Genetics

(Lecture 3&4)

1- Which one of the following pairs is mismatched ?

a. Patau syndrome: 47,XX,+13

b. Edward syndrome: 47, XX, +18

c. Down Syndrome: 47, XX, +18

Answer : C

2- A female with a flattened face, small head, short neck, protruding tongue, small ears, and a poor muscle tone (hypotonia). She probably has a genetic disorder that's caused by _____

a. Trisomy 21

b. Monosomy X

c. Trisomy X

Answer : A

3- Which of the following genetic changes is associated with a female whose karyotype is 46,XY ?

a. SRY gene mutations (deletions, translocations, …etc).

b. Monosomy X

c. Triple X syndrome

Answer : A

4- Chronic myeloid leukemia is caused by _____

a. t(9;22); a translocation that fuses part of the ABL1 gene from chromosome 9 with part of the BCR gene from chromosome 22, creating a gene called BCRABL1.
b. Having 3 copies of chromosome number 18.

c. Trisomy 13.

Answer : A

5- The most common aneuploidy that infants can survive with is _____ (most compatible with life)

a. Trisomy 18 (Edwards syndrome)

b. Monosomy X (Turner syndrome)

c. Trisomy 21 (Down syndrome)

Answer : C

6-The most stable chromosome of the following is :

a. 46 ,OY

b. 47 , XY – trisomy 14

c. 45, XY, t (14,21)

Answer : C

7- A patient with klinefelter syndrome can be seen as :

a. A male with 47 XXY

b. A female with 47 XXY

c. A female with 45 OX

Answer : A

8-Which of the following human triploid is possible to be found in adults :

a. 92, XXXY

b. Triploid cannot be found in adult human because it is incompatible with life

c. 23, XY

d. 92, YY

e. 69, XXY

Answer : B

9-Trisomy 47,XYY is a syndrome with signs and symptoms that range from being barely noticeable to learning disabilities, speech delay, low muscle tone. How would you expect this syndrome to have occurred?

a. Dispremy

b. Endomitosis

c. Fertilization by two sperms

d. Chromosomal rescue

e. Nondisjunction of paternal gametes

Answer : E

10-Which of the following karyotype is expected to be associated with abnormal phenotype [Note: "t" is translocation, and "del" is deletion]:

a. Deletion of the "P" arm of chromosome 5. Karyotype is 46,XX,del5p

b. Balanced double Robertsonian translocation between both chromosomes 13 and both chromosomes 14. Karyotype is 44,XX,t(13q;14q) *2.

c. Deletion of the "P" arm of chromosome 22. Karyotype is 46,XX,de22p

d. Balanced Robertsonian translocation between one chromosomes 13 and one chromosome 14. Karyotype is 45,XX,t(13q;14q)

e. 46,XY

Answer : A

11-What is the possibility for a couple to have a child with Edwards syndrome if the fathers' homologous chromosomes 18 fail to disjoin during meiosis 1?

a. 25%

b. 0%

c. 50%

d. 100%

e. 75%

Answer : C

12-46 XX male syndrome is a rare condition, described by De la Chapelle et al. in 1964 where the individual has phenotypically male characteristics. It occurs in one out of every 20,000-25,000 newborn males. This condition could be related to:

a. Turner syndrome

b. Translocation of acrocentric chromosomes

c. SRY gene

d. Rb gene

e. Kleinfelter syndrome

Answer : C

13-If one of the parent who carries balanced reciprocal translocation mates with a partner with normal karyotype. What is the risk of having a fetus with abnormal chromosomal complement ?

a. 1/5

b. 1

c. 1/3

d. 1/2

e. 1/4

Answer : D

14-A child person with clinical features that include: cardiovascular, brain with neurological, renal, gastrointestinal, respiratory, and skeletal malformations, craniofacial abnormalities such as prominent occiput, hand and feet anomalies including clenched hand. This patient is most probably affected with:

a. Trisomy 18

b. Trisomy 21

c. Turner Syndrome

d. Partial Trisomy 21

e. Klinefelter Syndrome

Answer : A

15-In the routinely performed karyotype (G-banding). Which of the following would you expect to have more clinical impact and lead to a disease ? (Lecture 2)

a. A duplication of a region with a light band

b. A duplication of a region with a light band

c. No clinical consequences will be observed due to a single band chromosomal aberration

d. A deletion of a region with a light band

e. A deletion of a region with a dark band

Answer : D

16-A person whose karyotype is 45,XX,t(15q;21q). If she mates with a 46, XY normal individual. What is the possibility that they will have a zygote which will develop into Down syndrome? ("t" is translocation):

a. 1/6

b. 1/4

c. 1

d. 1/2

e. 1/3

Answer : A

17-47 XYY occurs due to :

a. chimeric event

b. nondisjunction event from both parents

c. paternal nondisjunction

d. uniparental disomy (UPD)

e. maternal nondisjunction

Answer : C