta • 'multiple pigmented nevi. • -triangular shaped face, • -looks sad. • 'possible epicanthus. • -downward slanting • palpebrale fissures. • 'high-arched palate. • 'cubitus valgus . • 'short 4th metacarpal. • 'hypoplastic nails • 'Streak ovaries. • 'infantile external genitalia. • 'hypoplastic uterus. • -amenorrhea and sterility. • 'absence of breast development. • 'rare pubic pilosity. | <ul style="list-style-type: none"> ○ Reduced facial hair • normal or rare, feminine shaped pubic pilosity • Testicular atrophy • Hypogonadism • Reduced testosterone levels • Infertility • Educational difficulties • Intelligence can be 10-15 points lower than siblings • but usually in normal range <p>Behavioural problems.</p> |
|--|---|---|---|

DOWN SYNDROME:



Incidence of trisomy 21 at the time of :

- chorionic villus sampling (10-11 weeks),
- amniocentesis (16 weeks)
- and term.

✚ The incidence of trisomy 21 increases with increasing maternal age.

DOWN SYNDROME

epicanthus is a skin fold of the upper eyelid, covering the inner corner (medial canthus) of the eye..



Palpebral fissure is the anatomic name for the separation between the upper and lower eyelids.

Brushfield spots are small white or grayish/brown spots on the periphery of the iris

brachymesophalangia Abnormal shortness of the middle phalanxes of the digits

clinodactyly → انحراف الاصابع

palmar crease → the line which found in the hand .. normally 3 in down syndrome it is 1

**TURNER SYNDROME**

nucha → The back of the neck

pterygium colli → webbed neck; a thick skin fold on the side of the neck from the mastoid region to the acromion.

Pilosity → كثافة الشعر