Closed book

- Time closed book part: **30 minutes**
- Total time assessment: **60 minutes**
- When you are finished with the closed book part, you need to submit this by using the button 'Submit' (at the bottom of your screen). You will get a warning if you did not give an answer for all the questions. Click 'Cancel' to return to the questions to fill in the missing answers.
- After submitting the closed book part, you cannot return to the questions and the given answers of the closed book part. This is the same for the open book part.
- After submitting the closed book part you will automatically continue with the open book part.
- You are allowed to use your books after the signal of the invigilators.
- You are not allowed to take the assessment, or parts of it, with you. Doing so will be judged as fraud.

1. Which of the following is a continuous DNA molecule with associated proteins that contains the genetic information in a cell?

- (a) genome
- (b) centriole
- (c) chromosome
- (d) centromere

If choice c is selected set score to 1.

2. *Mutations in two different genes may cause red-green colour blindness. Unbalanced translocations of these genes are the cause of this type of colour blindness.*
Which diagnostic technique is used to demonstrate the presence of unbalanced translocations?

- (a) array CGH
- (b) GWAS
- (c) linkage tests
- (d) protein electrophoresis

If choice a is selected set score to 1.

3. Which of the following statements about unbalanced chromosomal aberrations is correct?

- (a) chromosomal material is gained
- (b) chromosomal material is lost
- (c) chromosomal material is either gained or lost
- (d) chromosomal material is neither gained nor lost

If choice c is selected set score to 1.

4. *The court appoints a father as guardian of his adult daughter who is being treated in a psychiatric hospital. After the daughter committed suicide, her father requests that he be given access to her medical file. The hospital refuses the request, stating that (1) his guardianship became void with his daughter's suicide, so that he can no longer derive any rights from it; and (2) that simply submitting the file of the deceased daughter would constitute a breach of medical confidentiality.*
The hospital's argument is legally sound.

- (a) True
- (b) False

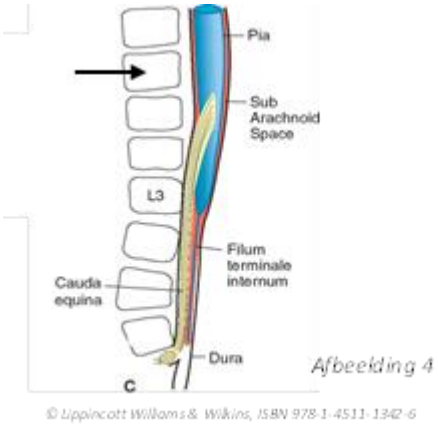
If choice b is selected set score to 1.

5. *A certain X-linked chromosomal abnormality is consistent with the following clinical picture: short broad neck, below-average body height, oedema of dorsum of hands and feet*
Which of the following X-linked chromosome abnormalities is most likely the case?

- (a) 45,X (Turner syndrome)
- (b) XXY (Klinefelter's syndrome)
- (c) XXX (trisomy X)
- (d) 47,XYY syndrome

If choice a is selected set score to 1.

6. What structure is indicated by the arrow in the figure?



Afbeelding 4

- (a) C8 vertebra
- (b) L1 vertebra
- (c) S5 vertebra
- (d) T12 vertebra

If choice d is selected set score to 1.

7.

- (a) caudal
- (b) cranial
- (c) dorsal
- (d) transverse

If choice b is selected set score to 1.

8. Which germ layer is the origin of the vertebrae?

- (a) ectoderm
- (b) endoderm
- (c) mesoderm

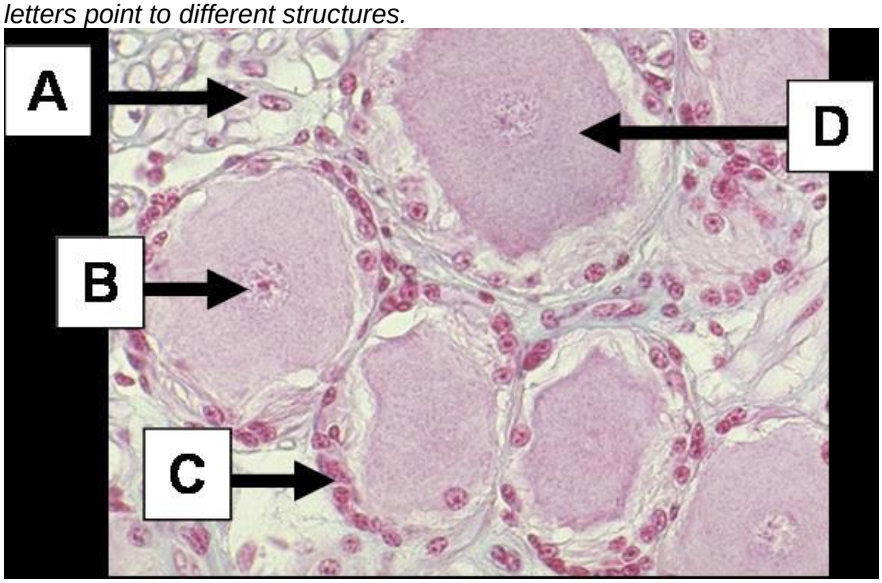
If choice c is selected set score to 1.

9. *There are four primary types of tissue.*
Which of the following is not a primary tissue type?

- (a) epithelium
- (b) connective tissue
- (c) muscle tissue
- (d) adipose tissue

If choice d is selected set score to 1.

10. *This picture iss showing a ganglion (400 x magnification) . Fours arrows indicated with different letters point to different structures.*



Which arrow point to the nucleus of a mantle cell?

- (a) Arrow A
- (b) Arrow B
- (c) Arrow C
- (d) Arrow D

If choice c is selected set score to 1.

11. *Various cells must be active one after the other before a muscle can contract. A shared characteristic of these cells is that they all have a resting membrane potential.*
Which type of ion channel creates the resting potential?

- (a) leak channel
- (b) mechanically activated channel
- (c) voltage-dependent channel
- (d) transmitter-activated channel

If choice a is selected set score to 1.

12. *A neonate has skin erosions and blisters. A skin biopsy is taken to determine the blister level. Immunofluorescence staining reveals keratin in both the blister roof and the blister base.*
What is the cleavage level?

- (a) intraepidermal above the basal membrane
- (b) subepidermal in the basal membrane
- (c) subepidermal below the basal membrane

If choice a is selected set score to 1.

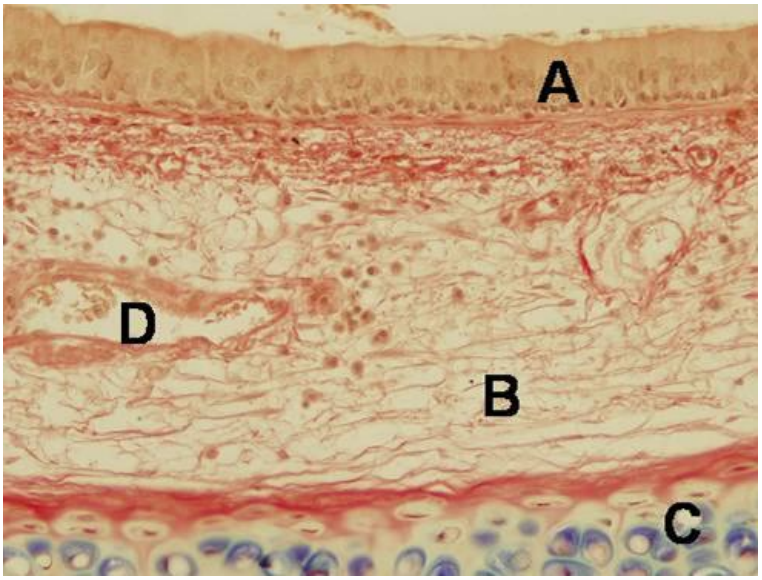
13. *This is a micrograph (magnification 1000x) of a Fallopian tube. Fallopian tubes transport the egg cells from the ovary to the uterus.*

What type of epithelial membrane specialization is indicated by the arrow?

- (a) cilia
- (b) brush border
- (c) microvilli

If choice a is selected set score to 1.

14. Below is a micrograph (magnification 200x) of a histological section.



Which letter indicates loose connective tissue?

- (a) A

- (b) B
- (c) C
- (d) D

If choice b is selected set score to 1.

15. *A 16-year-old boy with a history of hepatitis A has been nauseous for a week. His sclerae have become yellow too. The physical examination reveals pain in the right upper abdomen. Blood tests show elevated levels of the hepatic enzymes AST and ALT.*
Which hepatocyte impairment explains the elevated hepatic enzyme levels?

- (a) autolysis by lysosomes
- (b) clumping of nuclear chromatin
- (c) cell membrane defects
- (d) swelling of mitochondria

If choice c is selected set score to 1.

16. *The DNA of a patient who is suspected of having epidermolysis bullosa dystrophica is tested with the next-generation sequencing panel for EB diagnostics. The report of the DNA laboratory includes the following: 'homozygous mutation c.425A>G, p.Lys142Arg found in the* COL7A1 *gene'.*
At nucleotide (base pair) level, this is an instance of:

- (a) base pair substitution (point mutation)
- (b) base pair deletion
- (c) duplication
- (d) insertion

If choice a is selected set score to 1.

17. *Ms Pieterse is 28 years of age. She is 20 weeks pregnant with her first child. An ultrasound scan reveals a large lumbosacral cèle. She is otherwise in good health. Six months before she became pregnant, she began taking an additional 0.5 mg of folic acid per day; she stopped doing so several weeks ago.*
Besed on the level of the cèle it can reasonably be expected that there will be a complete paralysis of both legs.

- (a) True
- (b) False

If choice a is selected set score to 1.

18. How many autosomes does a human cell contain?

- (a) 1
- (b) 22
- (c) 44
- (d) 46

If choice c is selected set score to 1.

19. *A pharmacist is preparing the drugs for a client in sight of the other costumers*
This pharmacist is violating his legal obligation in respecting the physical privacy of this client

- (a) True
- (b) False

If choice b is selected set score to 1.

20.
A rapid increase phase is typical for an action potential
What is causing the increase?

- (a) Opening of potassium channels
- (b) Opening of sodium channels
- (c) Closure of potassium channels
- (d) Closure of sodium channels

If choice b is selected set score to 1.

Open book

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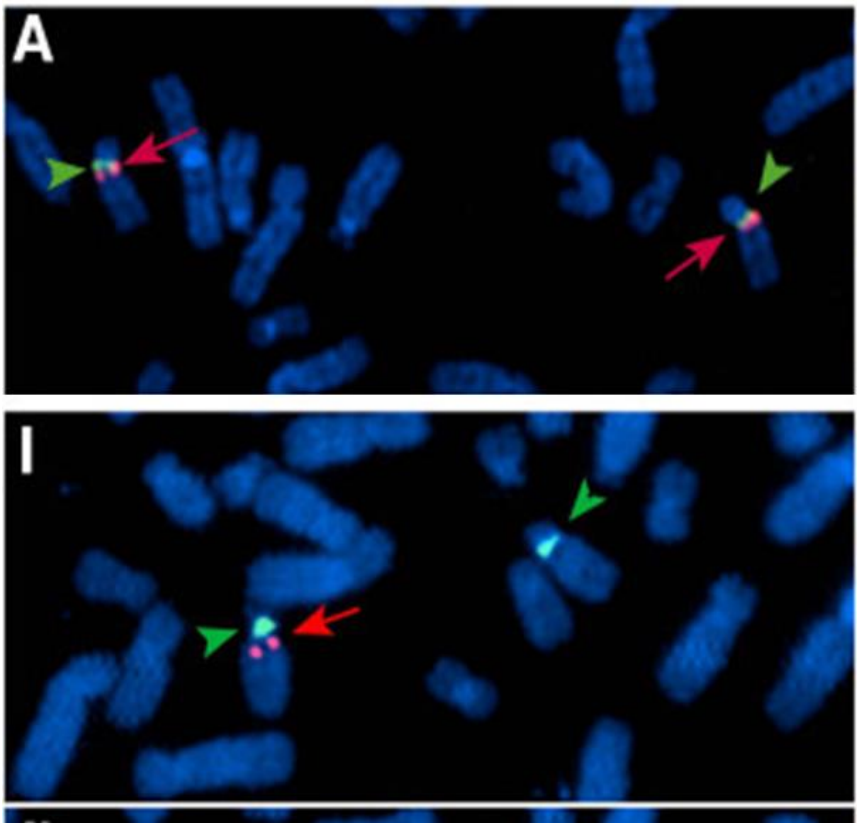
- After submitting the open book part, you cannot return to the questions and the given answers of the open book part.
- After submitting the open book part, you will get a first provisional score of your complete assessment. No right can be derived from this score. The final score will be published on Nestor.
- You are allowed to use your books after the signal of the invigilators.
- You are not allowed to take the assessment, or parts of it, with you. Doing so will be judged as fraud.
- Before you leave the examination hall you have to report yourself at the desk at the front side of the hall, so we can check if we received your answers on the assessment in good order.

1. *A new-born boy has trouble drinking due to a poor sucking reflex. He hardly responds to stimuli, does not cry a lot, sleeps a lot and has several typical external features such as a narrow forehead and almond-shaped eyes. It is suspected that he has Prader-Willi syndrome.*
FISH is used to make the diagnosis. Two probes are used: one is specific to chromosome 15 and labelled green and the other (red) is specific to area 15q11-13. Panel A shows the result of a control, while panel I is that of the neonate.



The FISH result confirms a 15q11-13 deletion, which could be consistent with Prader-Willi syndrome.

- (a) correct
- (b) incorrect

If choice a is selected set score to 1.

2. The result of the FISH test confirms the diagnosis of the Prader-Willi syndrome

- (a) correct
- (b) incorrect

If choice a is selected set score to 1.

3. The FISH result will be the same if the boy has Angelman syndrome.

- (a) correct
- (b) incorrect

If choice a is selected set score to 1.

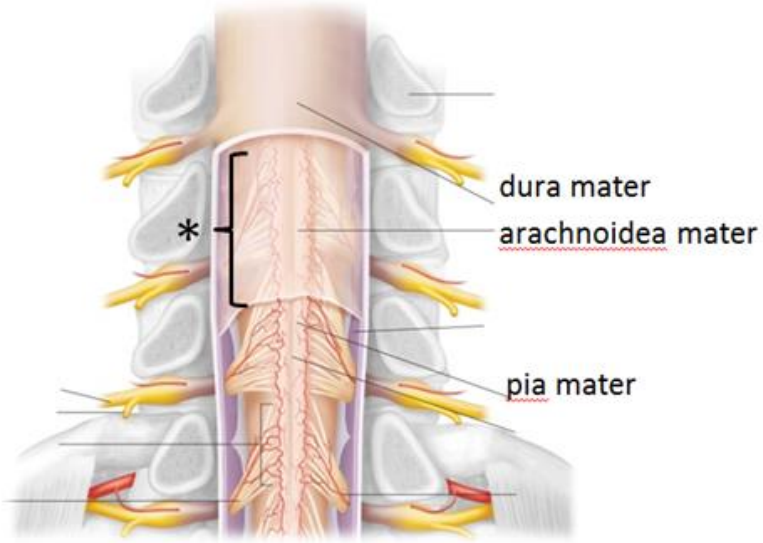
4. *The picture below shows an embryo*

Which of the following statements at this embryonic stage is correct?

- (a) cephalocaudal folding has started
- (b) the caudal neuropore is closed
- (c) the neural folds are starting to fuse
- (d) the embryo's length is about 5 mm

If choice a is selected set score to 1.

5.



Which space is indicated by the asterisk (*)?

- (a) epidermal space
- (b) epidural space
- (c) subdural space
- (d) subarachnoid space

If choice c is selected set score to 1.

6. *In the disease Myastenia gravis auto-antibodies are causing weakening of the muscles.*
These autoantibodies:

- (a) Bind to the myofibrils in the sarcomers
- (b) Block binding of acetylcholine to the receptor in the sarcolemma
- (c) Inhibit the synthesis of acetylcholin
- (d) Stimulate the release of Ca2+ in the sarcoplasma

If choice b is selected set score to 1.

7. *A man with haemophilia A marries a homozygous woman with a normal chromosome pattern.*
What is the probability that this couple will have a son with hemophilae?

- (a) 100%
- (b) 75%
- (c) 50%
- (d) 0%

If choice d is selected set score to 1.

8. Which part of the adult spinal cord is a remain of the lumen of the neural tube?

- (a) Canalis centralis

- (b) Canalis vertebralis
- (c) Dorsal horn
- (d) Ventral horn

If choice a is selected set score to 1.

9. *Keratitis-ichthyosis-deafness (KID) syndrome is a very rare genetic disease that is associated with deafness. KID is caused by a mutation in the GJB2 geen that encodes for the protein connexion 26*
Which intercellulair connections will not function correctly in patients with KID?

- (a) desmosomes (macula adhaerens)
- (b) gap junctions
- (c) tight junctions (zonula occludens)
- (d) zonula adhaerens

If choice b is selected set score to 1.

10. In a patient with a dominant inherited epidermolysis bullos simplex “with mottled pigmentation” the mutation p.PRO25Leu is found in the KRT5 gene. This mutation is causing the disease by a so-called dominant-negative effect.
What is meant by this dominant-negative effect?

- (a) The mutation alone is not enough to cause the disease, another mutation is necessary
- (b) The mutation has a strong negative effect on the function of the mutated gene product
- (c) The mutated gene product disturbs the function of the wildtype gene product
- (d) The fertility of the carrier is decreasing because of the mutation

If choice c is selected set score to 1.