

Group 1
1. Tetrad
2. Crossing over
3. Chromosomal abnormality
4. Interchromosomal recombination

Group 2
a. mixing of the alleles due to random separation of homologous chromosomes during the anaphase I.
b. change in the number and /or the structure of chromosomes or in both.
c. structure formed by homologous chromosomes pairing during prophase I.
d. exchange of chromosomal fragments between homologous chromosomes during prophase I.

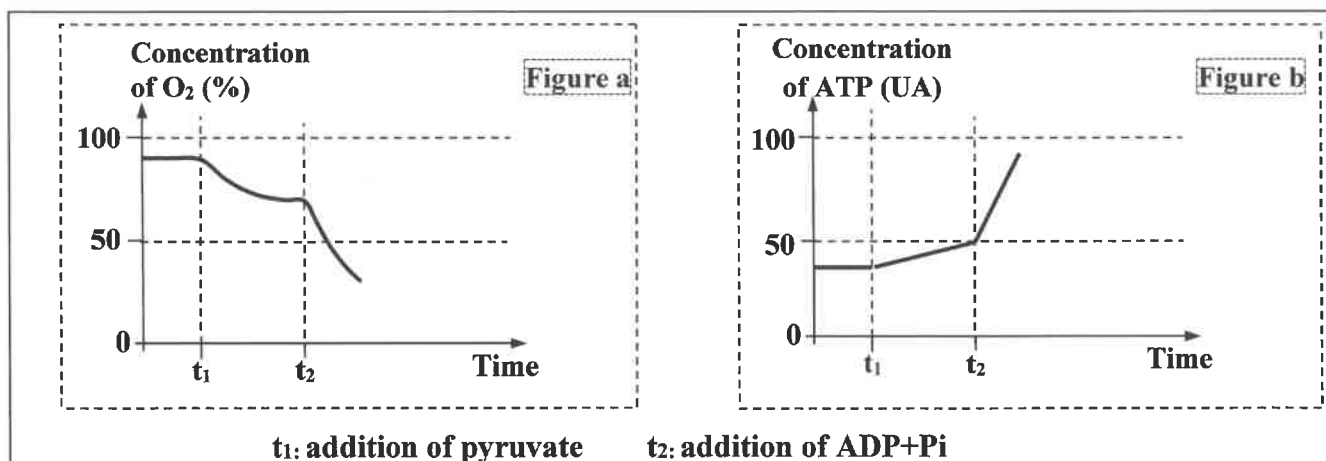
Section II: Scientific reasoning and communication in graphic and written modes (15 pts)

Exercise 1 (5.5 pts)

On the night of Wednesday, August 12, 2015, two terrible explosions shook the industrial zone of the port of the city of Tianjin in China causing more than a hundred dead and more than 700 injured. Many dangerous chemical substances are usually stored in this area including sodium cyanide the source of a very toxic gas, hydrocyanic acid (HCN) leading to death by cell and tissue asphyxiation. In order to understand the effect of hydrocyanic acid on respiratory metabolism and its relationship to asphyxiation, the following data are proposed:

• **Data 1:**

We place a mitochondrial suspension in a suitable rich-oxygen medium, then we follow the evolution of the concentration of O₂ and ATP in the medium. The document 1 shows the experimental conditions and obtained results.



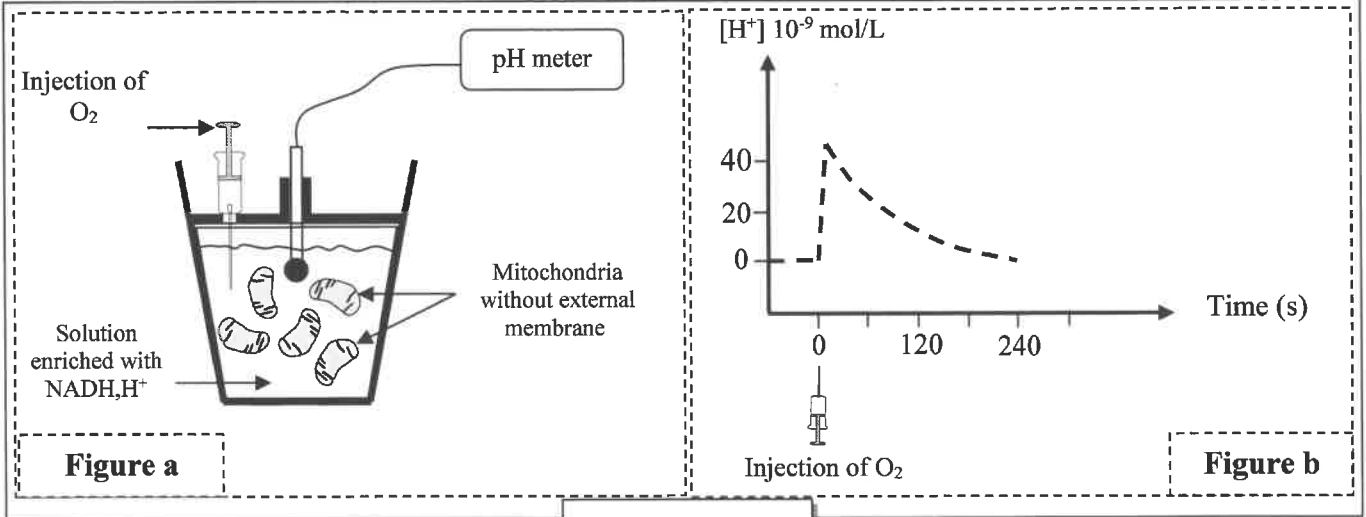
NB: The mitochondria initially contain a small amount of ATP+ Pi.

Document 1

1. Based on the document 1, **describe** the variation of concentrations of O₂ and ATP in the medium, then **deduce** the effect of adding pyruvate and ADP + Pi on mitochondrial respiratory metabolism. (1.5pts)

• **Data 2:**

Mitochondria, deprived of their external membranes, are placed in a solution without oxygen and enriched with electron donors (NADH, H⁺). The variation in the concentration of H⁺ protons in the solution is measured before and after the injection of a limited amount of oxygen. **Figures a and b** of document 2 present respectively the conditions and results of this experiment.



Document 2

2. Based on the data in document 2, **describe** the evolution of the concentration of protons (H^+) in the solution, then **deduce** the effect of oxygen injection on the movement of protons (H^+) through mitochondrial inner membrane. (1pt)

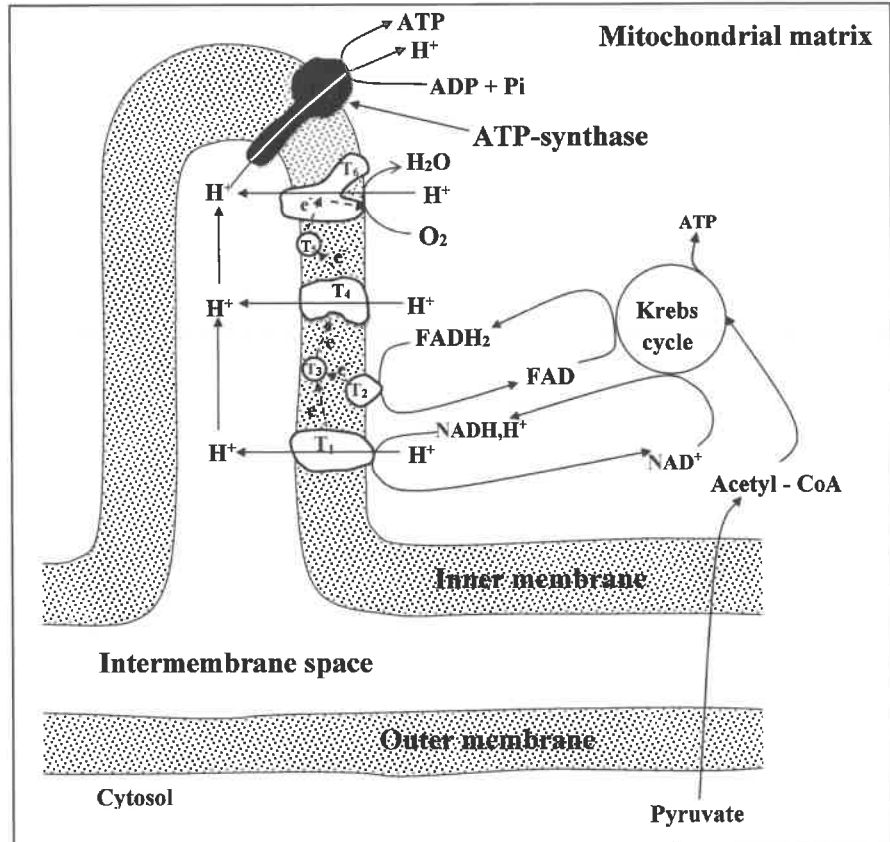
• **Data 3:**

The scheme in document 3 summarizes the reactions of mitochondrial respiratory metabolism and the relationship between pyruvate degradation and ATP synthesis.

3. By exploiting the document 3, **explain** the variation of concentrations of O_2 , protons (H^+) and ATP registered in experiments of documents 1 and 2. (2pts)

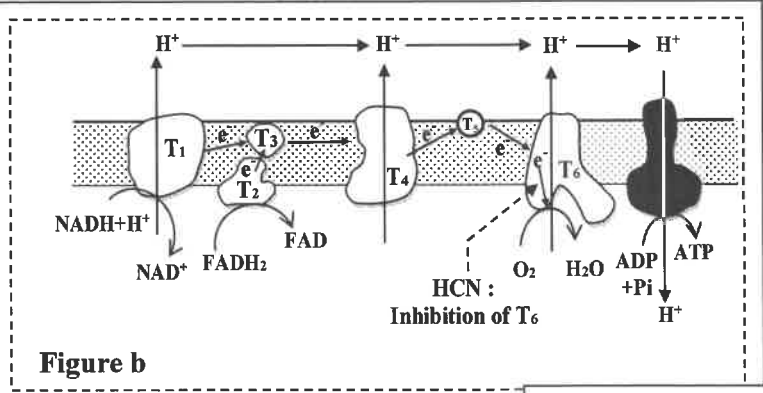
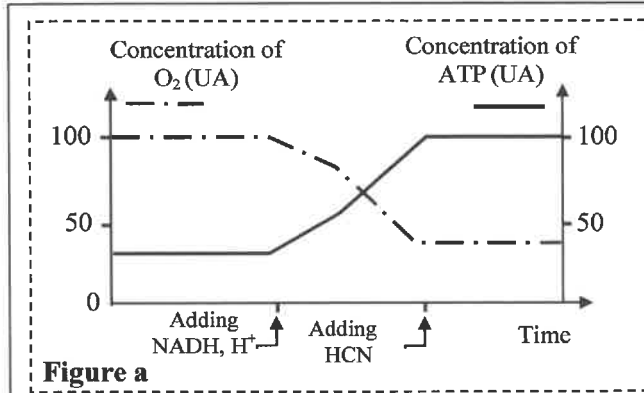
• **Data 4:**

In order to understand the relationship between the exposure to hydrocyanic acid (HCN) and the asphyxiation states recorded following the explosions in the industrial zone in the port of the Tianjin city, the data in Document 4 is proposed.



T₁, T₂, T₃, T₄, T₅, T₆: Transporters of electrons Document 3

The **figure a** in document 4 shows the evolution of concentrations of O_2 and ATP in a mitochondrial suspension placed in a suitable medium rich in O_2 and $ADP + Pi$ following the addition of $NADH, H^+$ and HCN. The **figure b** of the same document represents the mechanism of oxidative phosphorylation at the mitochondrion and the site of action of HCN.



Document 4

4. By exploiting data of document 4, **explain** the asphyxiation related to the exposure to hydrocyanic acid. (1 pt)

Exercise 2 (6.5 pts)

Tay-Sachs disease is a hereditary neurodegenerative disease which one of the forms occurs around the age of 2 to 3 years. Among its main symptoms: loss of motor skills, epileptic crisis, balance disorders, hypersensitivity to noise, mental retardation and sometimes decreased vision. Children affected by this disease usually die around the age of 5 years. In order to understand the genetic origin of this disease, the following data is proposed:

• Data 1:

Research has linked this disease to the lack of **Hexosaminidase A (HEX-A)** enzyme activity in cytoplasmic vesicles called lysosomes. In the normal case, this enzyme ensures the degradation of a substance called **Ganglioside (GM2)**. In the abnormal case the accumulation of **GM2** in the lysosomes is toxic for the nerve cells causing their degeneration. The document 1 represents the future of **Ganglioside GM2** in nerve cells and the appearance of these cells in a healthy individual and in an affected individual.

Molecule level	Cell level (nerve cell)	Individual level
<p>functional HEX-A</p> <p>Ganglioside GM2 → Ganglioside GM3 + GNA Complex</p>	<p>Nucleus</p> <p>Lysosome</p>	Healthy individual
<p>Non-functional HEX-A</p> <p>Ganglioside GM2 → Accumulation of Ganglioside GM2</p>	<p>Nucleus</p> <p>Giant lysosome</p>	Affected individual by Tay-Sachs

Document 1

1. Based on document 1, **show** the protein-trait relationship. (0.75pt)

• **Data 2:**

The synthesis of **HEX-A** enzyme is controlled by a gene **HEX-A** which exists in two allelic forms: The normal allele responsible for functional **HEX-A** enzyme synthesis and the abnormal allele responsible for non-functional **HEX-A** enzyme synthesis. The document 2 present a fragment of untranscribed strand of DNA for each of the two alleles. The document 3 presents the table of the genetic code.

	1270	Reading direction						1290
Fragment of normal allele	↓	CGT	ATA	TCC	TAT	GCC	CCT	GAC
Fragment of abnormal allele		CGT	ATA	TCT	ATC	CTA	TGC	CCC TGA C

Document 2

1 st letter	2 nd letter	U	C	A	G	3 rd letter			
U	UUU	Phe	UCU	Ser	UAU	Tyr	UGU	Cys	U
	UUC		UCC		UAC		UGC		C
	UUA	Leu	UCA		UAA	STOP	UGA	STOP	A
	UUG		UCG		UAG		UGG	Trp	G
C	CUU	Leu	CCU	Pro	CAU	His	CGU	Arg	U
	CUC		CCC		CAC		CGC		C
	CUA		CCA		CAA	CGA	A		
	CUG		CCG		CAG	CGG	G		
A	AUU	Ile	ACU	Thr	AAU	Asn	AGU	Ser	U
	AUC		ACC		AAC		AGC		C
	AUA	ACA	AAA		AGA	A			
	AUG	Met	ACG		AAG	Lys	AGG	Arg	G
G	GUU	Val	GCU	Ala	GAU	Ac.asp	GGU	Gly	U
	GUC		GCC		GAC		GGC		C
	GUA		GCA		GAA	GGA	A		
	GUG		GCG		GAG	GGG	G		

Document 3

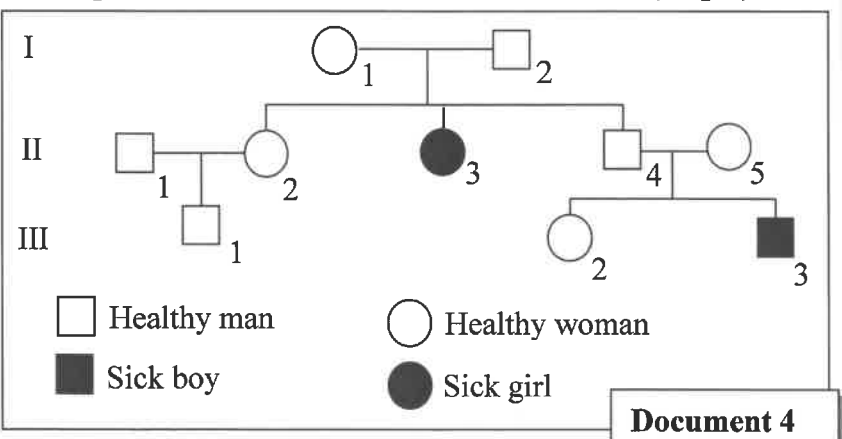
2. Based on documents 2 and 3, **determine** mRNA and the amino acids sequences corresponding to each of the two alleles, then **explain** the genetic origin of the disease. **(1.5pts)**

• **Data 3**

The document 4 presents a pedigree of a family whose members are affected by Tay-Sachs disease.

3. Based on the pedigree of document 4, **determine** the mode of transmission of this disease. **(1pt)**

4. a. Give by **justifying** your answer, genotypes of individuals I₂, II₂ and III₃. **(1pt)**



Document 4

(Use the symbols N and n for the two alleles of the studied gene)

b. The couple II₄ and II₅ wish to have a third child, **determine** the probability that this couple will give birth to a healthy child. **Justify** your answer by Punnet square. **(0.75pt)**

الصفحة	NS 32E	الامتحان الوطني الموحد للبكالوريا - الدورة العادية 2021 - الموضوع
6		- مادة: علوم الحياة والأرض - شعبة العلوم التجريبية مسلك علوم الحياة والأرض (خيار إنجليزية)
6		

• **Data 4**

The Tay-Sachs disease is a rare hereditary disease, however in some North American populations it affects one child in 3600.

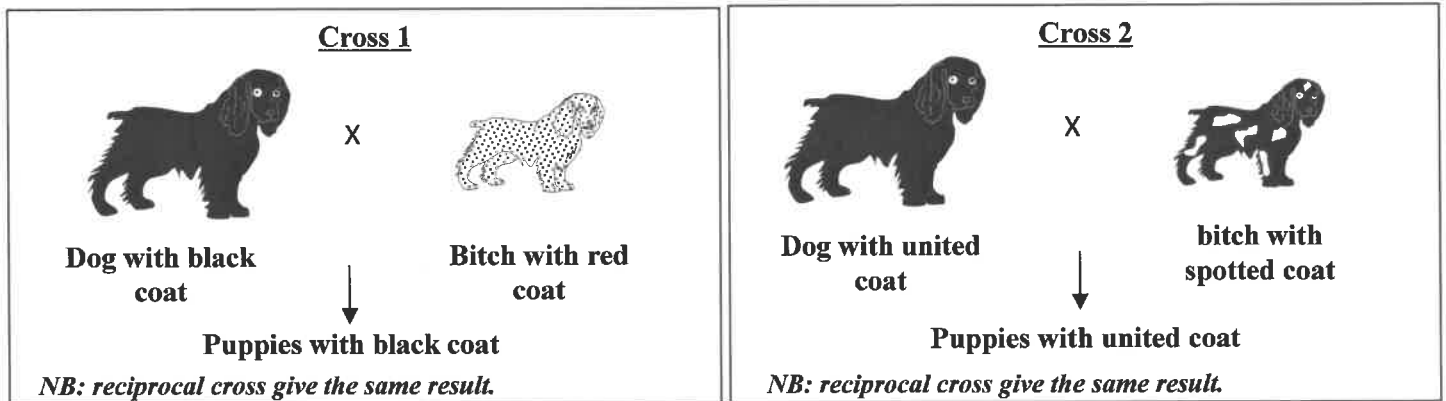
5. Based on the previous data and knowing that these populations are in the Hardy-Weinberg equilibrium:

- a. Calculate the frequency of the two allele N and n in these populations. (1.pt)
b. deduce the frequency of healthy carrier individuals in these populations. (0.5pt)

N.B: Give only four digits after the decimal point in numerical applications.

Exercise 3 (3 pts)

To study the transmission of two hereditary traits in cocker dog: the color (black or red) and type (united or spotted) of coat, we suggest the following crosses:



1. What do you deduce from the results of the two crosses 1 and 2? (1pt)

• **Cross 3:** The cross between dog with united and black coat and bitch with spotted and red coat yielded the following results:

- 25% puppies with black and united coat;
- 25% puppies with red and spotted coat;
- 25% puppies with black and spotted coat;
- 25% puppies with red and united coat.

2. Determine, by justifying the answer if the two studied genes are linked or independents. (0.5pt)

• **Cross 4:** The cross between dog with black and united coat and bitch with red and united coat yielded the following results:

- 3 puppies with black and united coat;
- 3 puppies with red and united coat;
- 1 puppy with black and spotted coat;
- 1 puppy with red and spotted coat.

3. a. Determine the genotype of each of the parents in cross 4. Justify your answer (0.5pt)
b. Use Punnett square to Interpret the results obtained in cross 4. (1pt)

*Use the following symbols: -R and r for alleles responsible for the color of coat;
-B and b for alleles responsible for the type of coat.*

-End-