Topics for Self-Study: *Human karyotype, types of human chromosomes, cytogenetic examination in medicine*

Method	Coverage	Advantages	Disadvanteges	Resolution (limits)
Karyotype	All chromosomes	Cost, availability	Low resolution level	> 5 Mb
FISH	Selected locus (loci)	Cost, accuracy	Extremely limited coverage	100-300 kb (probe size)
MLPA	Selected loci	Accuracy	Limited coverage	20-300 kb
Array-CGH / SNP-array	All chromosomes	High accuracy, high resolution level	Expensive, difficult to interpret	approx.100 kb

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Task 1:

Analyse the following photographs showing the results of cytogenetic examinations for different individuals. The photographs can be found online (https://ublg.lf1.cuni.cz/en/clinical-cytogenetics). For every case – determine the **cytogenetic diagnosis** (following the ISCN rules) and **clinical diagnosis** (clinical prognosis for the particular cytogenetic finding).

Karyotype 1 (*3 months old boy with hypospadias and cryptorchidism*)

Cytogenetic diagnosis:
Clinical diagnosis:

Karyotype 2 (*Newborn girl with congenital heart defect and severe muscle hypotonia*)

Cytogenetic diagnosis:
Clinical diagnosis

Karyotype 3 (*15 years old girl with primary amenorrhea*)

Cytogenetic diagnosis:
Clinical diagnosis

Karyotype 4 (*25 year old male examined because of repeat abortions in his partner*)

Cytogenetic diagnosis:
Clinical diagnosis

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Karyotype 5 (*20 weeks old fetus with holoprosencephaly*)

Cytogenetic diagnosis:
Clinical diagnosis

Karyotype 6 (*36 years old male examined with his partner for the reproductive failure)*

Cytogenetic diagnosis:
Clinical diagnosis

Karyotype 7 (*3 years old boy with severe psychomotor retardation*)

Cytogenetic diagnosis:
Clinical diagnosis

Karyotype 8 (*39 years old father of a stillbirth baby*)

Cytogenetic diagnosis:
Clinical diagnosis

Task 2:

On the same webpage – you will continue with the analysis of 3 photographs showing the results of FISH analysis in 5 years old girl with short stature.

1. Analyse given photos of FISH test in which X-centromeric probe was used (green labeled). What is the result of this test?

2. Is the finding relevant to the phenotype of proband?

3. What is the most likely mechanism of origin of this pathology?

4. What does the severity of phenotypic manifestation depend on?

Task 3:

The final task on the same webpage is to analyse the attached results of array-CGH analysis in fetus with orofacial cleft and heart defect.

1. Analyse the results of fetal arrayCGH test. Which CNV is pathogenic?

2. What is the clinical diagnosis of fetus?

3. Does the finding correspond to the reason for referral?

4. Would this finding influence prognosis of next pregnancy of this couple? (parents have normal karyotypes)