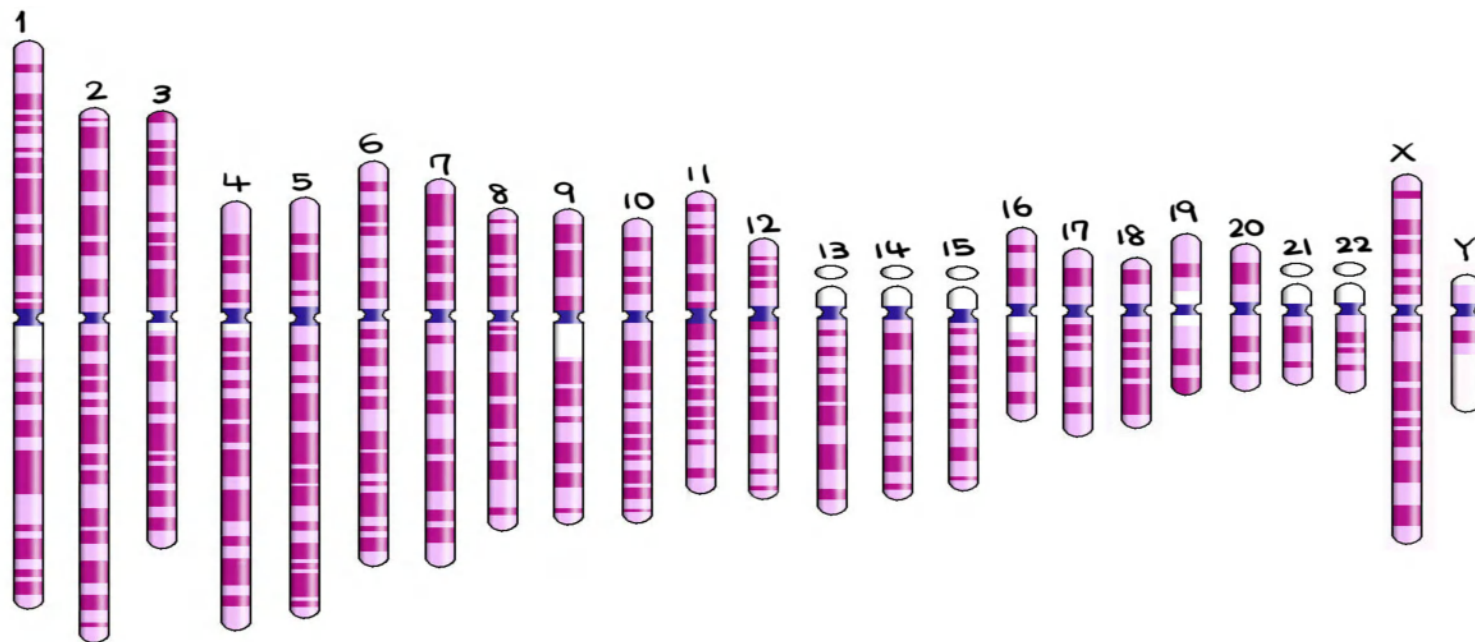


# Essential genetics 5

## Disease map on chromosomes



例

**Gaucher disease** 単一遺伝子病

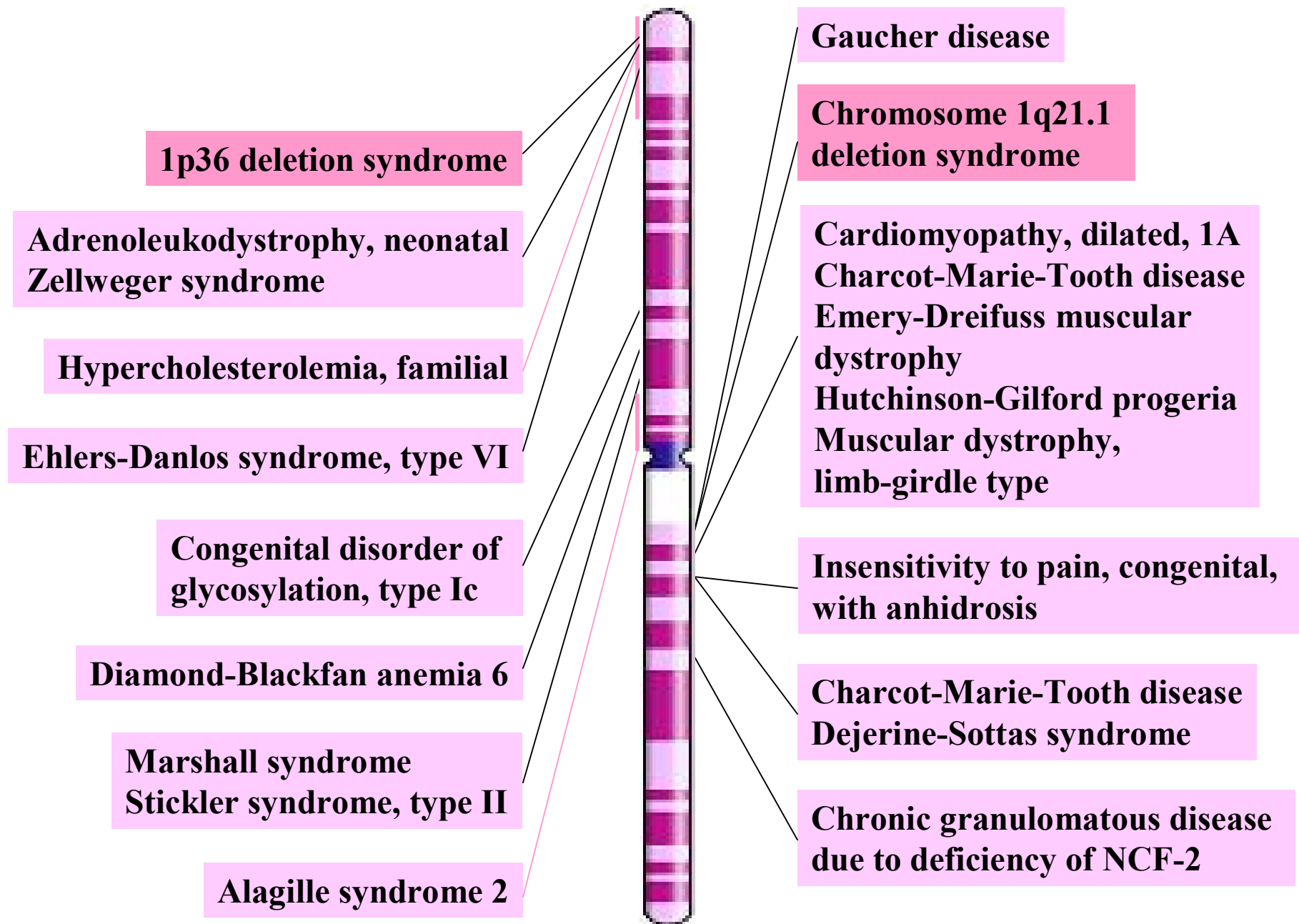
**Prader-Willi syndrome** 隣接遺伝子症候群, 欠失が主因となる疾患

**Trisomy 13** 複数の遺伝子の重複によって起こる疾患

遺伝子の座位あるいは欠失等の範囲を示す

天使病院  
臨床遺伝診療室  
外木秀文  
挿画 Koromo

# Disease map on chromosome 1



## Disease map on chromosome 2

**Epiphyseal dysplasia, multiple  
Spondyloepimetaphyseal dysplasia**

**Noonan syndrome  
Peters anomaly**

**Parkinson disease, familial**

**Seizures, benign familial  
neonatal-infantile**

**Ehlers-Danlos syndrome,  
type I, III, IV**

**Rhizomelic chondrodysplasia  
punctata, type 3**

**Split-hand/foot malformation**

**Brachydactyly, type D-E,  
Brachydactyly-syndactyly syndrome  
Synpolydactyly, type II and V**

**Leigh syndrome**

**Multiple pterygium syndrome  
Escobar syndrome**

**Brachydactyly, type A1**

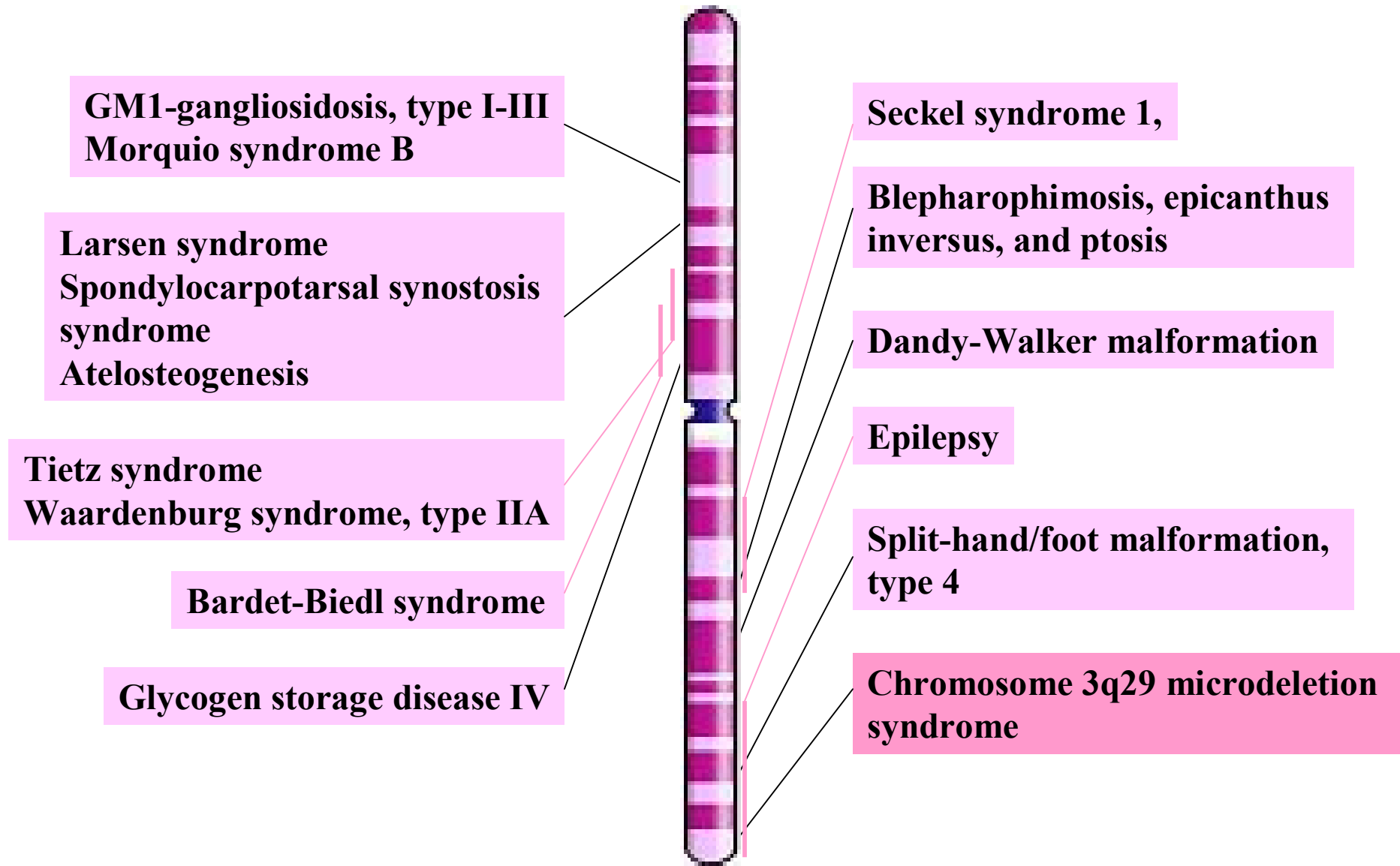
**Waardenburg syndrome**

**Alport syndrome,  
autosomal recessive**

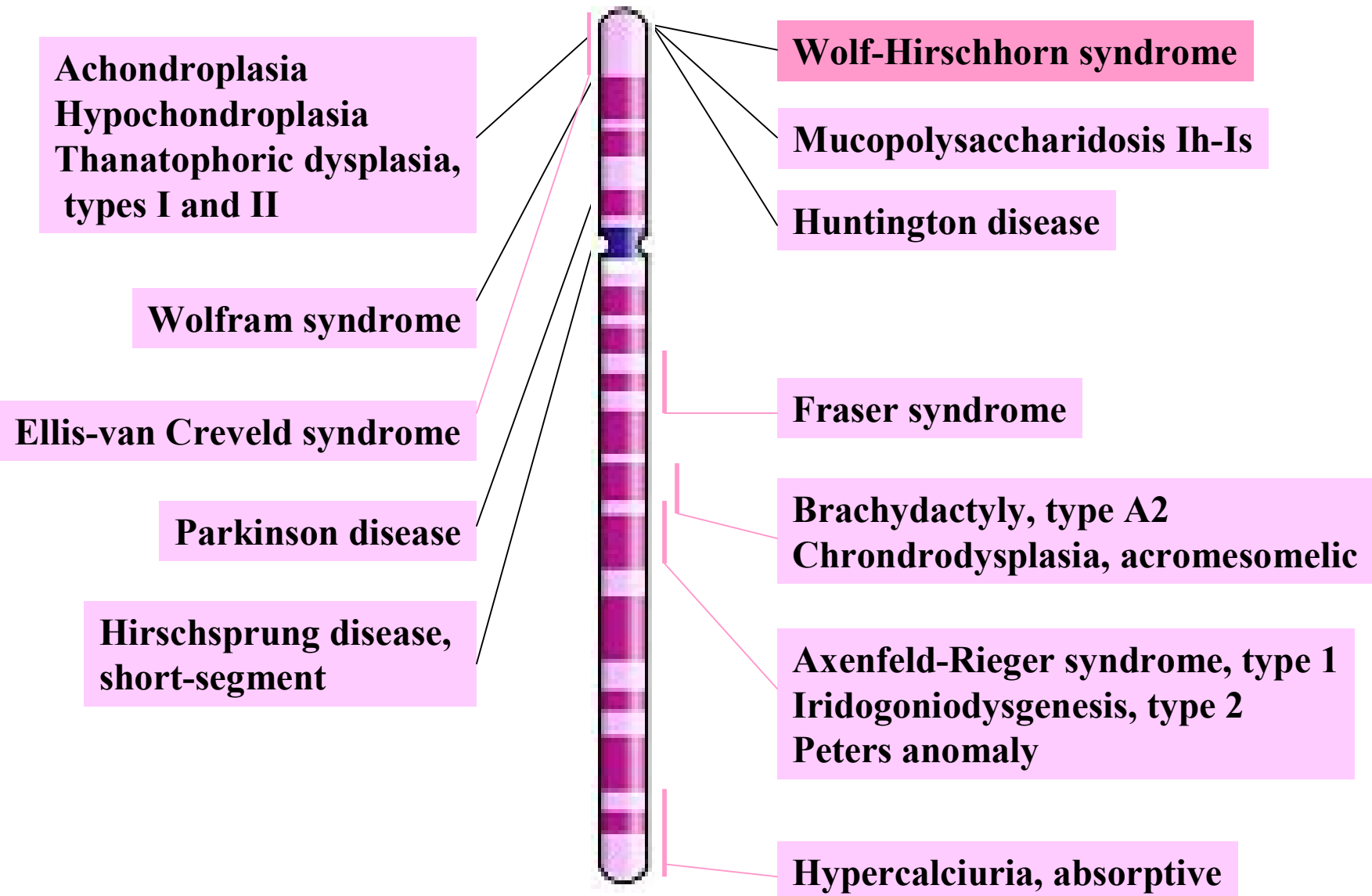
**Crigler-Najjar syndrome**



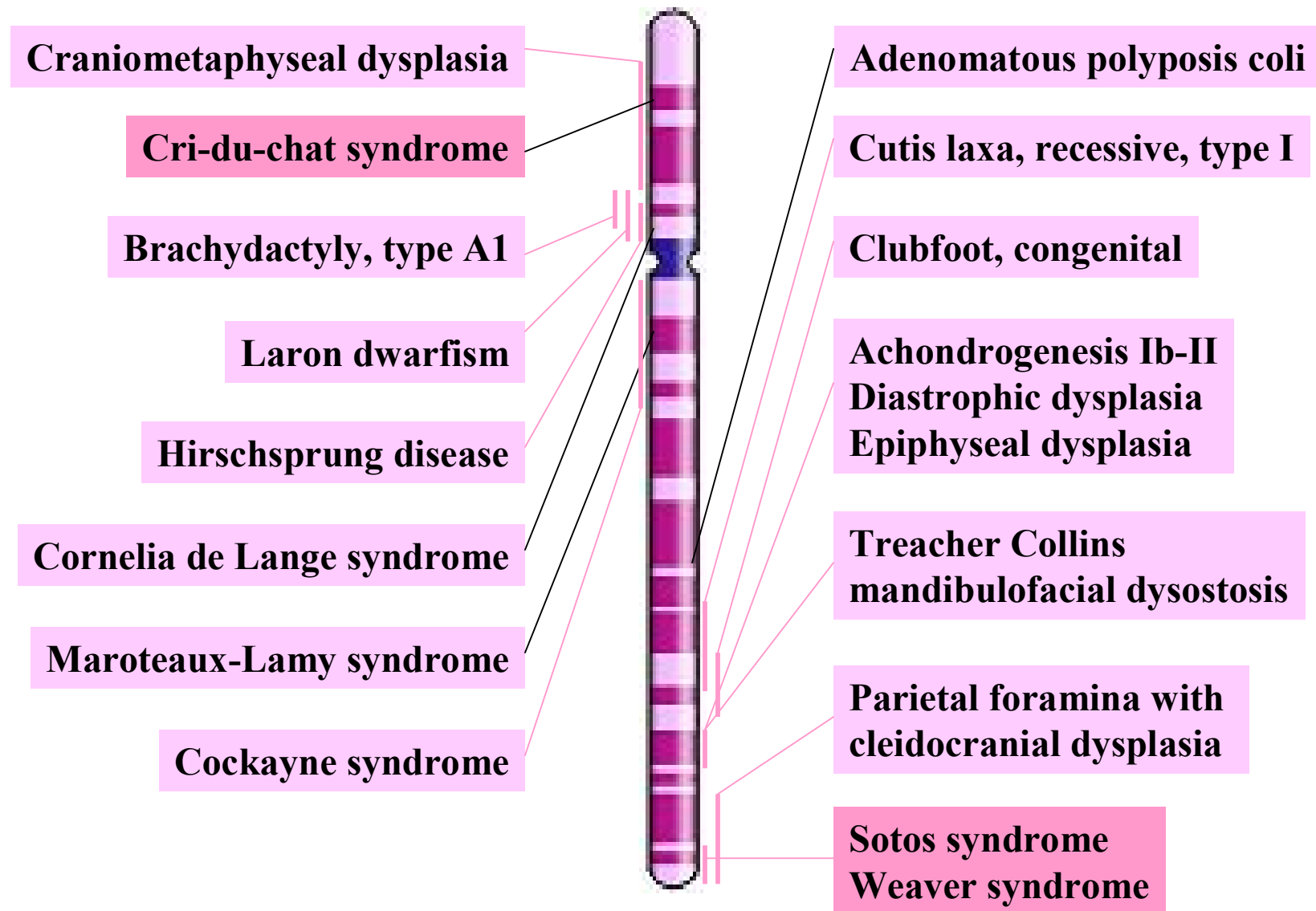
# Disease map on chromosome 3



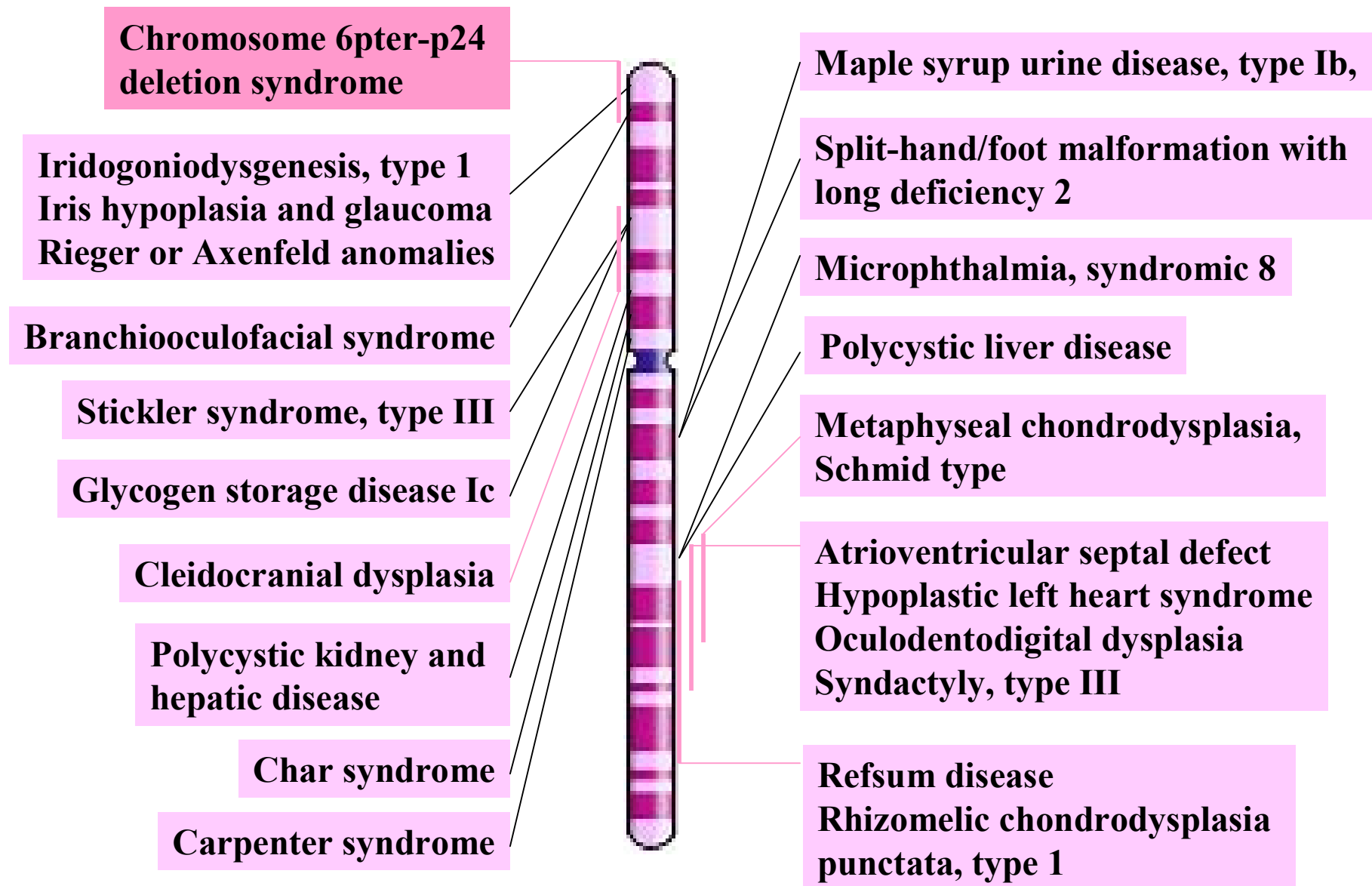
# Disease map on chromosome 4



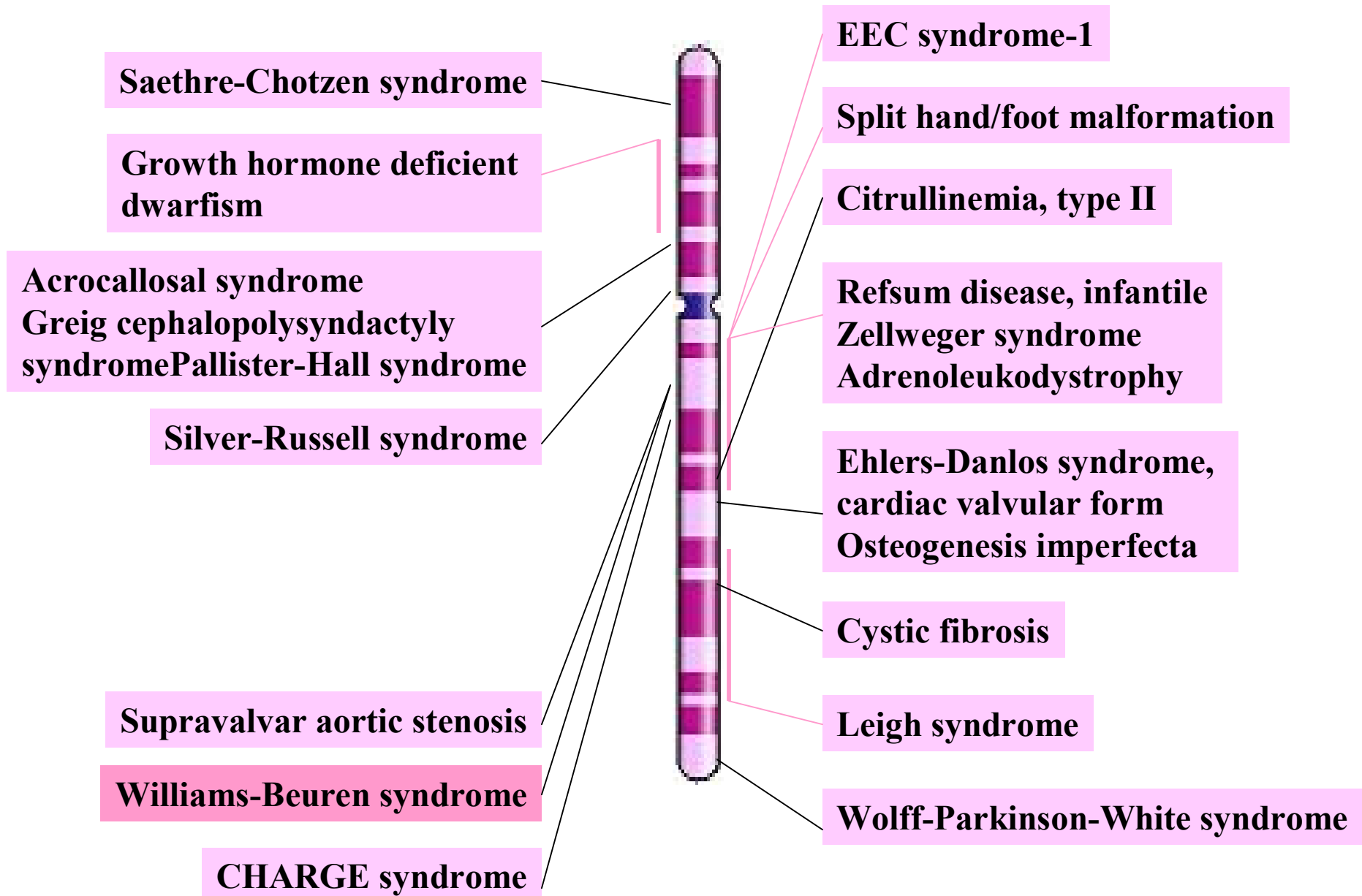
# Disease map on chromosome 5



# Disease map on chromosome 6

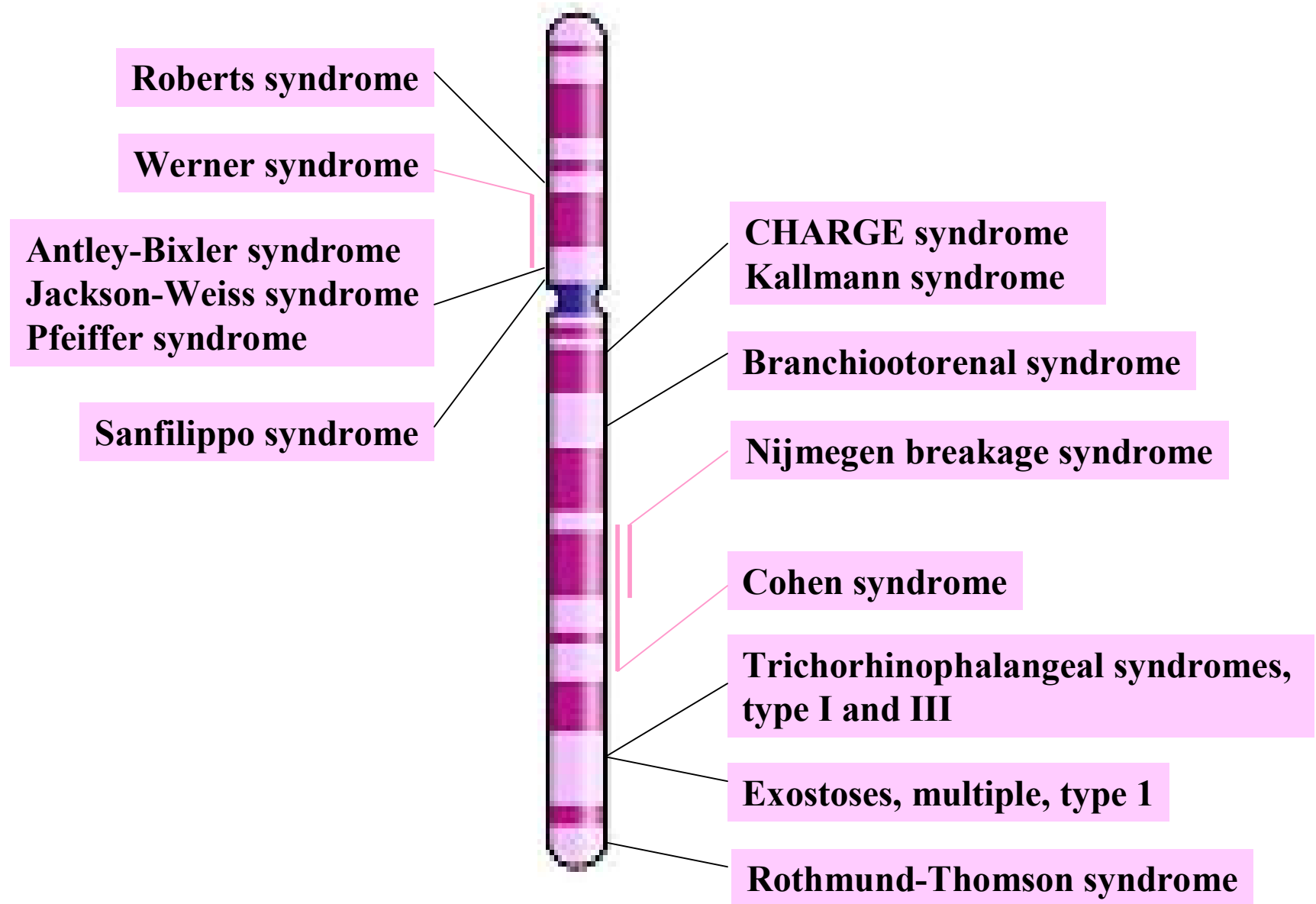


# Disease map on chromosome 7

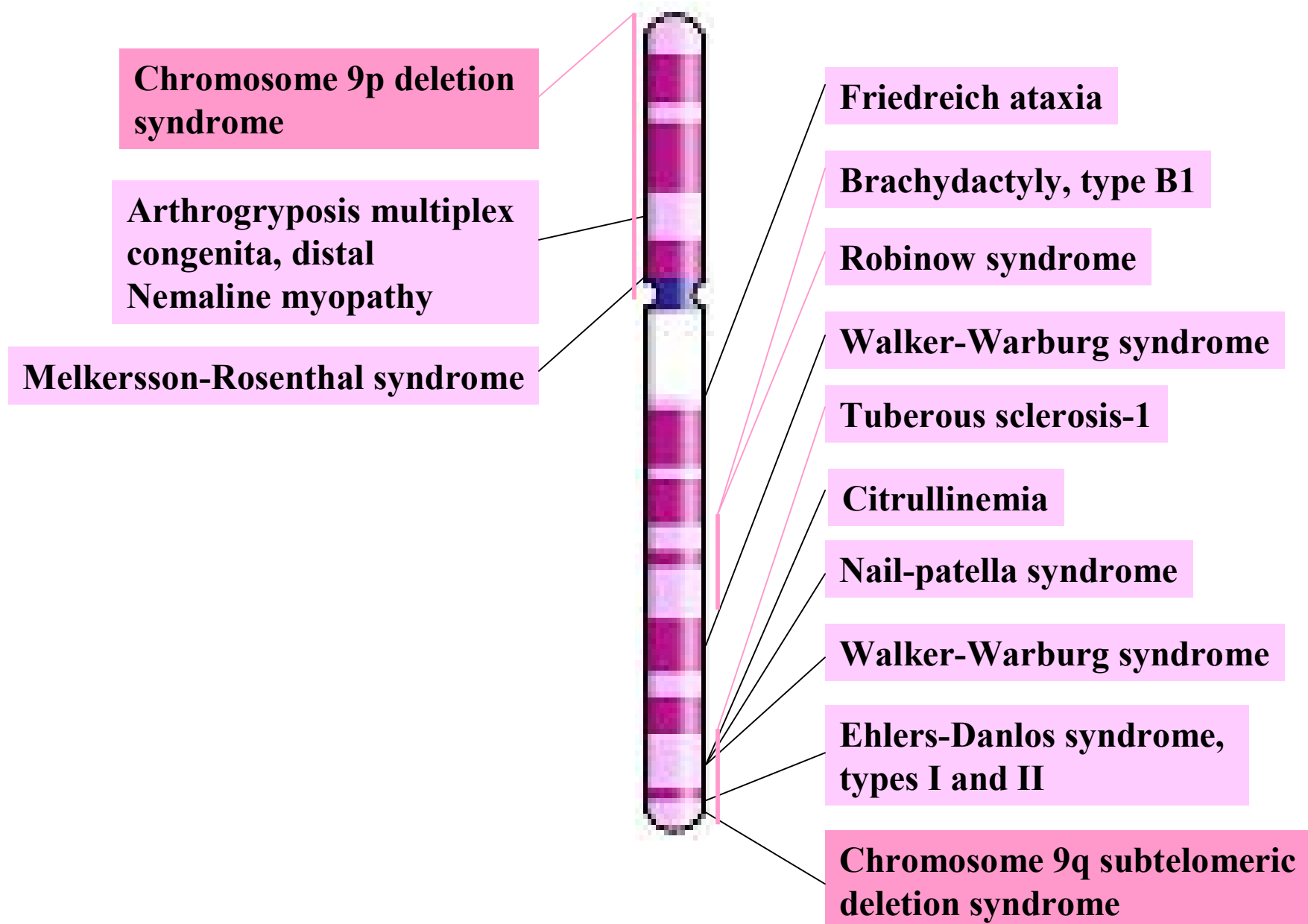




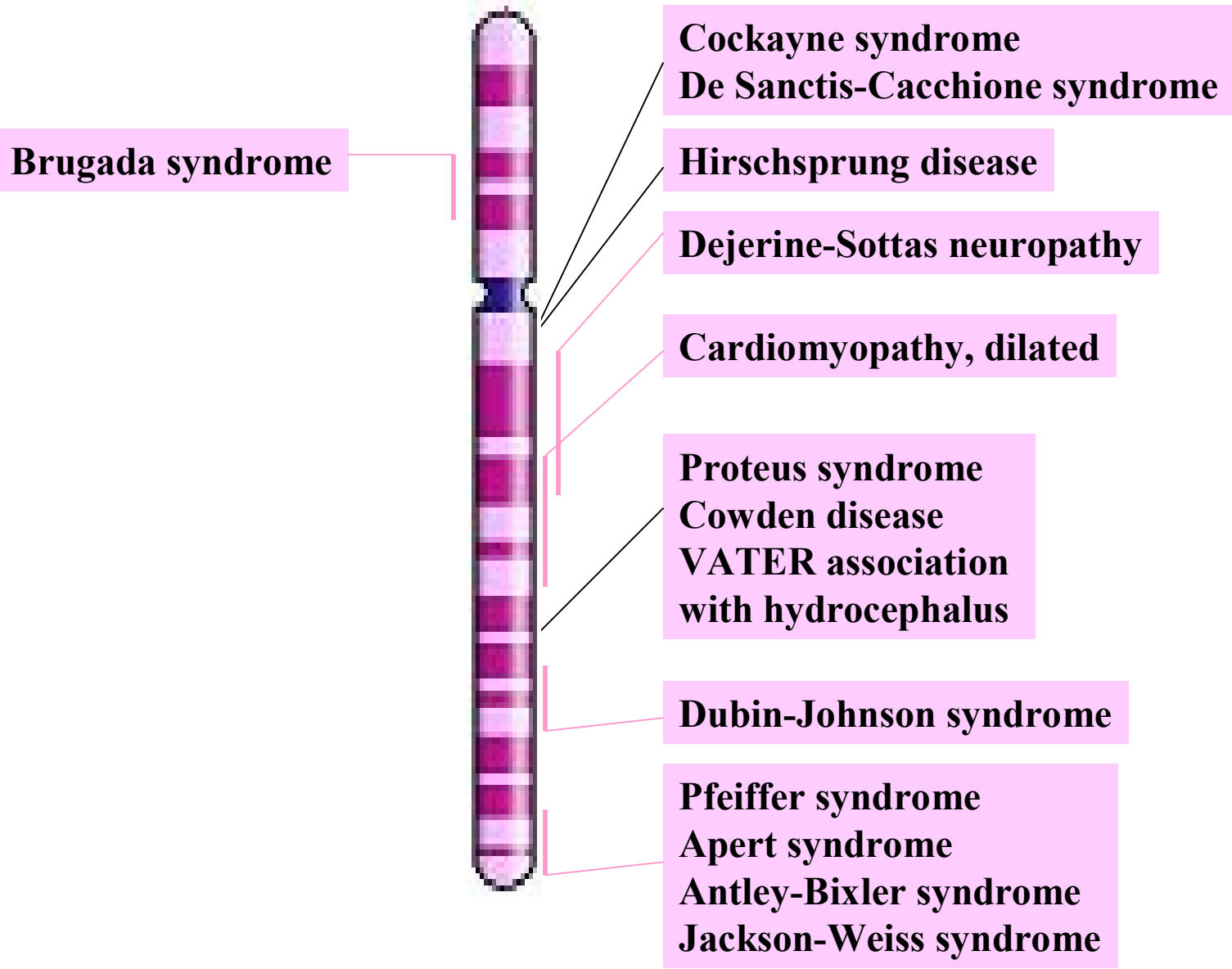
# Disease map on chromosome 8



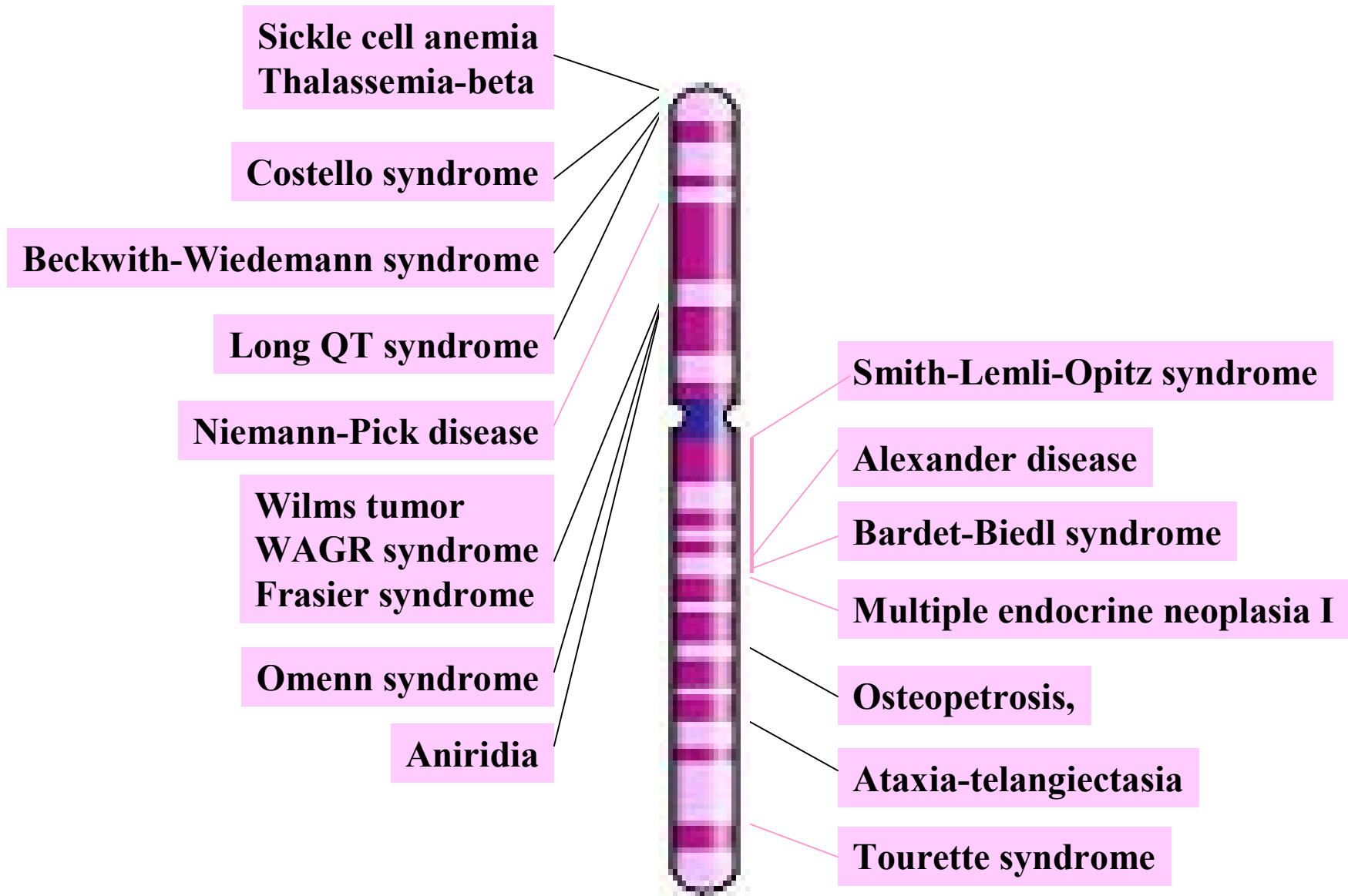
# Disease map on chromosome 9



# Disease map on chromosome 10



# Disease map on chromosome 11



# Disease map on chromosome 12

**Zellweger syndrome**

**Costello syndrome**



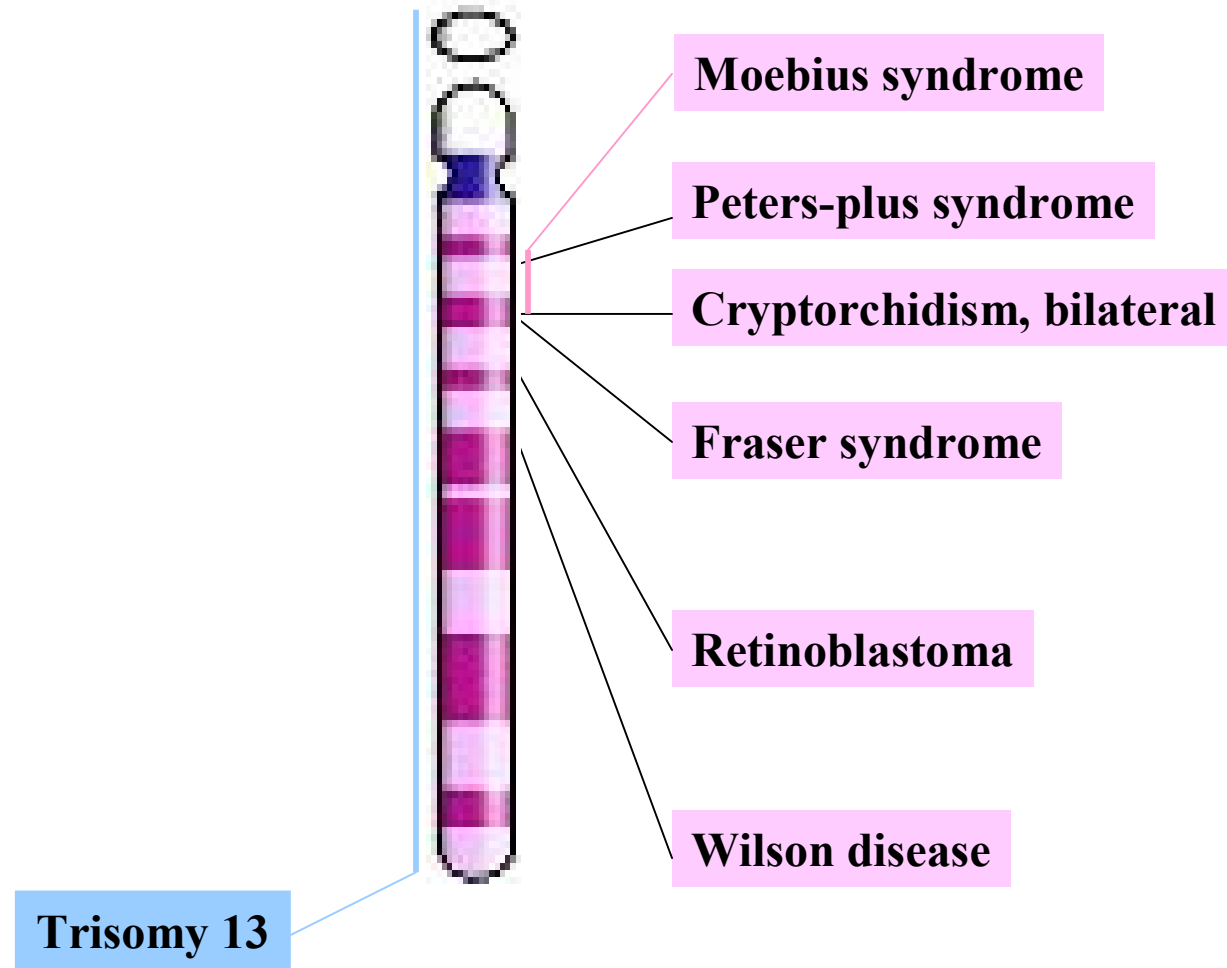
**Epidermolysis bullosa simplex**

**Stickler syndrome**  
**SED congenita**  
**Achondrogenesis**  
**-hypochondrogenesis, type II**

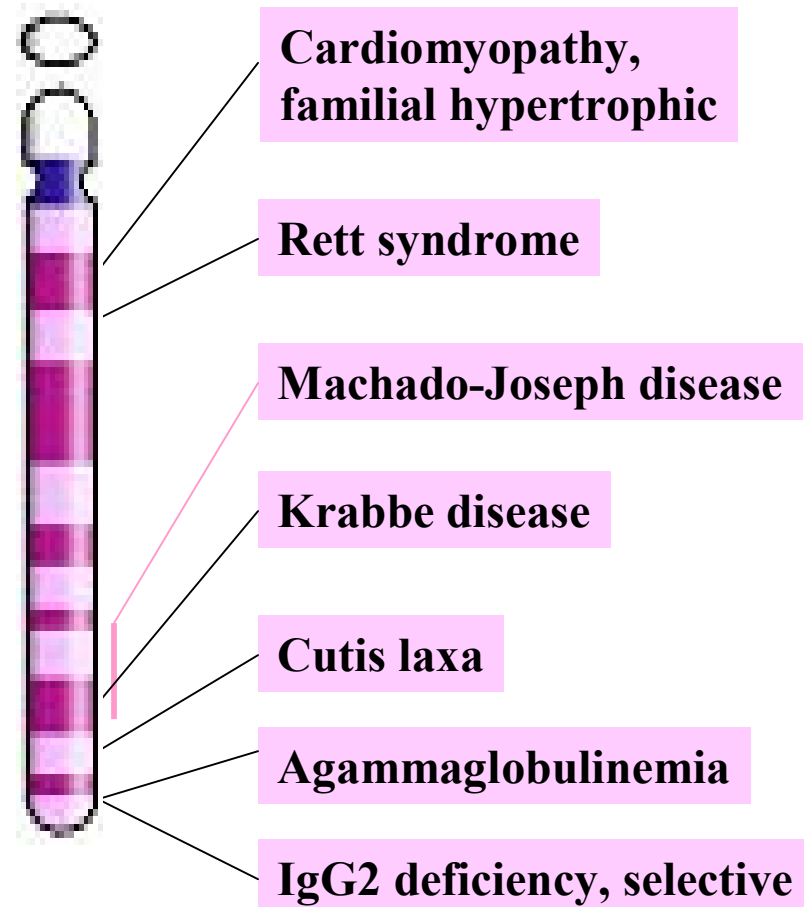
**Holt-Oram syndrome**

**Noonan syndrome**  
**Leopard syndrome**

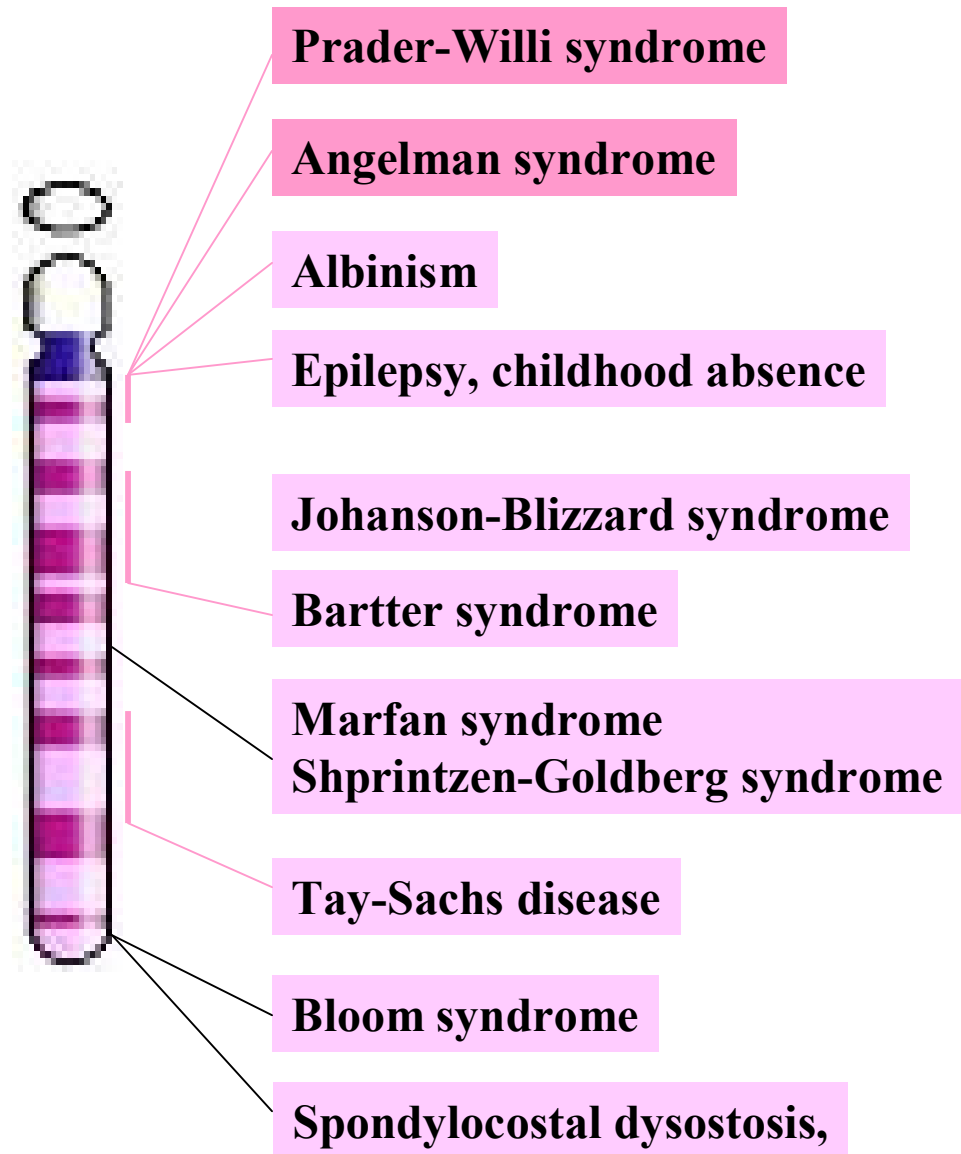
# Disease map on chromosome 13



# Disease map on chromosome 14

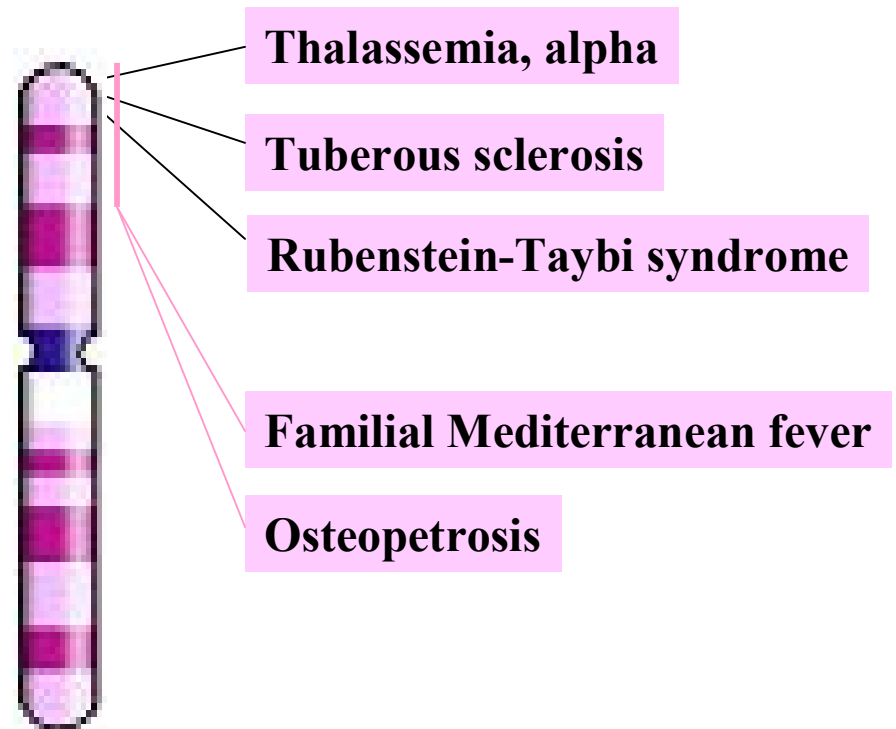


# Disease map on chromosome 15

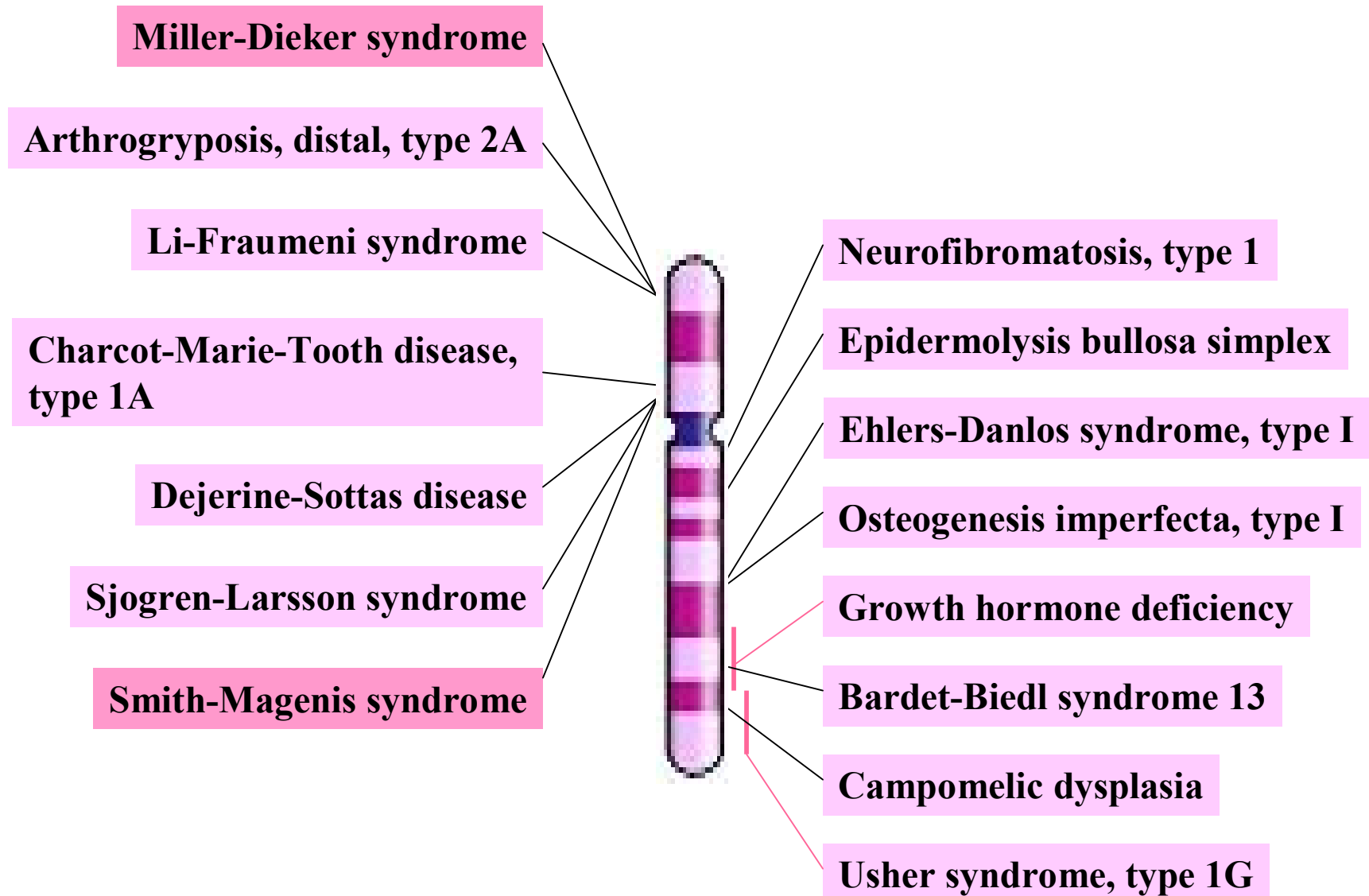




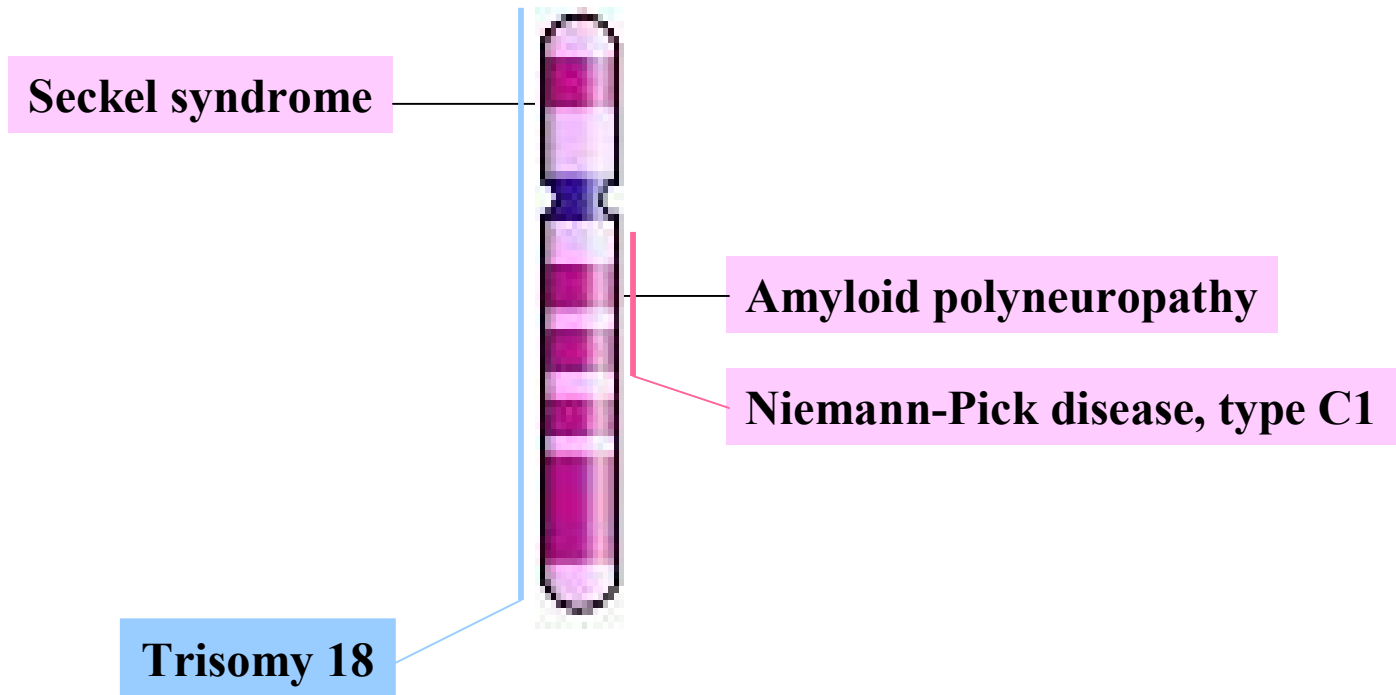
# Disease map on chromosome 16



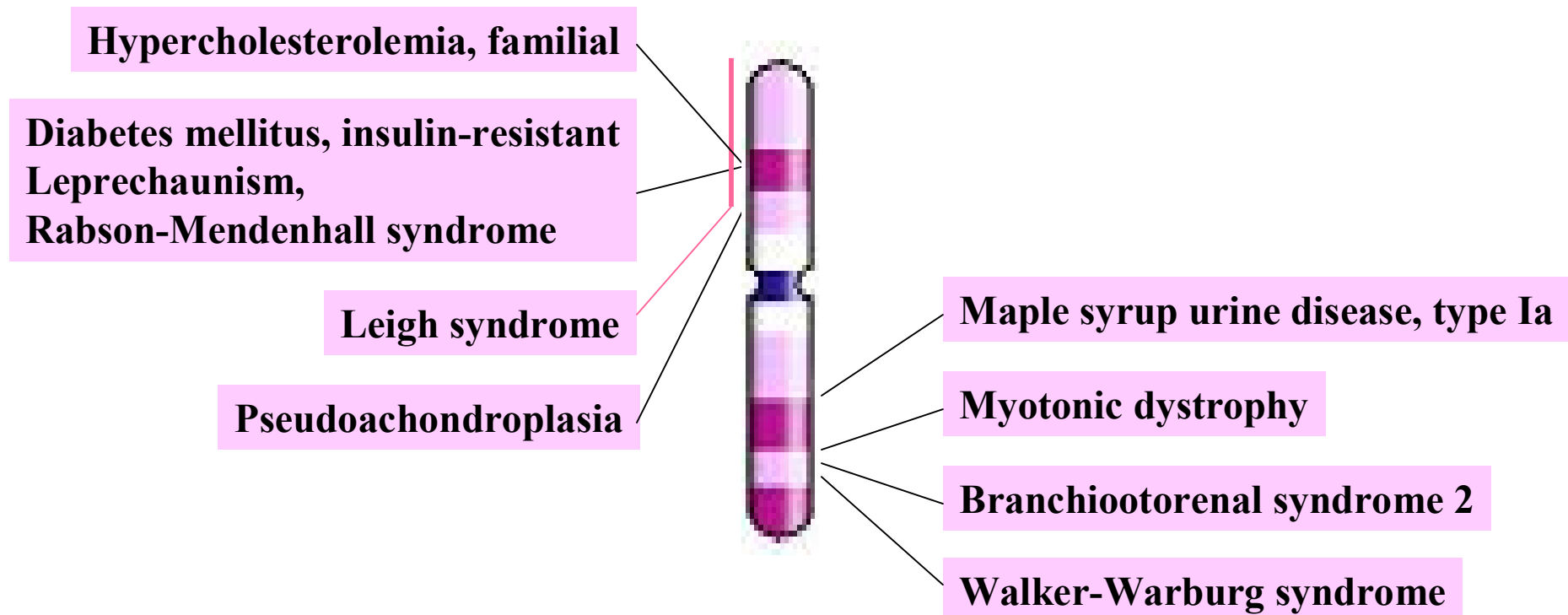
# Disease map on chromosome 17



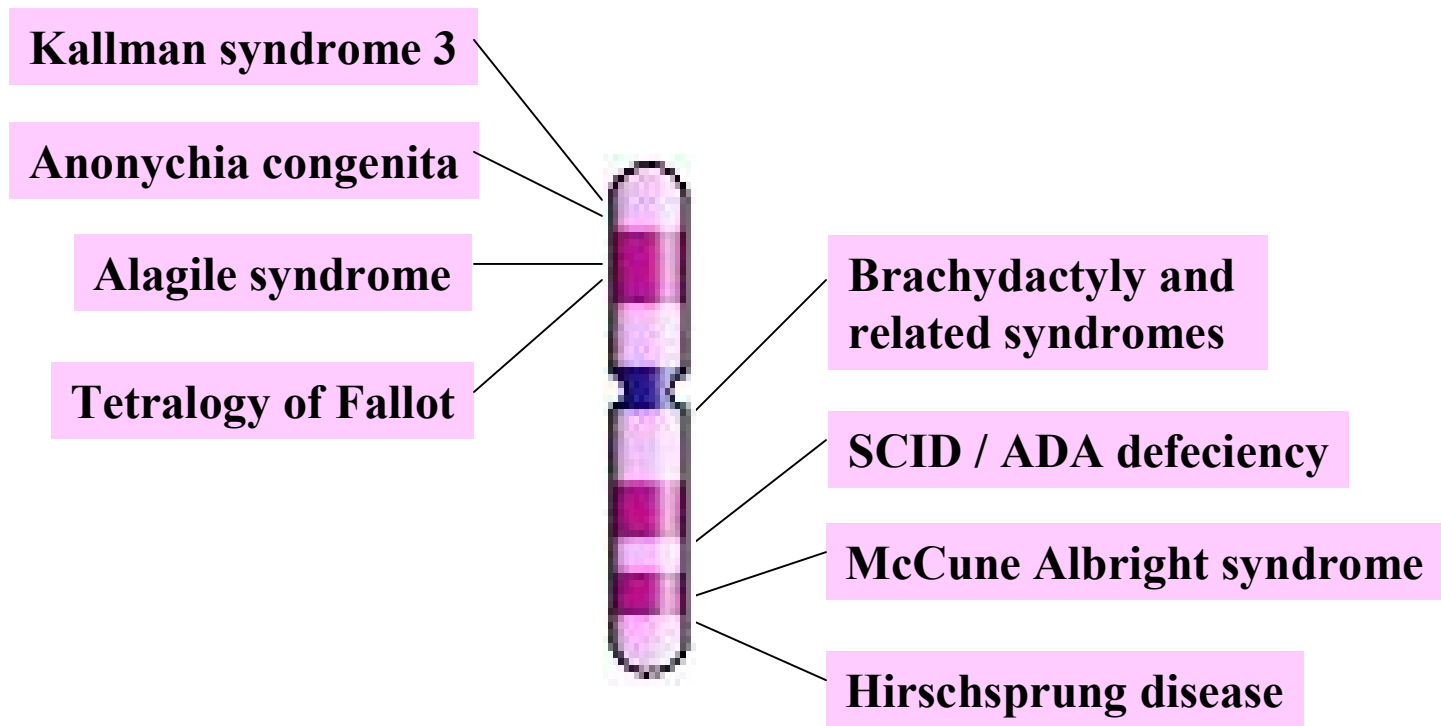
# Disease map on chromosome 18



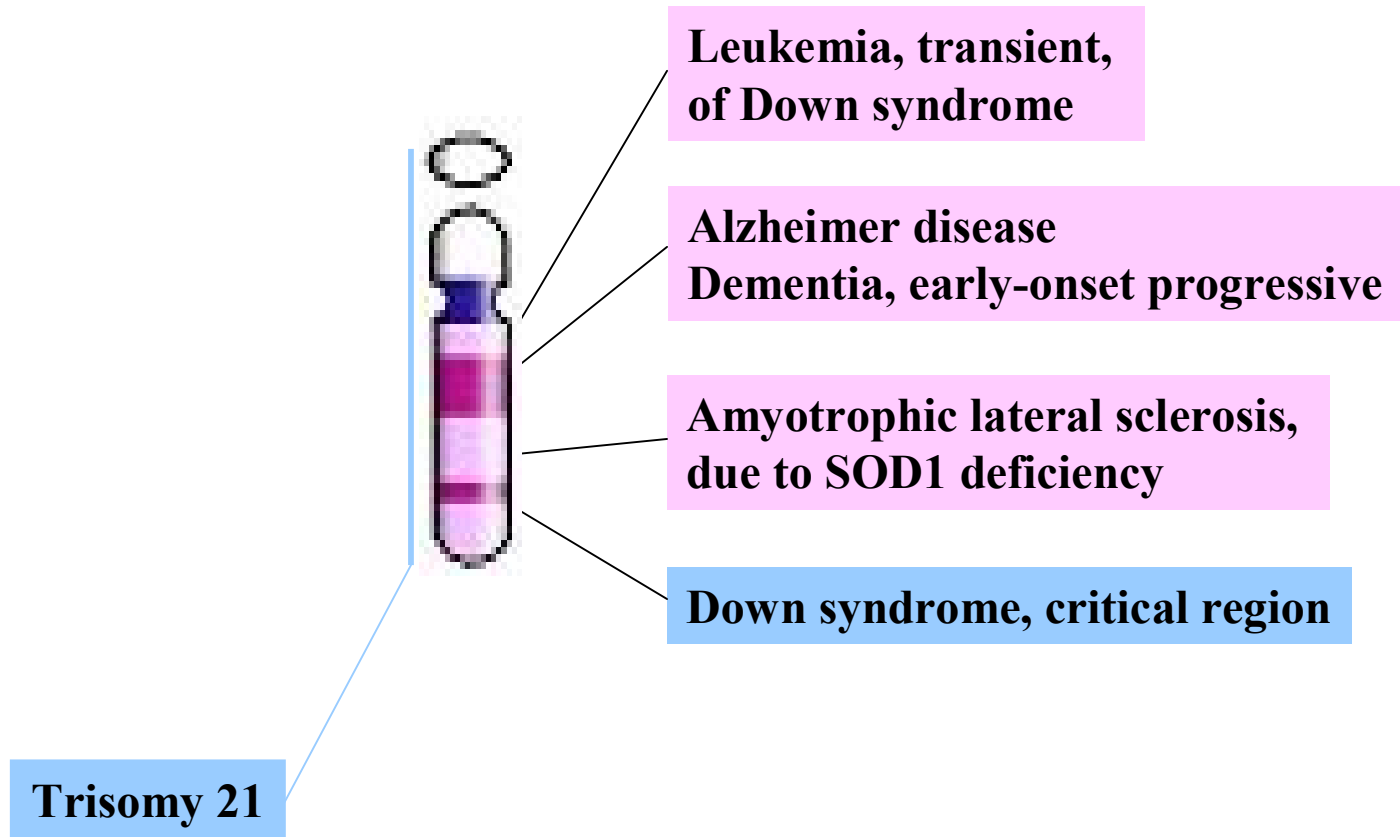
# Disease map on chromosome 19



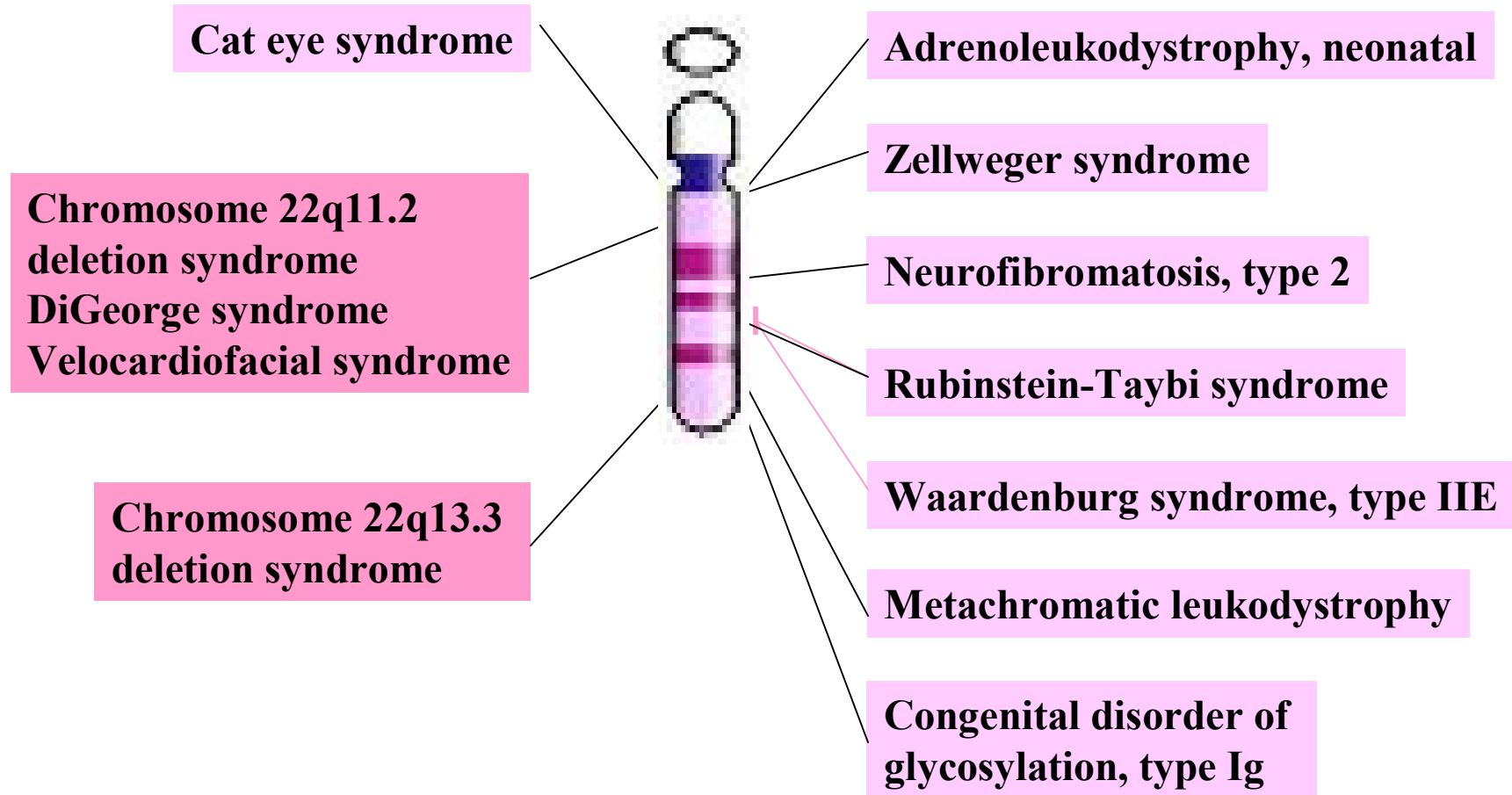
# Disease map on chromosome 20



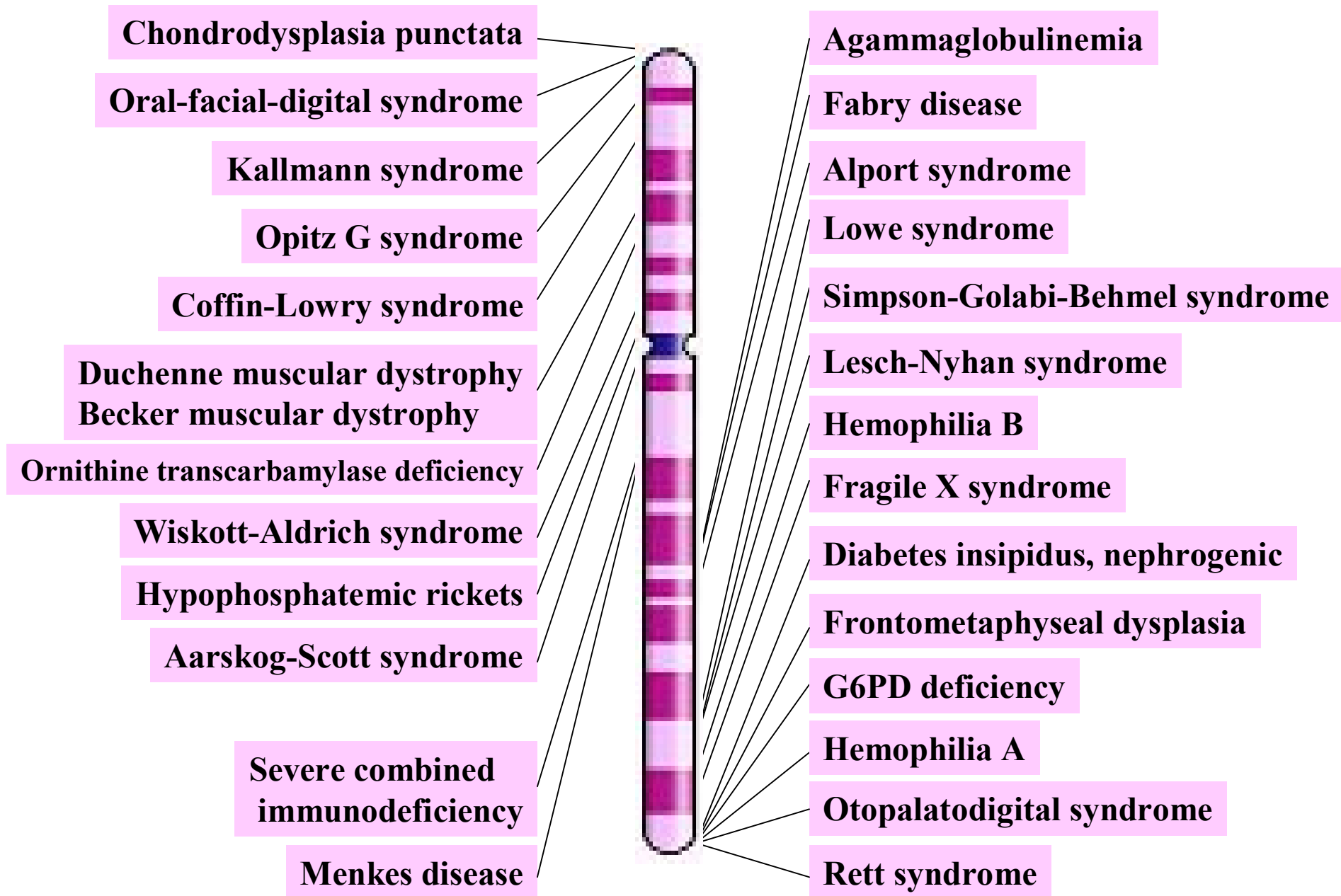
# Disease map on chromosome 21



# Disease map on chromosome 22



# Disease map on chromosome X





# Disease map on chromosome Y

