

# STRUKTURÁLNÍ ABERACE

ÚBLG 1.LF UK



# CHROMOSOMÁLNÍ ABERACE

## ▶ NUMERICKÉ



## ▶ MIXOPLOIDIE

- MOZAICISMUS
- CHIMÉRISMUS

# TYPY PŘESTAVEB

**BALANCOVANÉ  
(BEZ FENOTYPOVÝCH  
PROJEVŮ):**

INVERZE

TRANSLOKACE

INSERCE

**NEBALANCOVANÉ:**

DELECE

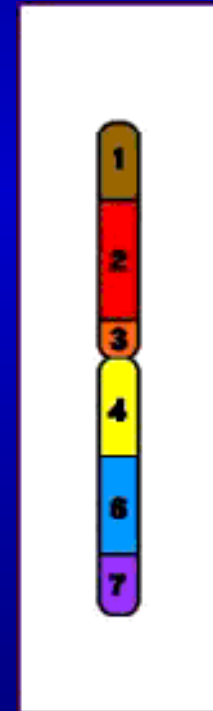
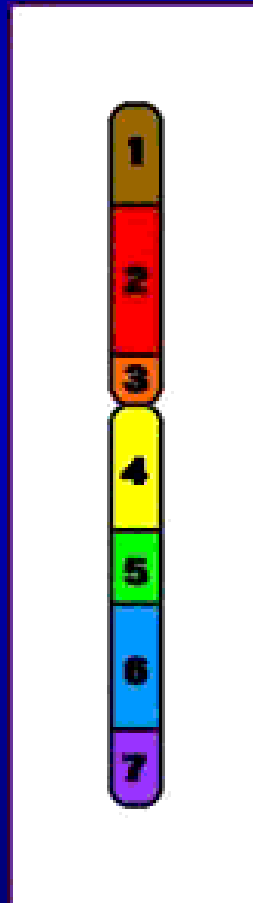
DUPLIKACE

RING CHROMOSOM

ISOCHROMOSOM

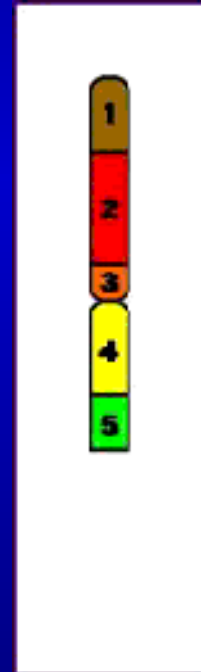
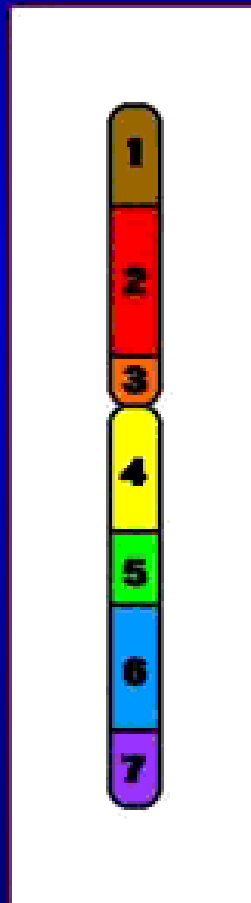
DICENTRICKÝ  
CHROMOSOM

# DELECE INTERSTICIÁLNÍ

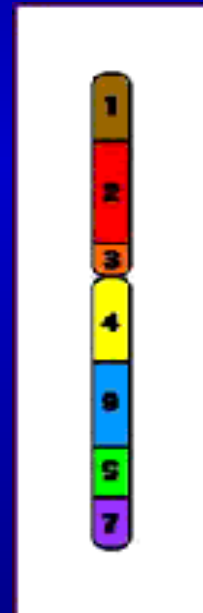
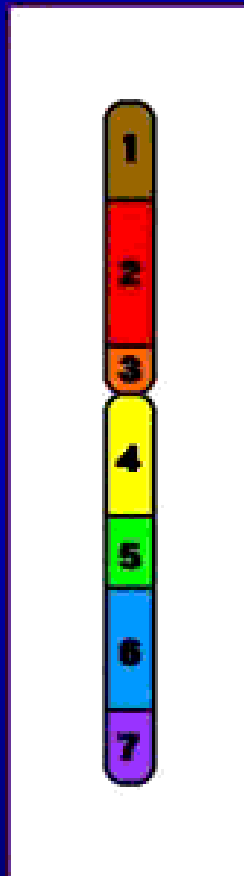


# DELECE

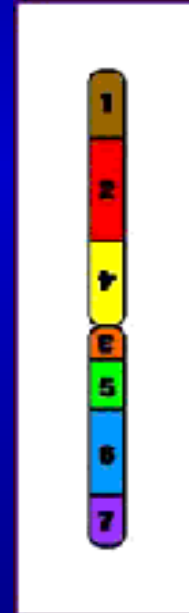
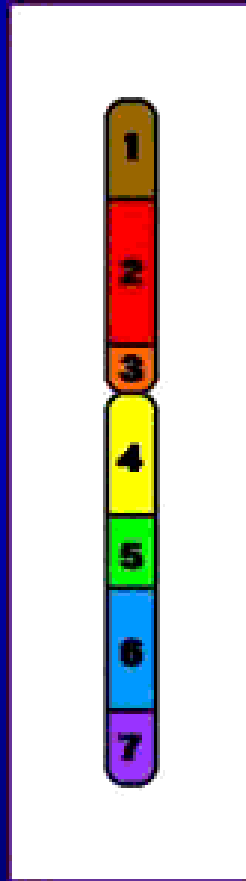
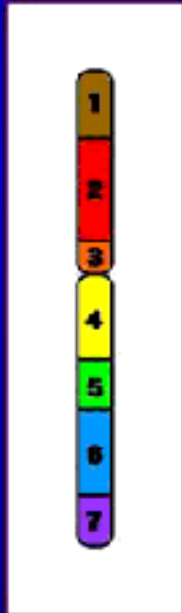
## TERMINÁLNÍ



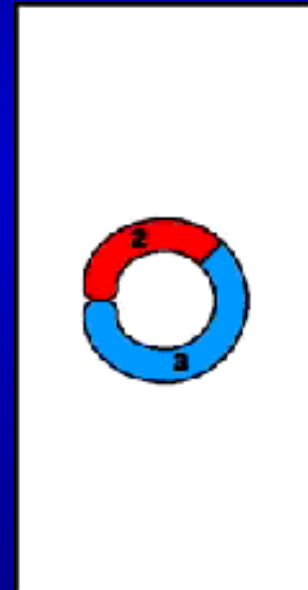
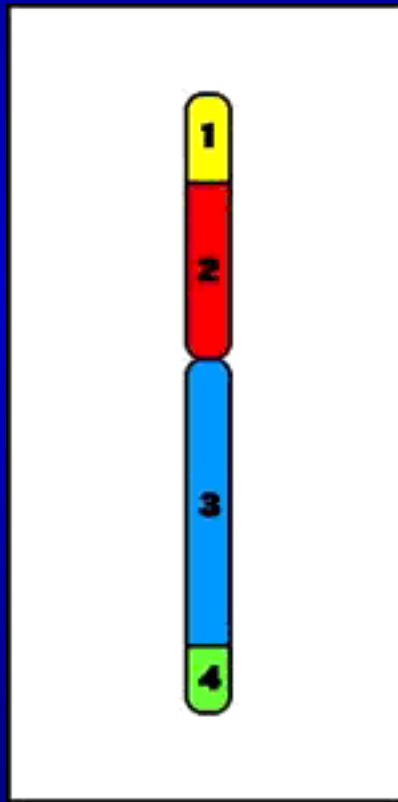
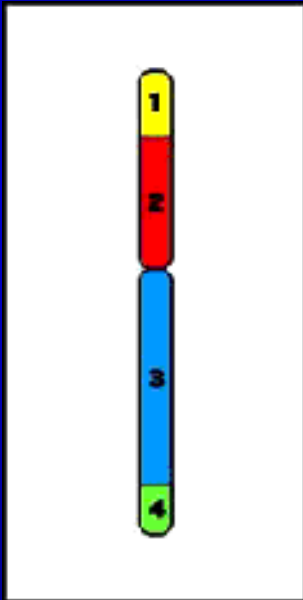
# INVERZE PARACENTRICKÁ



# INVERZE PERICENTRICKÁ

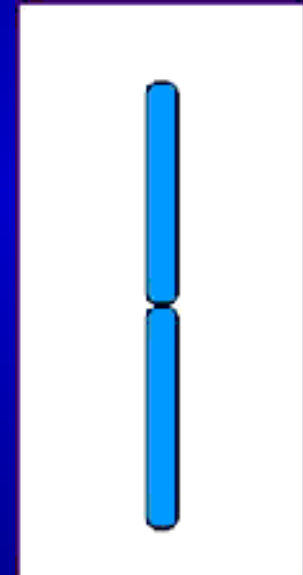
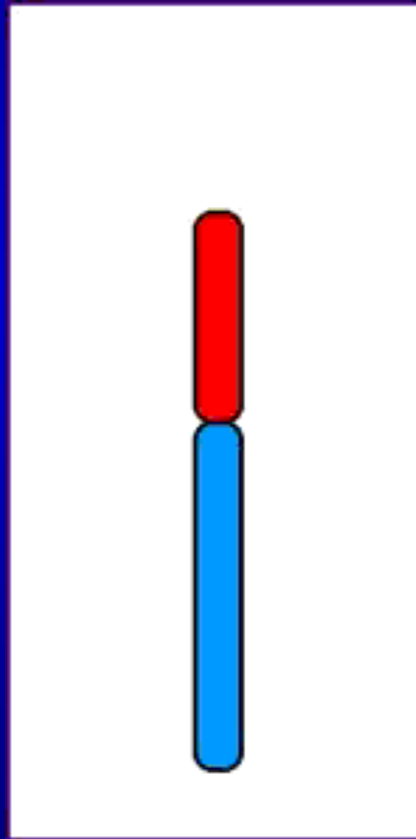
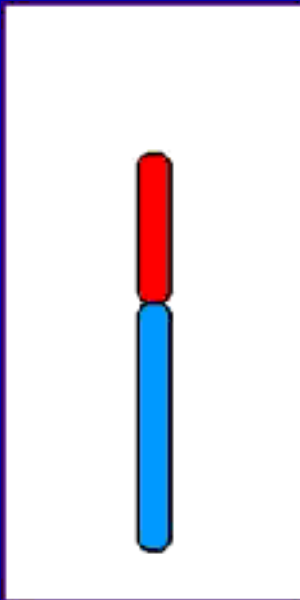


# RING CHROMOSOM

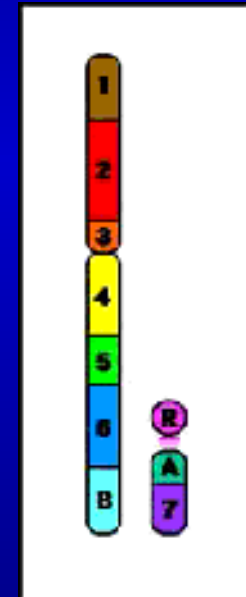
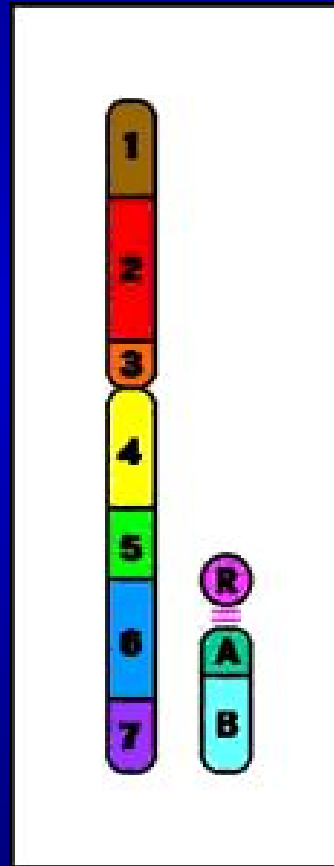
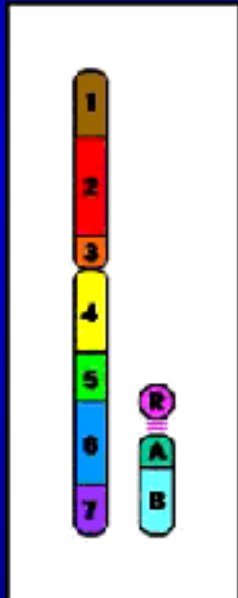




# ISOCHROMOSOM

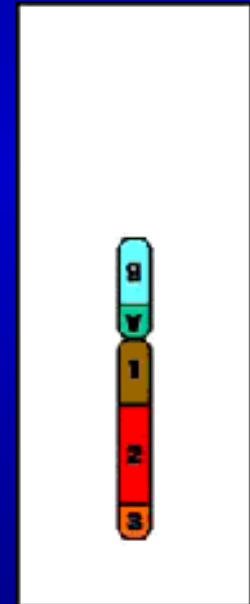
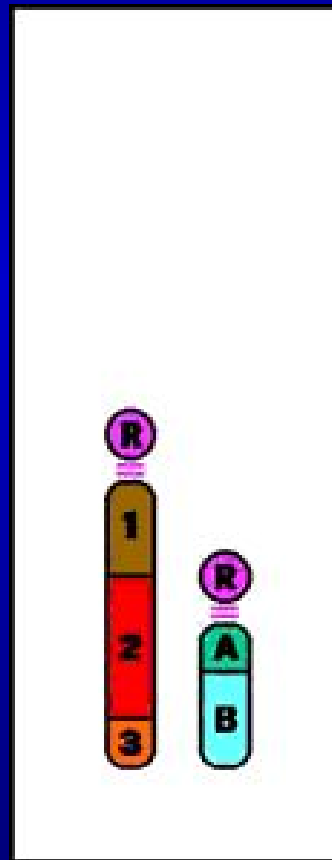
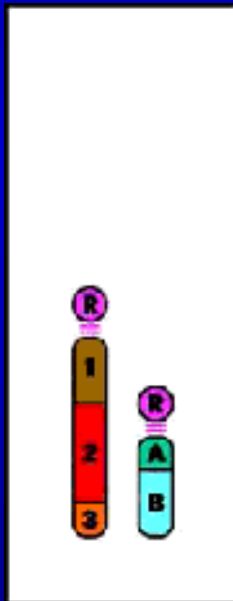


# TRANSLOKACE RECIPROKÁ



# TRANSLOKACE

## ROBERTSONOVA



# MEZINÁRODNÍ CYTOGENETICKÁ NOMENKLATURA ISCN - 2005, 2009 (příklady symbolů a zkratek)

**Normální lidský karyotyp:** 46, XX nebo 46, XY

**Numerické aberace:** 47, XXY; 45, X; 69, XXY

+/- před chromosomem nadpočetným/chybějícím 47, XX, +21

**Strukturní aberace – vybrané příklady:**

del - delece 46, XY, del(5)(p?) 46, X, del(X)(q?)

t - translokace (reciproká) 46, XY, t(2;8)(p?;q?)

der, rob - derivovaný chromosom (Robertsonova translokace)

45, XX, der(14;21) n. 45, XX, rob(14;21)

46, XY, der(21;21), +21 n. 46, XY, rob(21;21), +21

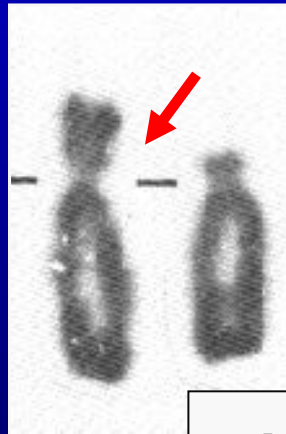
# TURNERŮV SYNDROM

## DELEČNÍ FORMA



46,X,del(Xp)

# CRI DU CHAT SYNDROM (SYNDROM „KOČIČÍHO KŘIKU“)



del(5p)

- mikrocefalie
- těžká tělesná a mentální retardace
- kulatý „měsícovitý“ obličej (v dětství)
- hypoplasie laryngu - vysoký pronikavý pláč (jako mňoukání kočky)



# PRADER-WILLIHO SYNDROM

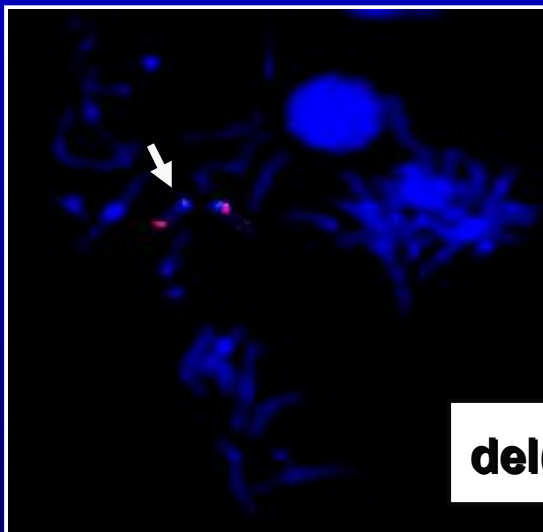


## NOVOROZENCI, KOJENCI:

- těžká hypotonie
- neprospívání, opoždění vývoje

## POZDĚJI:

- mentální retardace
- polyfagie - těžká obezita s následky (DM, poruchy oběhu,...)
- hypogenitalismus
- poruchy chování

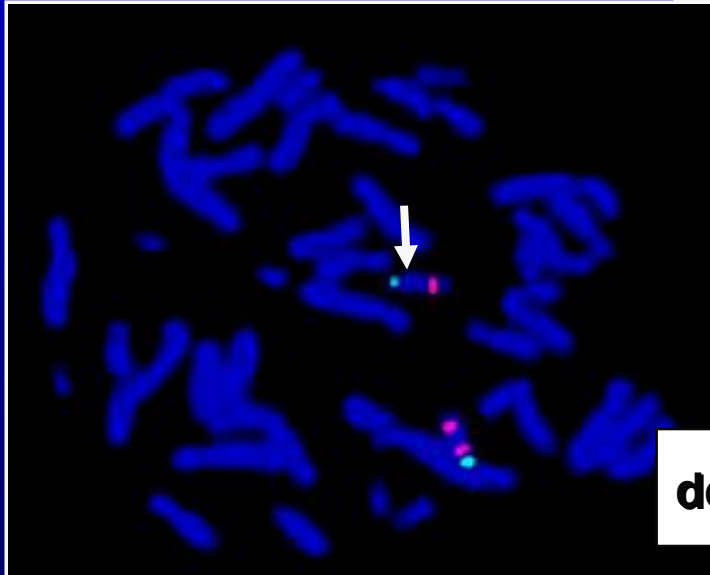


**del(15)(q11-13)pat**

# ANGELMANŮV SYNDROM



- těžká mentální retardace
- chybějící vývoj řeči
- nemotivované záchvaty smíchu
- „škubavé“ pohyby končetin
- toporná chůze
- syndrom „happy puppet“



**del(15)(q11-13)mat**



# DOWNŮV SYNDROM

## TRANSLOKAČNÍ FORMA



M. DOWN:

95% prostá trisomie chr. 21

4-5% translokační forma

(Robertsonova translokace)



<1% mozaicismus

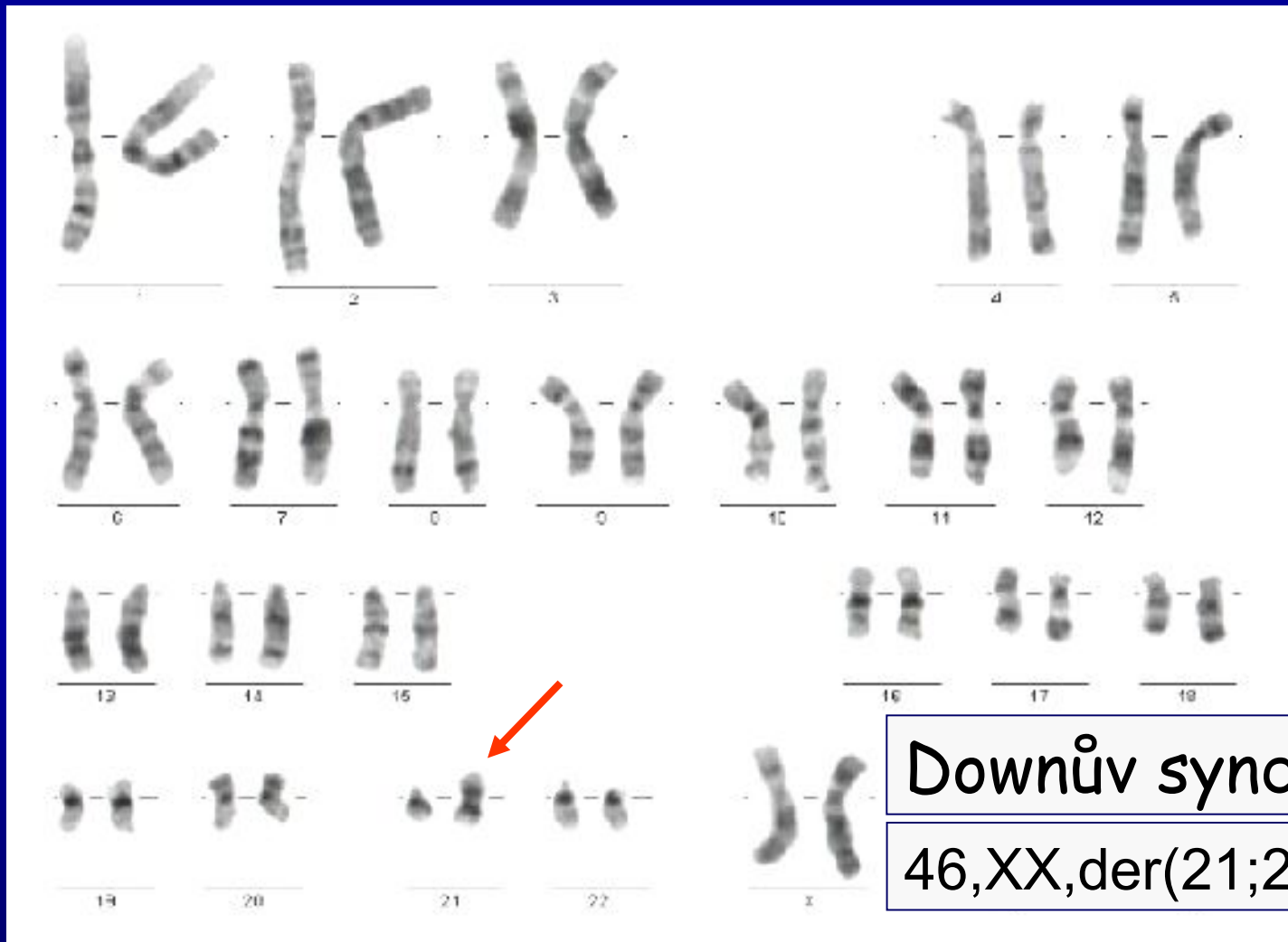
# DOWNŮV SYNDROM

## TRANSLOKAČNÍ FORMA



46,XY,der(14;21),+21

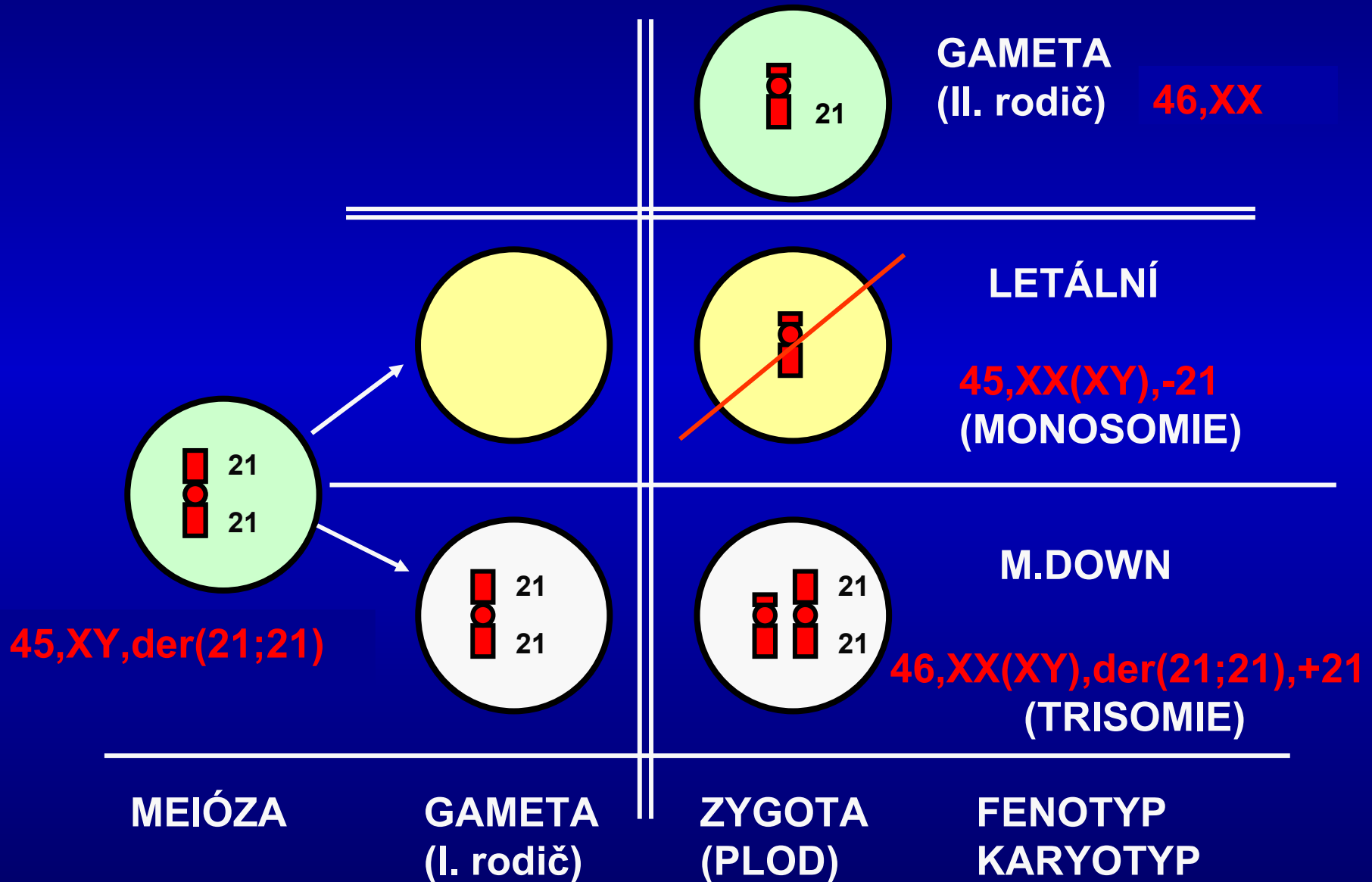
Zhodnot'te karyotyp novorozence se znaky  
Downova syndromu (př.12, str. 43):



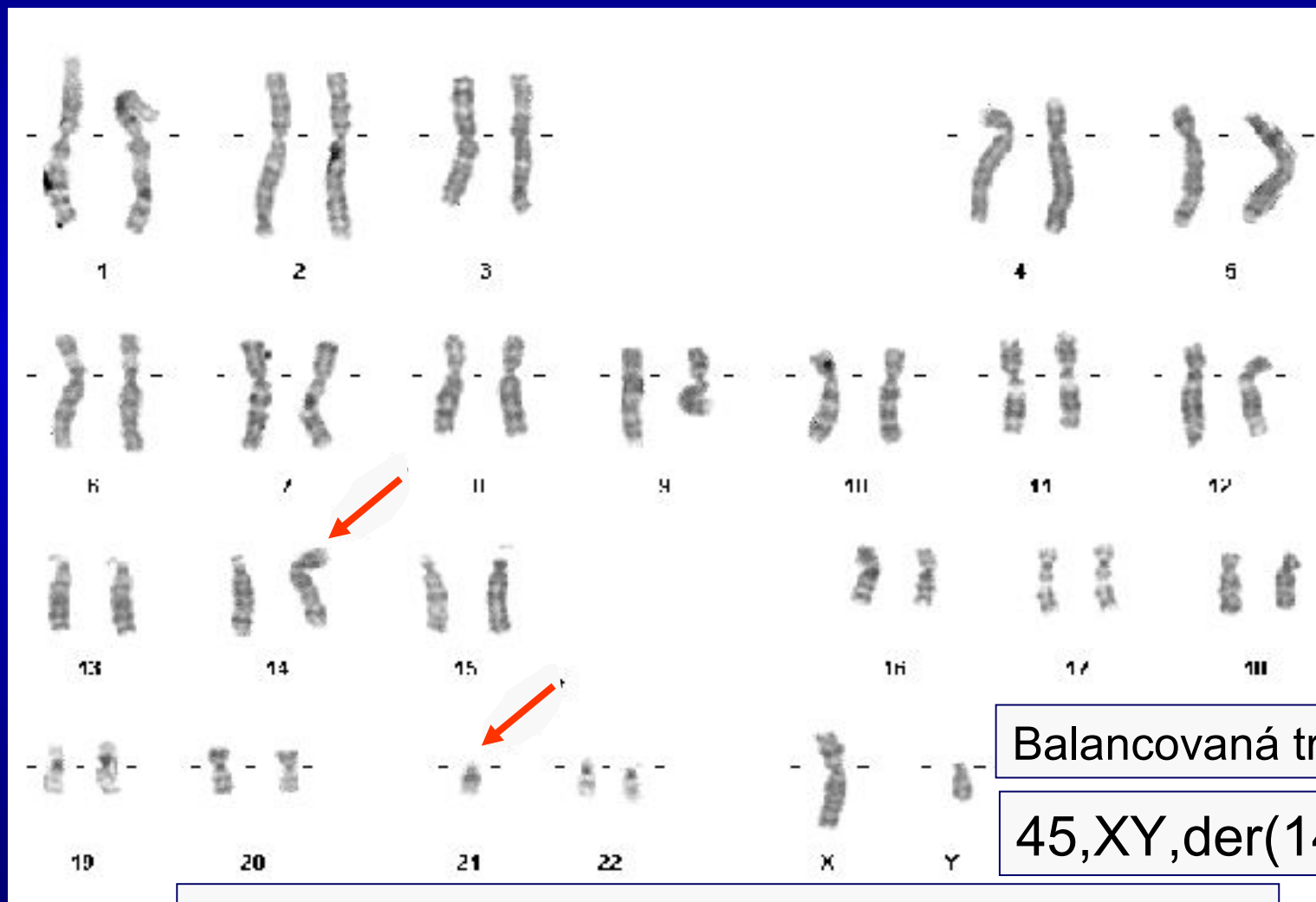
Downův syndrom

46,XX,der(21;21),+21

Riziko: teoretické 100%, empirické 100%



Zhodnot'te karyotyp otce dítěte se znaky Downova syndromu, matka 46,XX (př.11, str. 42):

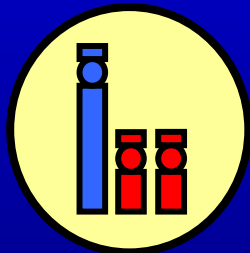
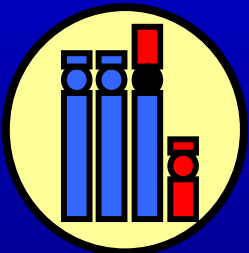
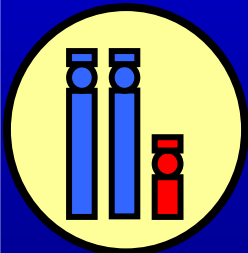
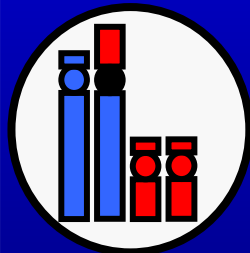
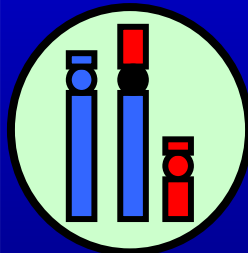
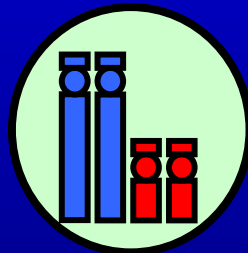
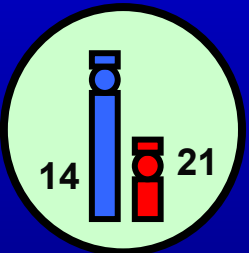
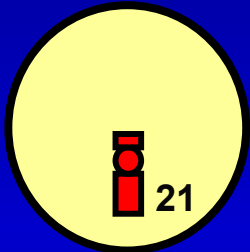
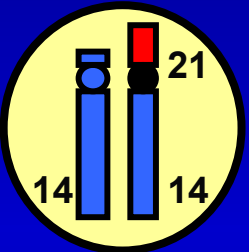
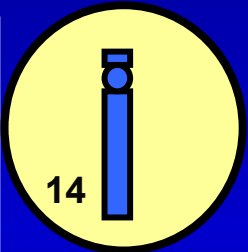
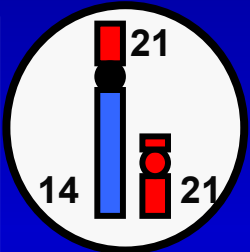
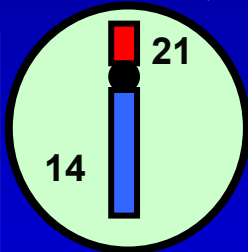
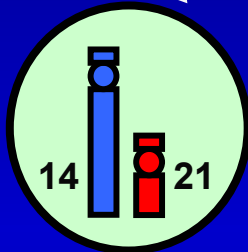
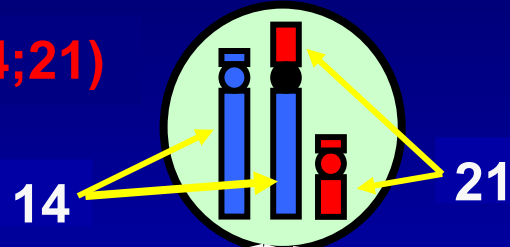


Balancovaná translokace

45,XY,der(14;21) - otec

46,XX(XY),der(14;21),+21 – dítě M. Down

45,XY,der(14;21)



46,XX

NORMA

BALANCOVANÁ  
TRANSLOKACE

M.DOWN

33,3%

LETÁLNÍ

# MORBUS DOWN

| PROBAND                                                     | RODIČE                                            |             | RIZIKO                                                                               |
|-------------------------------------------------------------|---------------------------------------------------|-------------|--------------------------------------------------------------------------------------|
| $47, X^X/Y, +21$                                            | $46, X^X/Y$                                       | $46, X^X/Y$ | > než populační<br>závisí na věku matky                                              |
| $46, X^X/Y, der(21;21), +21$                                | $45, X^X/Y, der(21;21)$                           | $46, X^X/Y$ | 100% TEORETICKÉ<br>100% EMPIRICKÉ                                                    |
| $46, X^X/Y, der(D;21), +21$<br>$46, X^X/Y, +21, der(21;22)$ | $45, X^X/Y, der(D;21)$<br>$45, X^X/Y, der(21;22)$ | $46, X^X/Y$ | 33,3% TEORETICKÉ<br>EMPIRICKÉ:<br>cca 5% - nositel otec<br>cca 15% - nositelka matka |
| $46, X^X/Y, der(D;21), +21$<br>$46, X^X/Y, +21, der(21;G)$  | $46, X^X/Y$                                       | $46, X^X/Y$ | NOVÁ MUTACE<br>NONPATERNITA                                                          |
| $47, X^X/Y, +21$                                            | $47, X^X/Y, +21 / 46, X^X/Y$                      | $46, X^X/Y$ | MOZAIKA – závisí na<br>zastoupení linií<br>aberovaných a normálních<br>buněk         |