GENETICS

LECTURE 3

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ISCN – An International System for Human Cytogenomic Nomenclature

46,XX[20]

number of chromosomes

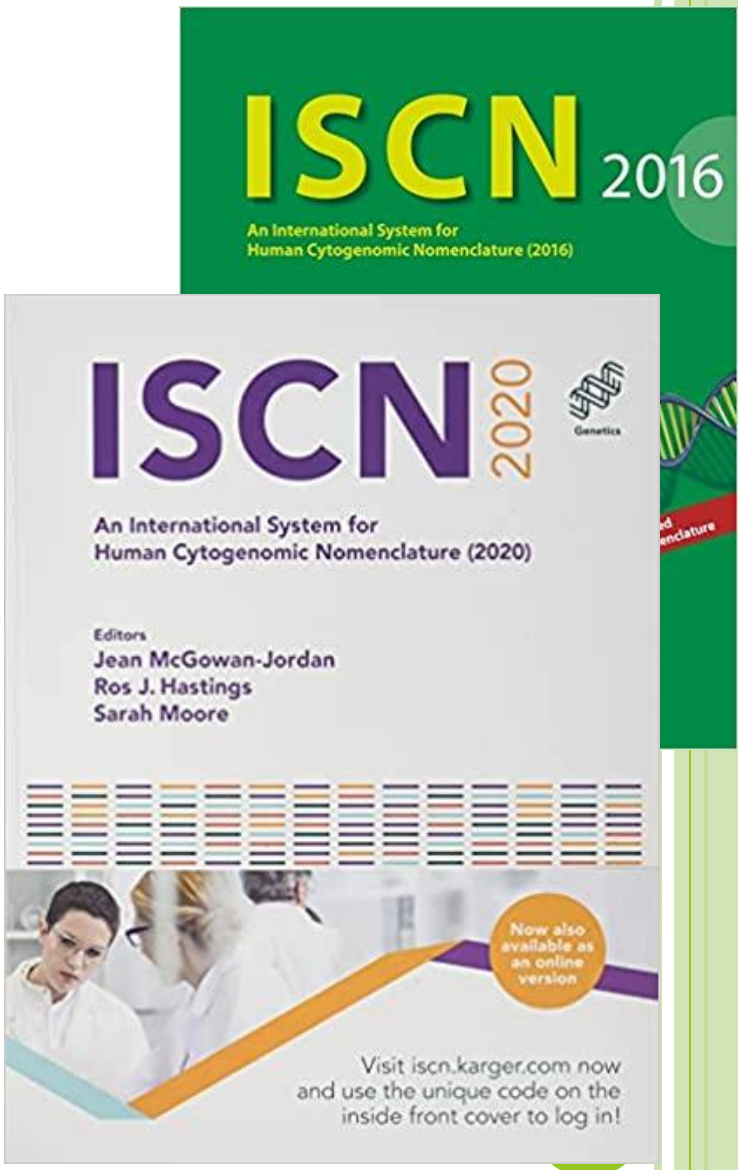
sex chromosomes

number of analysed metaphases

45,X

47,XXY

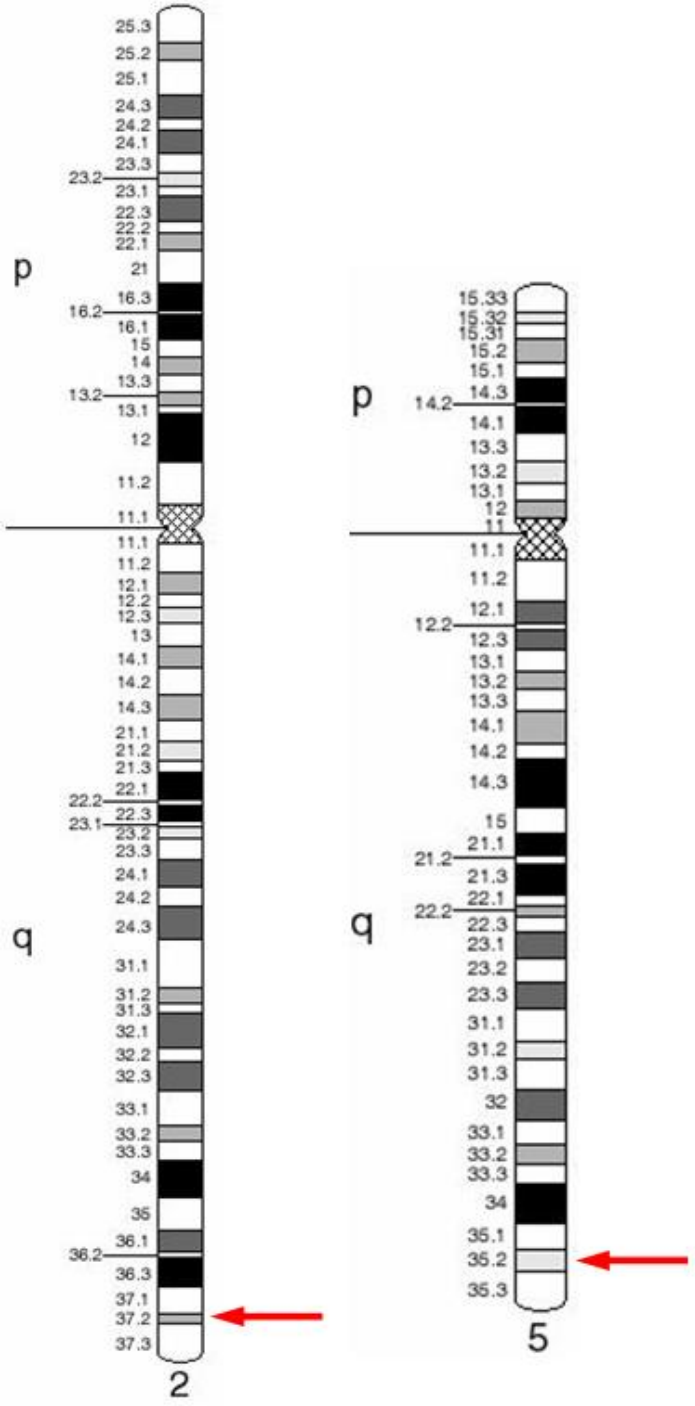
47,XY,+21



46,XX,t(2;5)(q37.2;q35.2)

type of aberration and
involved chromosomes

breaking points



The most common abbreviations:

- p – short arm
- q – long arm
- add – additional material of unknown origin
- ter – telomere
- pter – end of short arm
- qter – end of long arm
- cen – centromere
- del – deletion
- der – derivative chromosome
- dup – duplication
- i – isochromosome
- ins - insercion
- inv – inversion
- r – ring chromosome
- t – translocation

Indications for performing constitutional karyotype

- Persons with suspected recognizable chromosome syndrome (e.g. Down syndrome)
- Persons with an unrecognizable pattern of two or more malformations
- Persons with ambiguous genitalia
- Mental retardation or development delay in children who have multiple physical abnormalities
- Parents and children of persons with chromosomal translocations, deletions, or duplications

Indications for performing constitutional karyotype

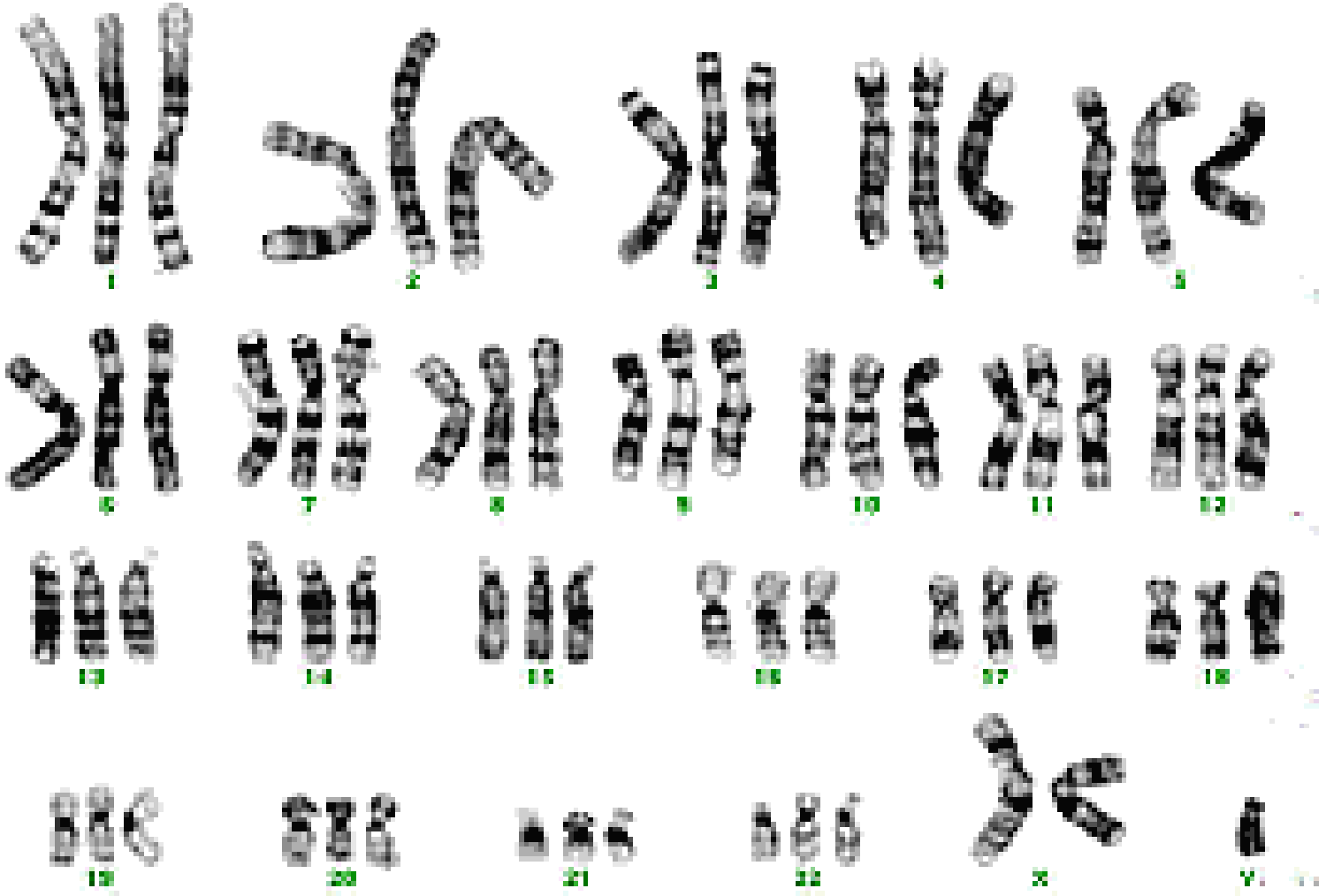
- Stillborn infants with malformation or with no recognizable reason for fetal death
- Recurrent miscarriages
- Primary infertility
- Females with proportionate short stature and primary amenorrhea
- Males with small testes or significant gynecomastia

Indications for performing acquired karyotype

- Diagnostics, determination of the diagnosis
- Establishment of prognosis (good or poor)
- Choice of the right treatment method
- Monitoring - assessment of treatment efficacy
- Assessment of the course of the disease - evolution

What kind of aberration is it???

1



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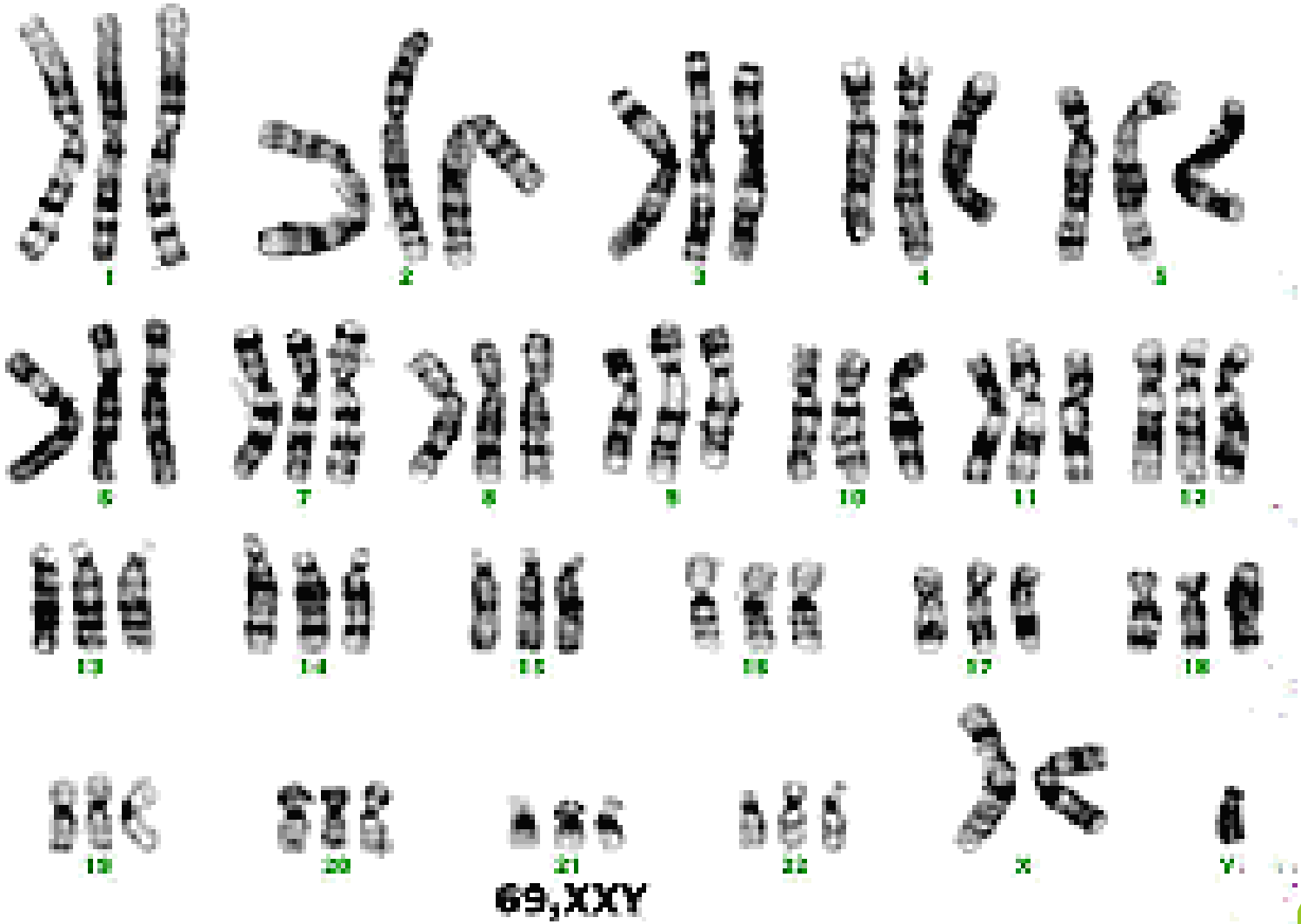

TRIPLOIDY

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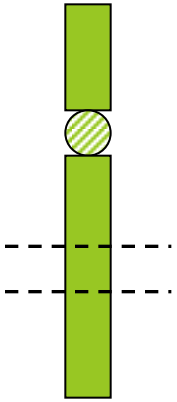
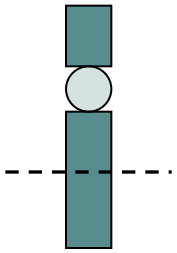
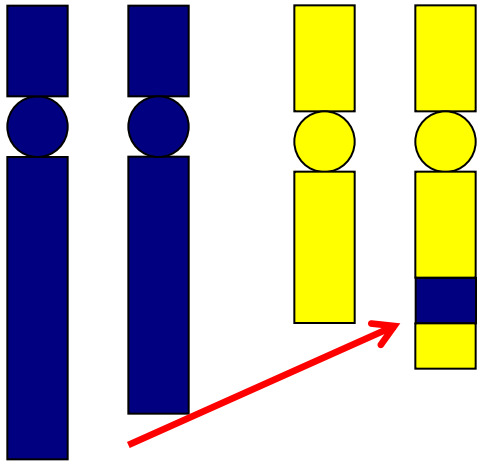


2 breaks in „donor” chromosome



1 break in „recipient” chromosome



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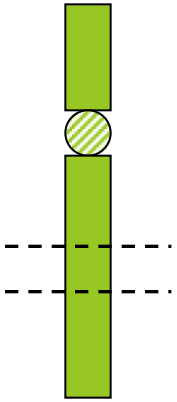
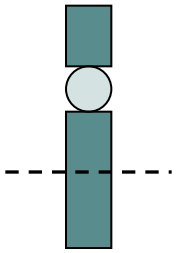
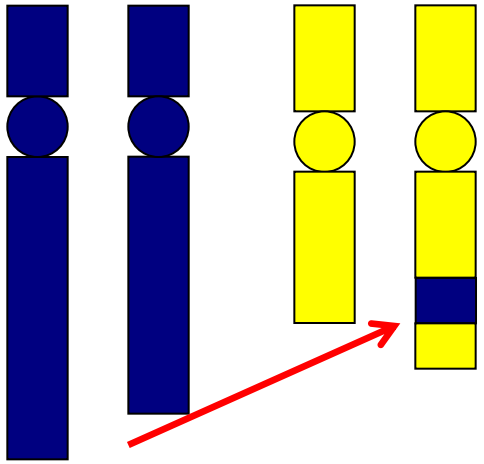


2 breaks in „donor” chromosome



1 break in „recipient” chromosome



INSERTION

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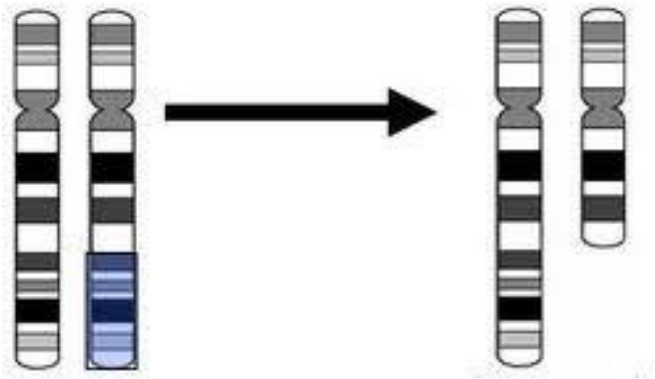

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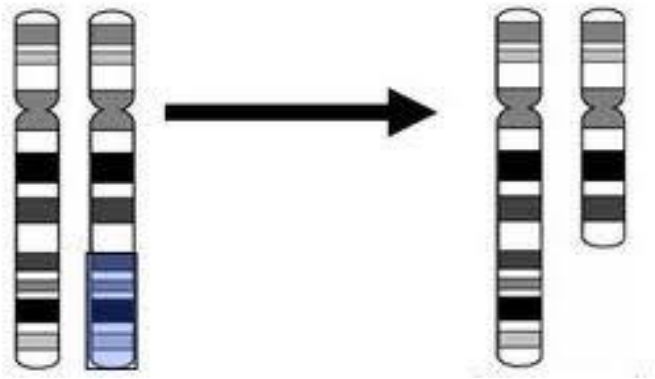

TERMINAL DELETION

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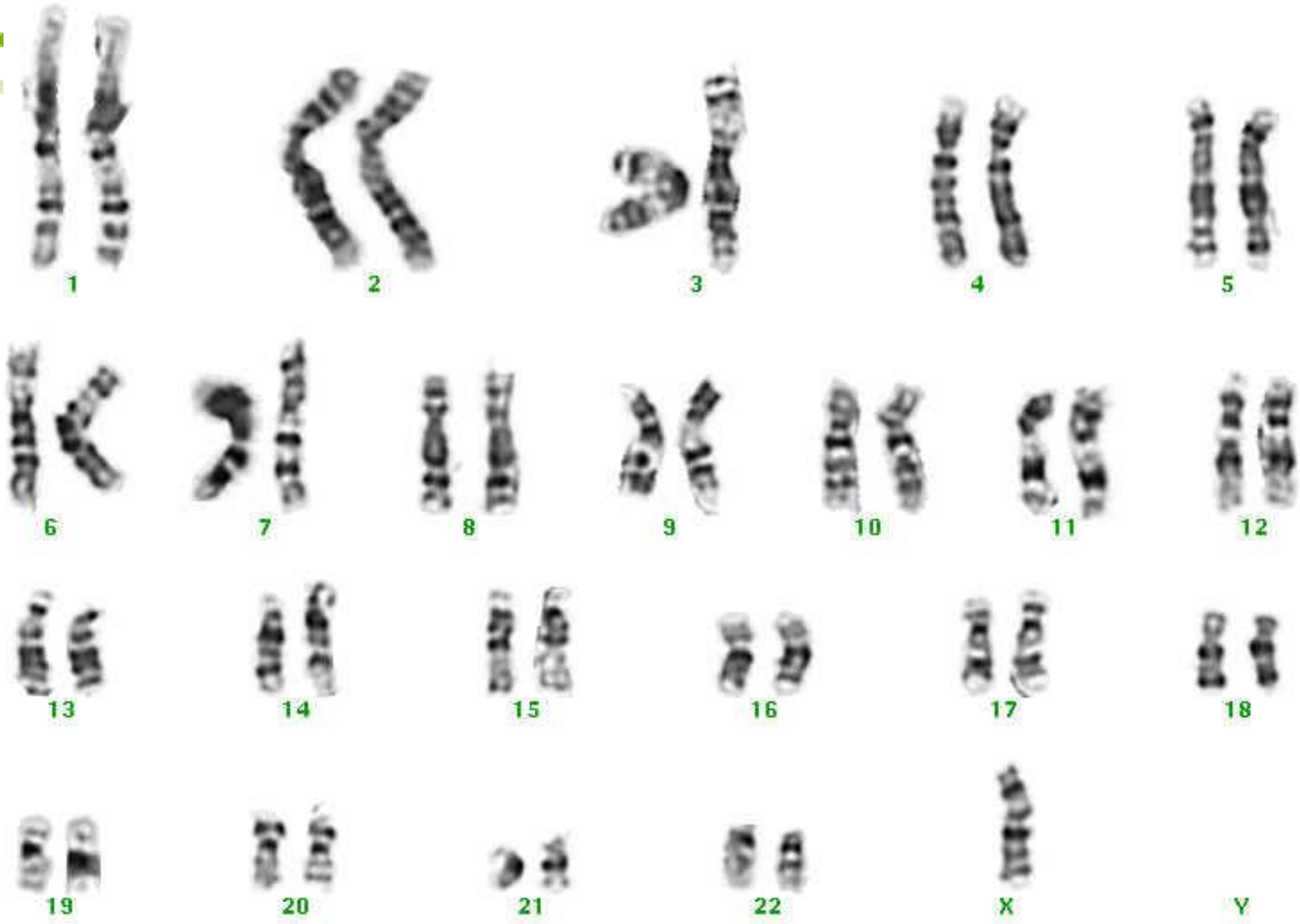

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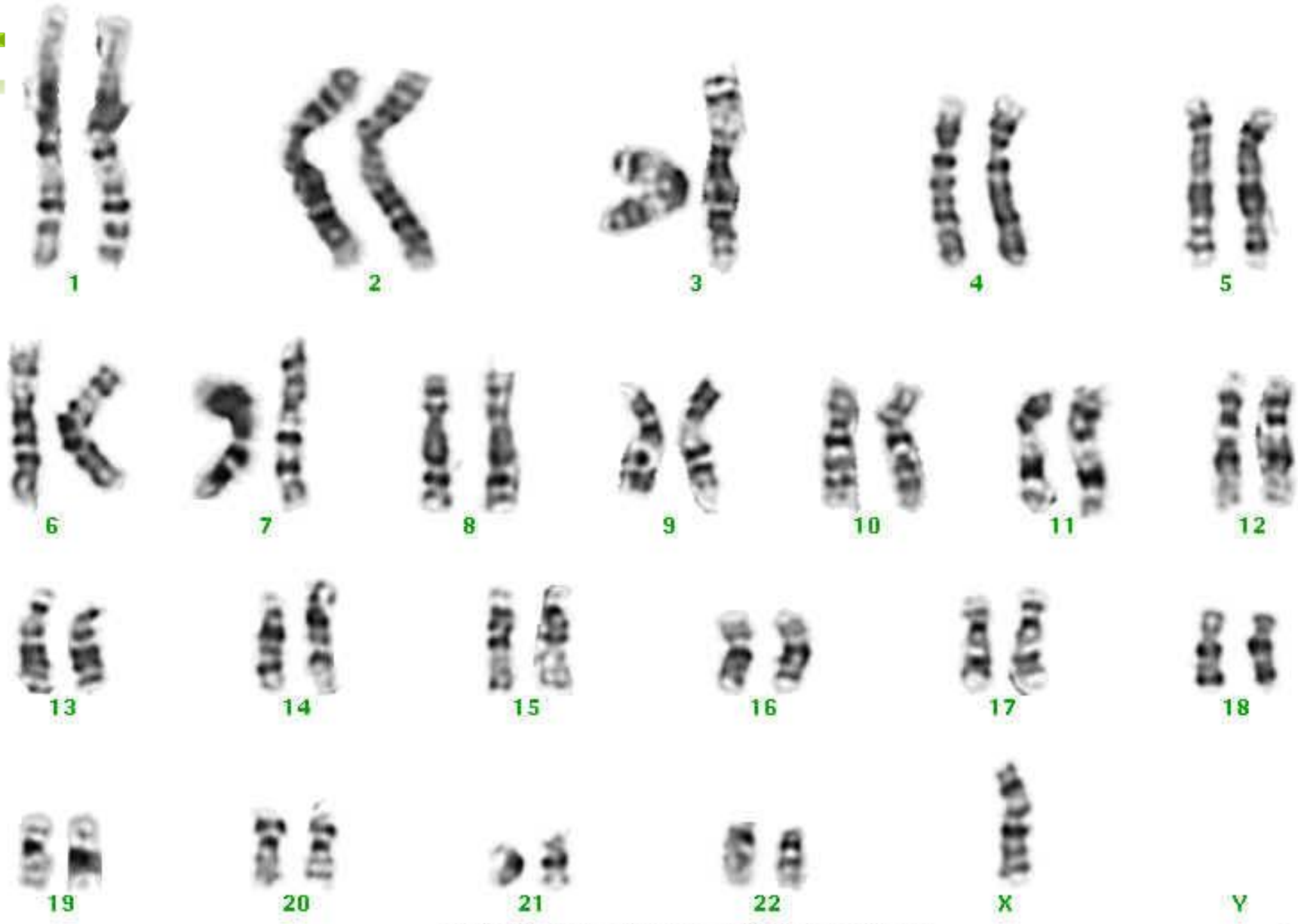
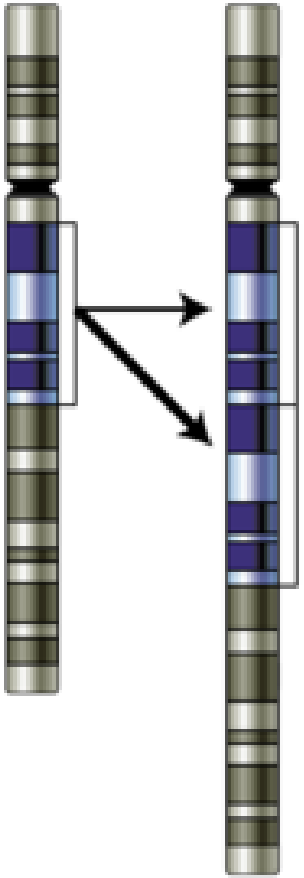


45,X (Turner syndrome)

MONOSOMY

5



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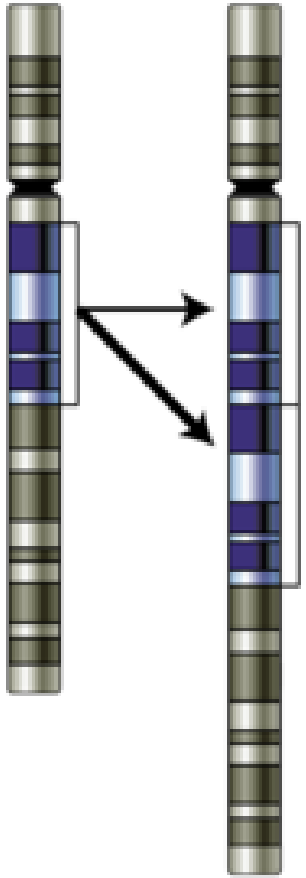

DUPLICATION

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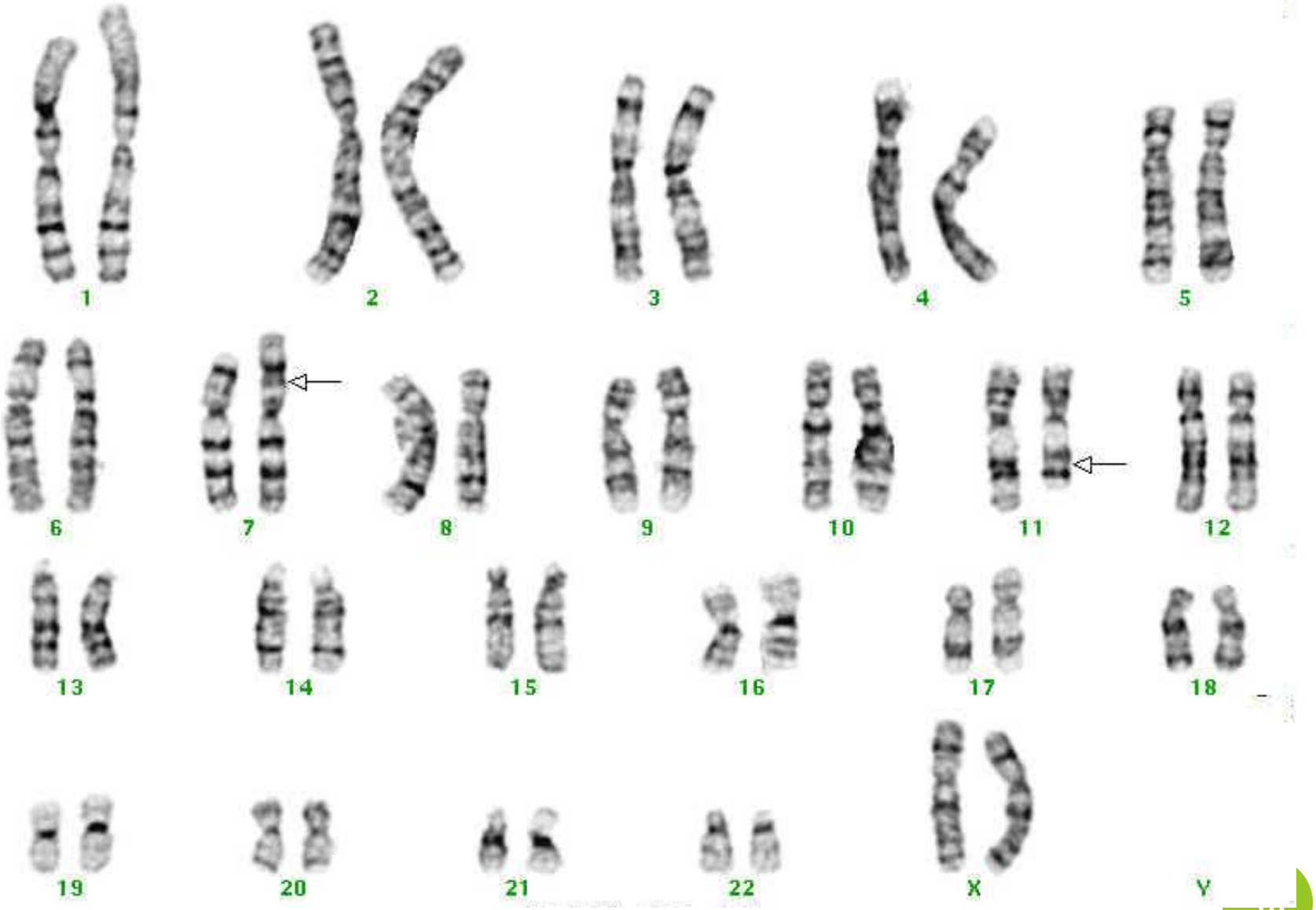

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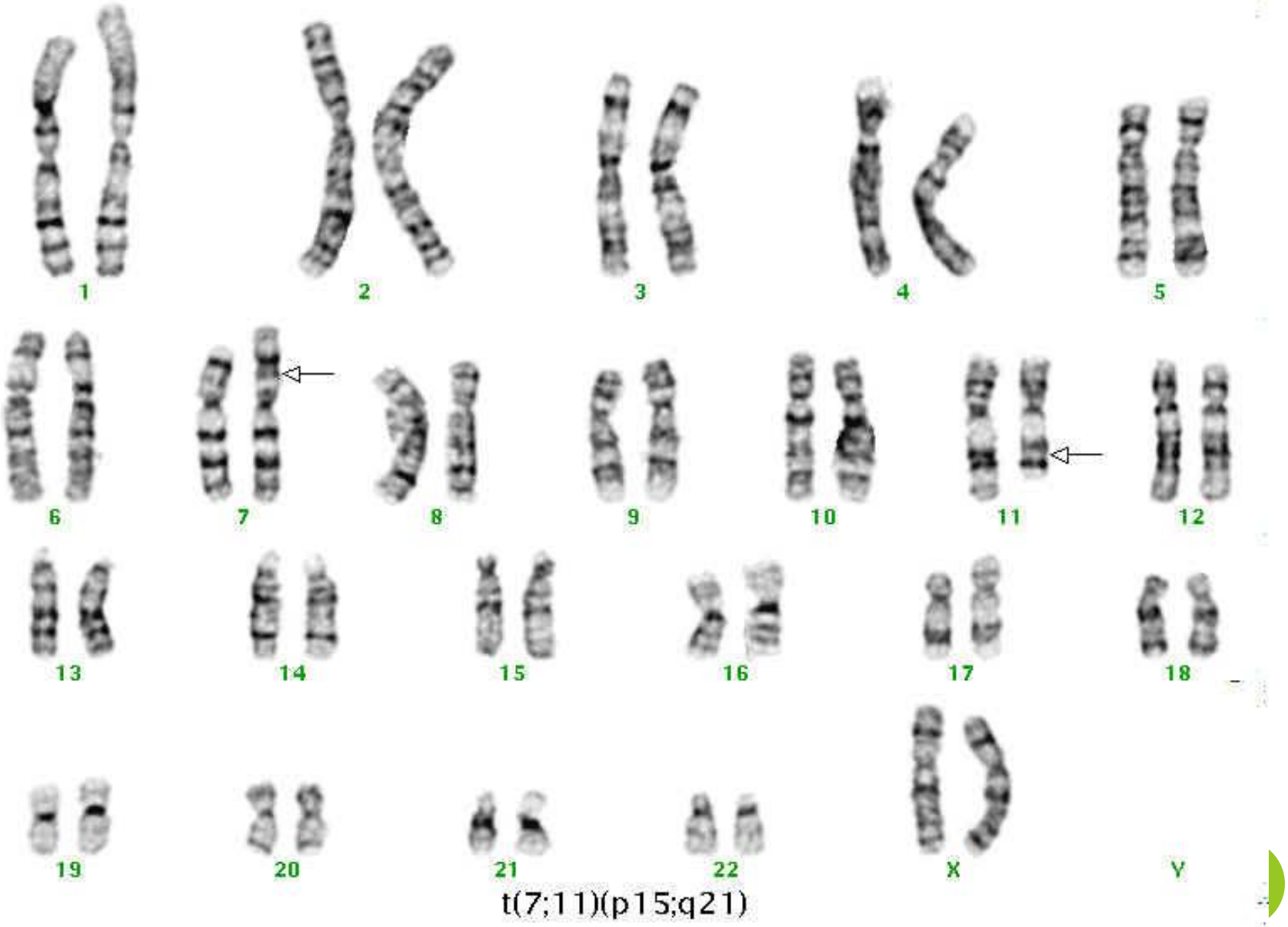

TRANSLOCATION

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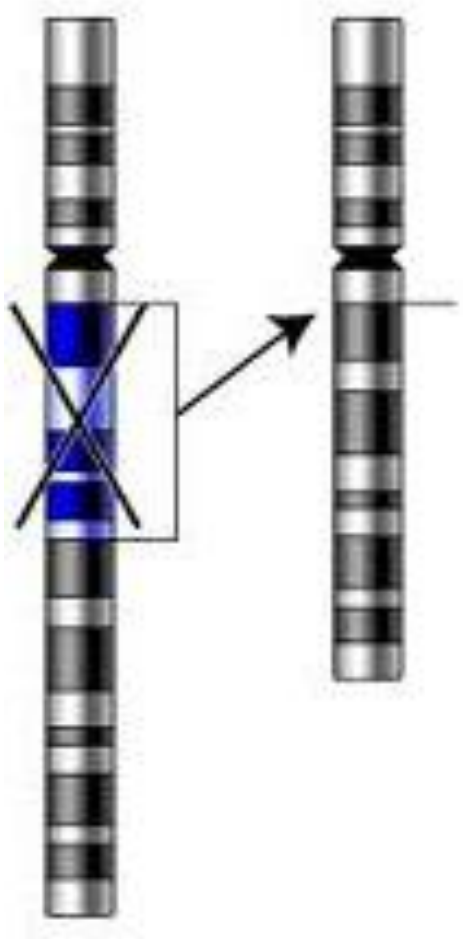

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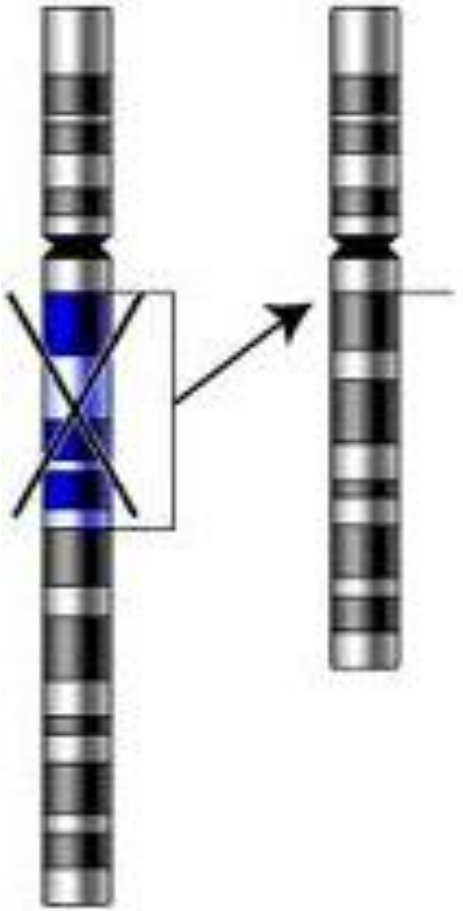
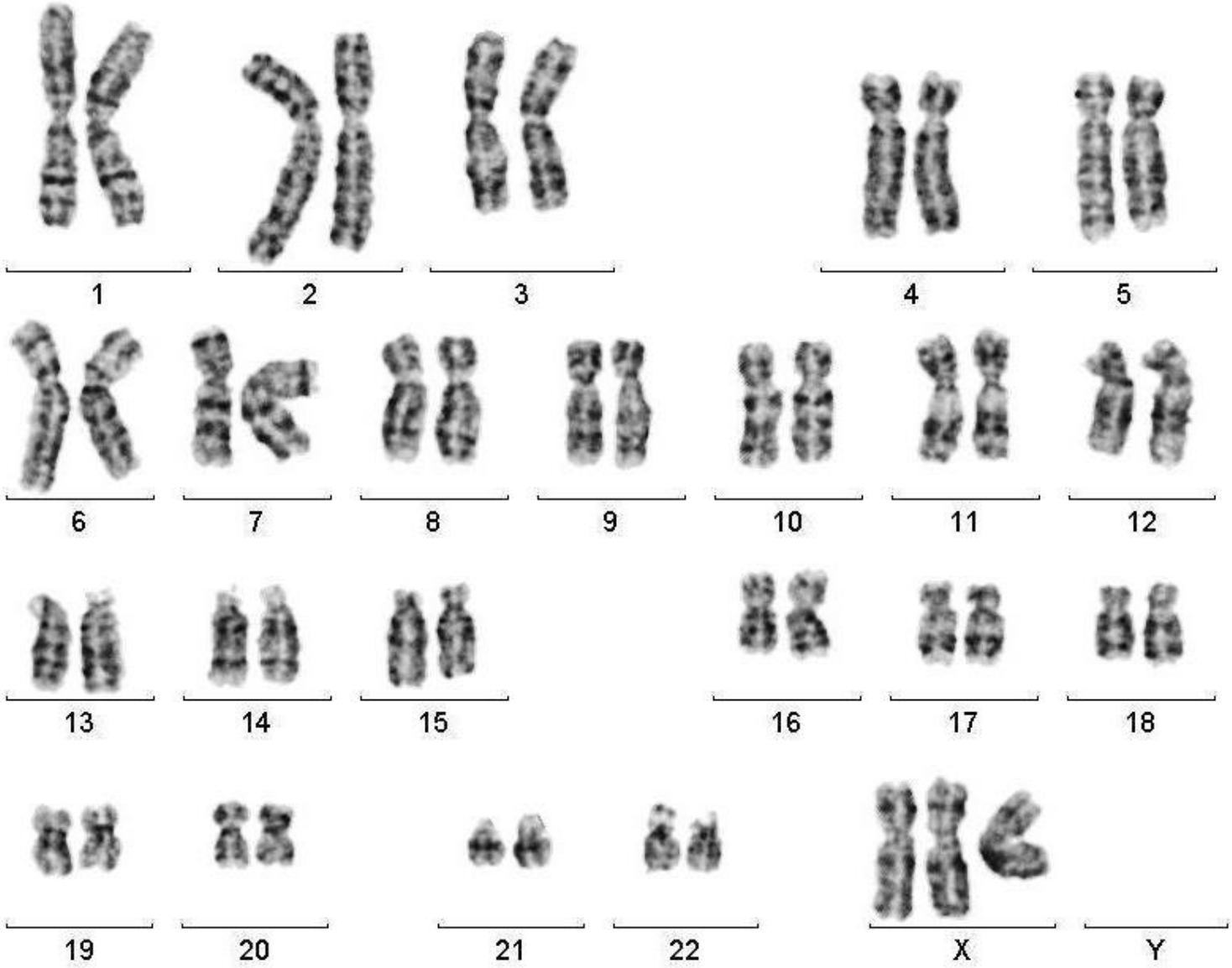


INTERSTITIAL DELETION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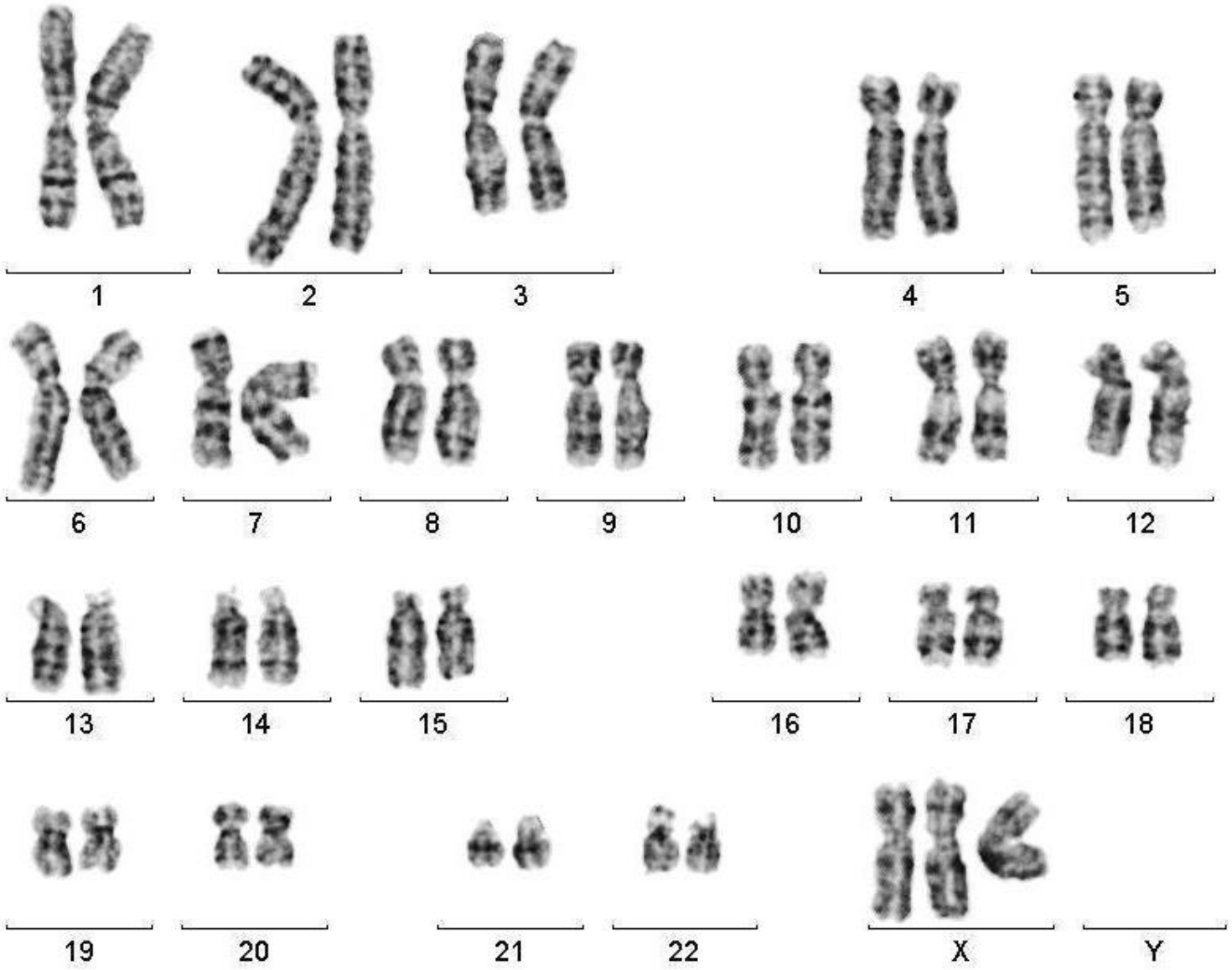

TRISOMY

Triple X syndrome – 47,XXX