

Childhood syndromes

Patau trisomy 13	Edward trisomy 18	Down trisomy 21
polydactyly	rocker-bottom feet	epicanthic folds
microcephaly	micrognathia	hypotonia
microphthalmia	low-set ears	low-set ears
cleft palate	overlapping fingers	single palmar crease
scalp lesions	developmental delay	protruding tongue
Fragile X	Noonan	Pierre-Robin
macro-orchidism	webbed neck	posterior tongue
macrocephaly	pectus excavatum	micrognathia
long face	short stature	cleft palate
large ears	pulmonary stenosis	OSA
learning difficulty	brachydactyly	
Prader-Willi	William	Turner 45,XO
hypogonadism	extroversion	bicuspid aortic valve
hypotonia	short stature	short stature
hyperphagia	learning difficulty	shield chest
obesity	aortic stenosis	amenorrhoea
short stature	hypercalcaemia	high-arched palate
Klinefelter 47,XXY		
tall stature		
hypogonadism		
infertility		
gynaecomastia		
high gonadotrophins		

Notes

- Down risk increases with maternal age: 1/380 at 35, 1/100 at 40, 1/30 at 45
 - risk of recurrence after one Down pregnancy increases 10-fold
 - associated with heart defects, hearing loss, cataracts, GORD, hypothyroidism, AML
- Treacher-Collins is similar to Pierre-Robin but with autosomal dominant inheritance
- Kallman is similar to Klinefelter but with anosmia