

Basic Cytogenetics

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A short history about Cytogenetics....

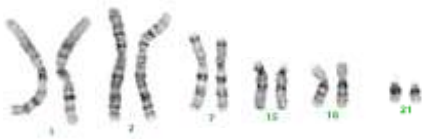
When?	Who?	What?
1888	Waldeyer	Used first the term chromosome
1912	Winiwarter	Founder of human cytogenetics
1923	Painter	Studied testicular material – 24 haploid chromosomes; Diploid number = 48
1952	Hsu	Hypotonic treatment to spread chromosomes
1956	Tjio and Levan	Correct number of human diploid chromosomes
1971	Caspersson	Banding techniques – Q bands
1977	Rudkin and Stollar	Hybridization with fluorescent probes



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Human Chromosomes



Group	Chromosomes	Type
A	1-3	Metacentric big
B	4-5	submetacentric big
C	6-12 e X	Submetacentric medium
D	13-15	Acrocentric medium
E	16-18	Submetacentric small
F	19-22	Metacentric small

Metacentric
Submetacentric
Acrocentric



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(GTL-bands by Trypsin using Lieshman)



C- Banding



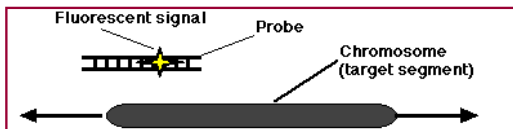
NOR staining



DA/DAPI staining



Fluorescence *in situ* hybridization - FISH



FISH

Microscopic view showing multiple fluorescently labeled probes hybridizing to a target sequence on a chromosome. Each chromosome in the karyotype pair can be identified.

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FISH probes

- Beta satellite
- Alpha satellite
- Classical satellite
- Telomeric sequences
- Unique gene sequences
- Partial chromosome paints
- Whole chromosome paints

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locus specific

centromere

painting

subtelomeric

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Reciprocal translocation

Robertsonian translocation

Aneuploidy screening

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Chromosome abnormalities

- Numerical
 - Aneuploidies
 - Polyploidies
- Structural
 - Deletions, duplications, inversions,
 - Markers, Dicentric, Rings chromosomes,
 - Reciprocal translocations
 - Robertsonian translocation

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Trisomies
most commons in newborns...

Trisomy 21 (Down Syndrome)- (47,XX ou XY, +21)

Trisomy 13 (Patau Syndrome)- (47,XX ou XY, +13)

Trisomy 18 (Edwards Syndrome) - (47,XX ou XY, +18)

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Trisomies
most commons in spontaneous abortions...

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45,X (Turner Syndrome)
Cause: 70-80% loss X paternal

47,XXX (Triple X Syndrome)
Cause: Meiosis I maternal

47,XXY (Klinefelter Syndrome)
Cause: 50% meiosis I paternal

47,XYY (XYY Syndrome)
Cause: Meiosis II paternal

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Meiosis, 1st division

Meiosis, 2nd division

Gametes

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DELETIONS

DUPLICATIONS

ISOCROMOSSOMES

RINGS

DICENTRICS

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PERICENTRIC INVERSION

Normal Inverted chromosome

Meiosis Loop

Deletions and duplications

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PARACENTRIC INVERSION

Normal Inverted chromosome

Meiosis Loop

Dicentric and acentric chromosomes

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RECIPROCAL TRANSLOCATION

11 der(11) 22 der(22)

ROBERTSONIAN TRANSLOCATION

14 21 der(14;21)

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RECIPROCAL TRANSLOCATION

Adjacent-1

Adjacent-2

Alternate

2:2 segregations

Alternate Adjacent-1 Adjacent-2

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SEGREGATIONS

ALTERNATE

Normal Balanced

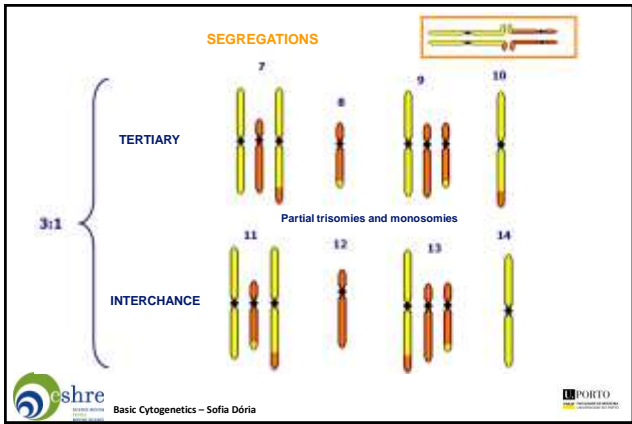
ADJACENT

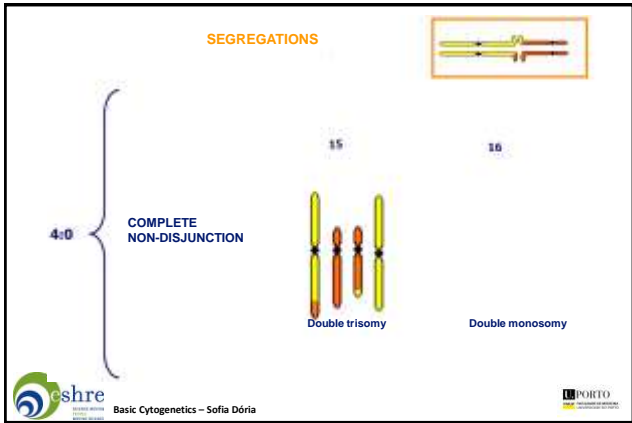
Adjacente 1 Adjacente 2

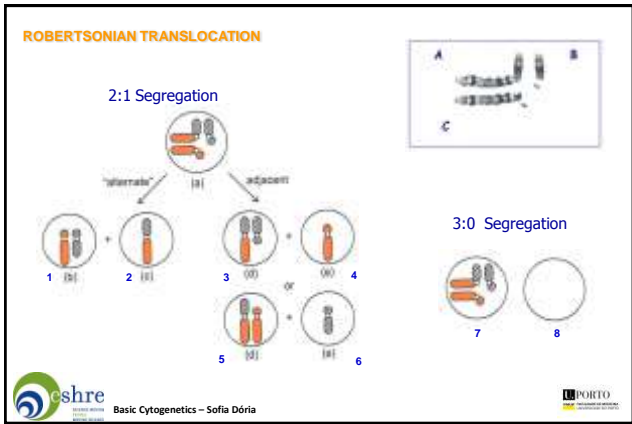
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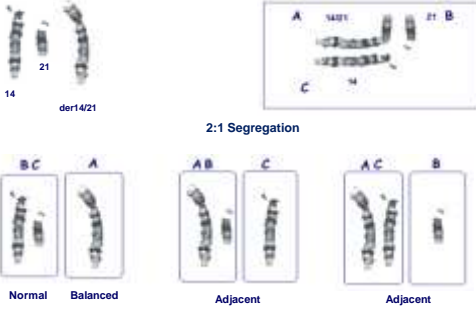
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ROBERTSONIAN TRANSLOCATION



Karyotyping 11857 patients with Infertility

Chromosome abnormalities found = 360 (3%)
(225 men, 135 women)

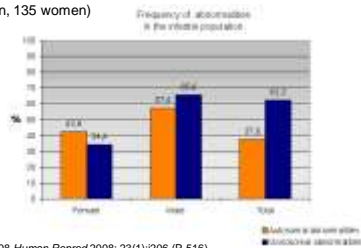
Most frequent Chromosome abnormalities:

- Sex aneuploidies = 140 (38.9%)
- Reciprocal translocations = 77 (21.4%)
- Robertsonian translocations = 34 (9.4%)

Dória S et al. 2008. *Human Reprod* 2008; 23(1):206 (P-516)

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Karyotyping 11857 patients with Infertility

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Abnormalities	Men	Women	Total
Structural abnormalities			
Reciprocal translocations	48	27	75
Non-reciprocal translocations	7	27	34
Inversions	4	9	13
Other structural abnormalities	7	5	12
Clonal abnormalities			
Structural abnormalities of the Y chromosome	8	0	8
Structural abnormalities of the X chromosome	8	22	30
Polyploid	2	1	3
Aneuploid	8	3	11
Normal karyotype	4	104	108
Normal karyotype	22	9	31
Not available	13	26	39
Total	118	168	286

Dória S et al. 2008. *Human Reprod* 23(1):206 (P-516)



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Thank you!



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