Cytogenetic analysis in prenatal diagnosis

INTRODUCTION

INDICATIONS

- Advanced maternal age
  - >35 years old

- Screening test results
  - Probability higher than 1/250

- Family history of genetic disorders

PREVALENCE OF TRISOMY 21 BY MATERNAL AGE AND GESTATIONAL AGE

- Trisomy 21
  - NT<br>  - hCG<br>  - alpha-FP<br>  - Inhibin A

- Trisomy 18
  - NT<br>  - hCG<br>  - alpha-FP<br>  - Inhibin A

- Trisomy 13
  - NT<br>  - hCG<br>  - alpha-FP<br>  - Inhibin A

G-BANDING

- Chromosomes are stained with Giemsa, giving a characteristic pattern for each chromosome.

- Detection rate depends on the resolution (35p, Xqter, 22q11.2, 16p11.2, 11p15.5).

FISH (FLUORESCENCE IN SITU HYBRIDIZATION)

- Hybridization of probe labeled with fluorescence (green within chromosomes either in metaphase or in interphase) followed by microscope slide.

- Advantages: Rapid diagnosis, results available in 4-5 h.

ARRAY CGH (COMPARATIVE GENOMIC HYBRIDIZATION)

- Genetic counseling is performed throughout the prenatal diagnosis process.

GENETIC COUNSELING

PRE-TEST COUNSELING

- Focus of Down syndrome
- Cancers: Breast cancer and leukemia are increased.
- Background risks and individual increased risks
- Risks of testing and risks associated with the procedure

- Stress: Clinicians should be sensitive to their psychological impact.

POST-TEST COUNSELING

- disclosure about the test and its limitations
- discussion about the parents' ability and willingness to have a child affected by a syndrome
- counseling on genetic testing and options

- Counseling must explain all possible options in a non-directive way

- COUNSELORS MUST RESPECT PARENTS' DECISION AND BE PREPARED TO SUPPORT THEM IN DECISION-MAKING PROCESS

CONCLUSIONS

- IPS is not performed on all pregnant women. It is used where the probability of having an affected fetus is >1/500.
- Genetic counseling must always occur before any invasive test.
- Array CGH has higher resolution and wider coverage than QF-PCR and FISH, respectively. Results obtained by array CGH must be confirmed using other methods. FISH can only be used to analyze low syndromes in each exon.
- Applied to select the best test depending on the syndrome of interest.
- Genetic counseling: Explain the risks, the syndrome's implications, and provide psychological support.

REFERENCES