Exercises of the National Book

- ***Exercise IV p.108***

Exercise IV

Karyotype analysis allows the detection of genetic diseases. The next table shows the results of the analysis of some normal and abnormal karyotypes. Only the affected pair of chromosomes is shown in this table.

a- At what stage of the cell cycle can we observe such chromosome shape?

b- Can we detect an abnormality before the baby's birth?

Documents 1 and ***2*** were obtained from the karyotypes of two children C_1 and C_2 who show the same troubles in development.

c- Bearing in mind that the genetic make-up (all the genes) carried by the chromosomes 14 and 21 together is the same for C_1 and C_2, compare these documents to each other and to ***document 4***. What can you conclude? What is the name of the abnormality the children suffer from?.

Documents 3 and ***4*** were respectively obtained from karyotypes of the mother (healthy carrier) and the father (healthy) of child C_2.

d- Why is the mother a normal carrier?

e- What are the possible gametes of the couple?

f- Estimate the risk for the couple to have a trisomic child.

Documents	chromosome 14	chromosome 21
Document 1 Child C_1		
Document 2 Child C_2		
Document 3 Mother of child C_2		
Document 4 Father of child C_2		

Exercise IV:

a- Metaphase of Mitosis

b- - By prenatal diagnosis

- Collecting cells from the chorionic villi or blood from the umbilical cord, or amniotic fluid.

c- C_1, C_2 and father have a pair of chromosome of 14, where both chromosomes 14 are of the same length in C1, and the father, but in C_2 , one of chromosome 14 is longer then the other (it shows tans location of chromosome 21). However, C_1 has 3 copies of chromosome 21 (free) equal to that of C_2 where the third copy is trans located to chromosome 14, more than the father who has 2 copies of chromosome 21.

Thus, C_1 and C_2 have abnormal karyotypes having extra chromosome 21 while the father has normal karyotype.

→ **Name of abnormality:** C_1 : free trisomy 21

C_2 : linked (translocated) trisomy 21

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d- Doc.3 show that the 2^{nd} copy of chromosome 21 is translocated on chromosome 14. So, she has abnormal karyotype but a normal phenotype because she has no excess or missing in genetic information where chromosomes are the carriers of the genetic information. The genetic information was conserved there was only translocation.

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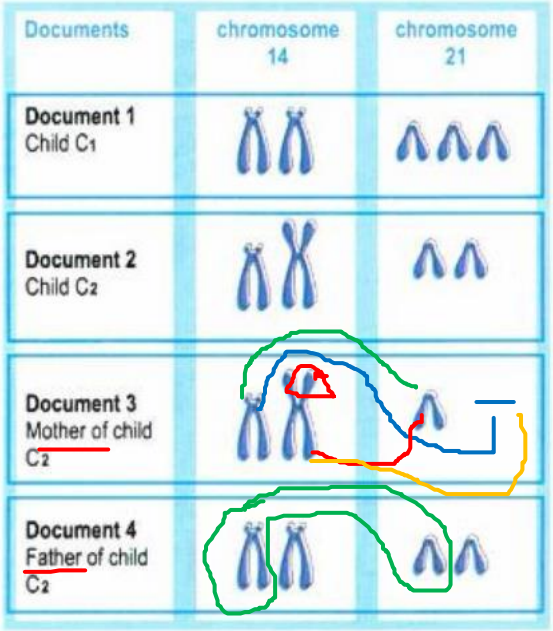
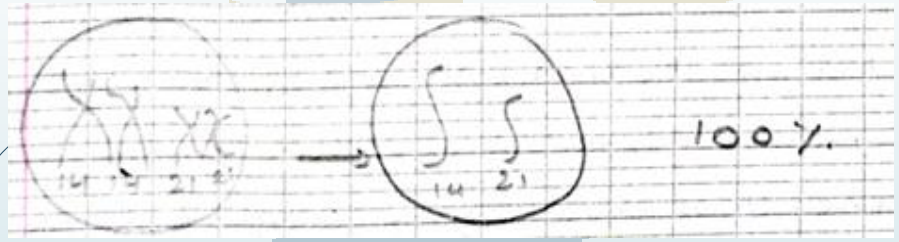


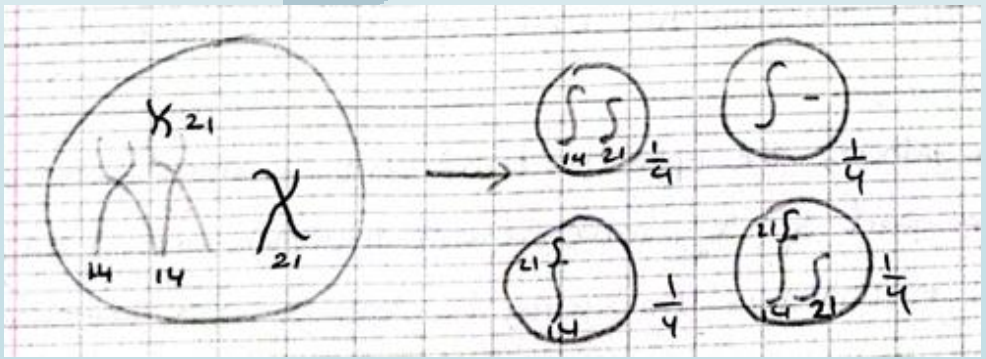
e- During miosis, the 2 homologous of a pair separate, each gamete will have 1 chromosome of each pair.

- The father gives only one type of gametes

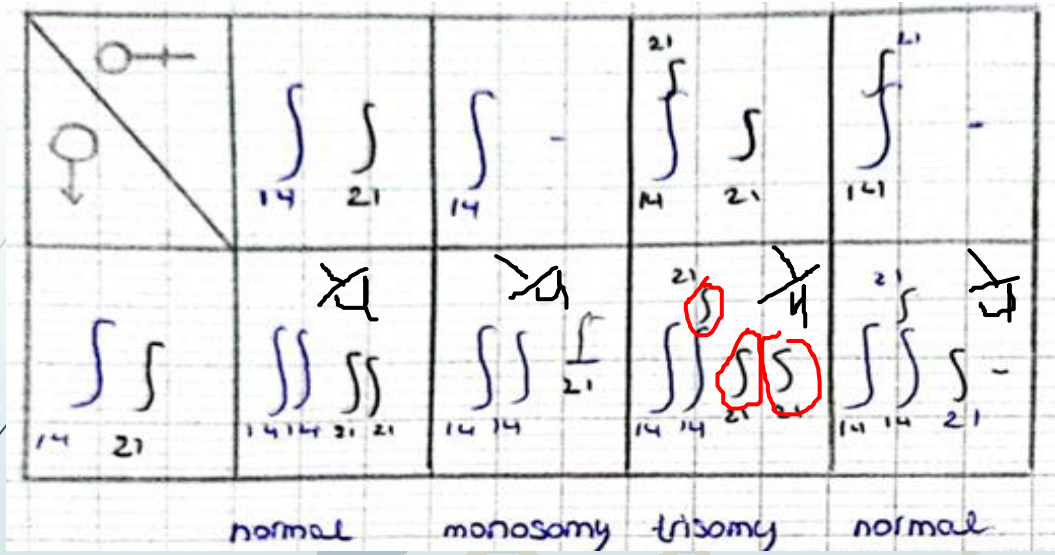
Title: A schematic diagram repr3esenting possible gametes of the father



Title: A schematic diagram representing possible gametes of the mother.



f- Table of cross



Risk of the couple to have trisomic child is $\frac{1}{4}$