Topics for Self-Study: *Causes and types of structural chromosomal aberrations, clinical syndromes caused by structural chromosomal aberrations, microdeletion syndromes, genomic imprinting.*

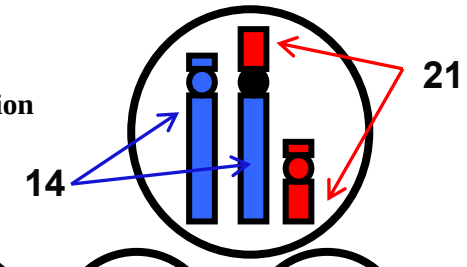
Selected deletion syndromes	
Syndrome	**Localization**
S. Cri-du-chat	del(5p)
Wolf-Hirschhorn s.	del(4p)
Turner s. (deletion form)	del(Xp)

Selected microdeletion syndromes	
Syndrome	**Localization**
Angelman s.	del(15)(q11q13)mat
Prader-Willi s.	del(15)(q11q13)pat
DiGeorge s.	del(22)(q11)

Karyotype formulas for selected structural rearrangements according to ISCN 2013	
Symbol	**Description and example of usage**
der	Derivative chromosome or Robertsonian translocation: 45,XY,der(14;21)
rob	Robertsonian translocation: 45,XY,rob(14;21)
t	Reciprocal translocation: 46,XX,t(4;15)
del	Deletion of chromosomal segment: 46,XY,del(5p) or 46,X,del(Xp)
inv	Inversion: 46,XX,inv(9)(p12q13)
mar	Marker chromosome: 47,XX,+mar

In the first part of this practical course you will be presented meiotic chromosomal segregation in a carrier of **rob(14;21)** balanced Robertsonian translocation. Draw all possibilities and mark the vital and lethal combination.

Parental combination

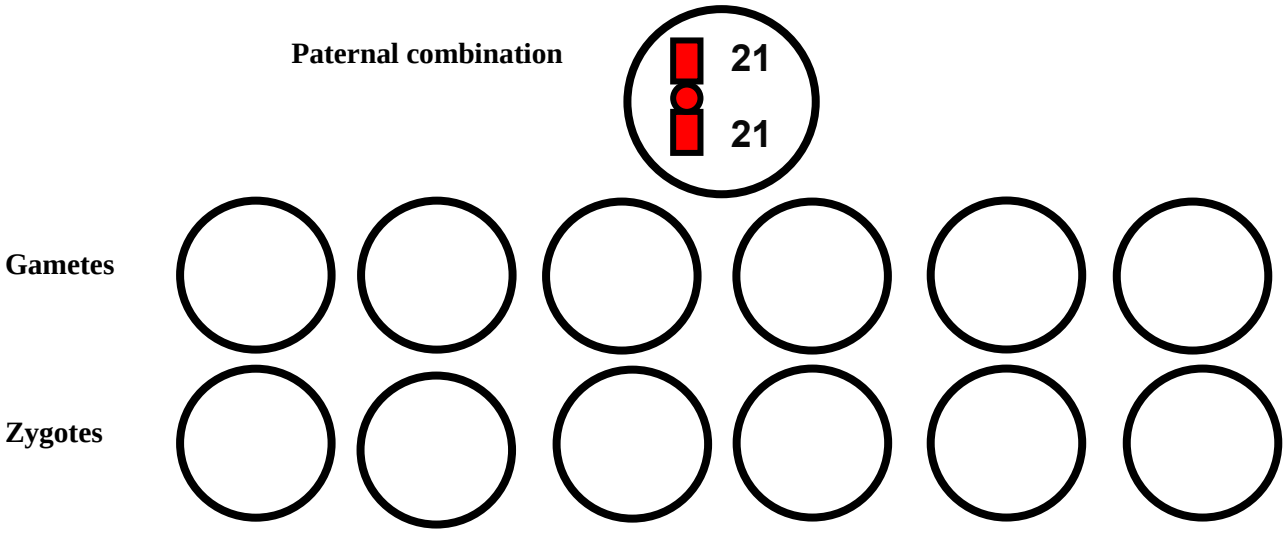


Gametes

Zygotes

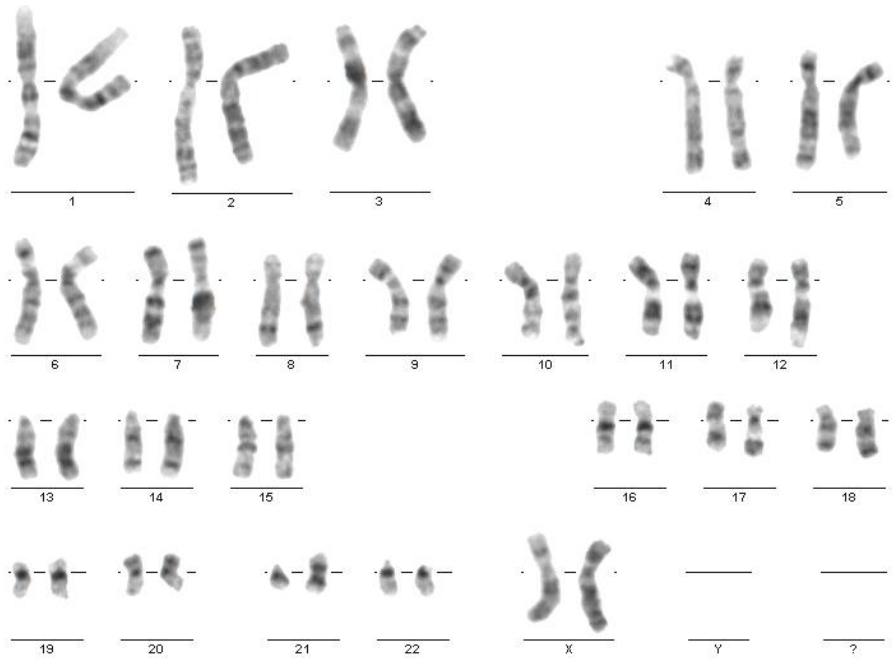
Theoretical risk of birth of child with Down syndrome:

Task 1: Draw all possibilities of meiotic chromosomal segregation in the **rob(21;21)** translocation carrier. Draw the corresponding zygotic chromosomal combinations (after fertilization with normal gamete). Solve the task analogous to the previous one. What is the theoretical risk of Down syndrome for the offspring of this rearrangement carrier?



Theoretical risk of birth of child with Down syndrome:

Task 2: A girl with translocation form of Down syndrome (her karyotype is shown under this text) was born to healthy parents. Cytogenetic examination of both parents showed that father is a carrier of rob(21;21) translocation. Write father's and daughter's karyotype and answer following questions:



Father's karyotype:

Daughter's karyotype:

Question 2-1: In the case of future pregnancy of this couple - would you indicate the invasive prenatal diagnostics? What is the expected proportion of pregnancies with Down syndrome fetus in this couple?

Question 2-2: What can modern medicine offer to this couple?

Task 3: Name the type of structural chromosomal aberration which changed the structure of the original chromosome **A-B-C-D•E-F-G-H** (Symbol • stands for centromere of the chromosome).

a) **A-B-C-D•E-F-H**

b) **A-B-C-E•D-F-G-H**

c) **A-B-C-D•D-C-B-A**

d) **A-C-B-D•E-F-G-H**

e) **A-B-B-C-D•E-F-G-H**

f) **B-C-D•E-F-G-H**

g) **A-B-D-C•E-G-H**

h) **A-B-C-C•E-F-G-H**