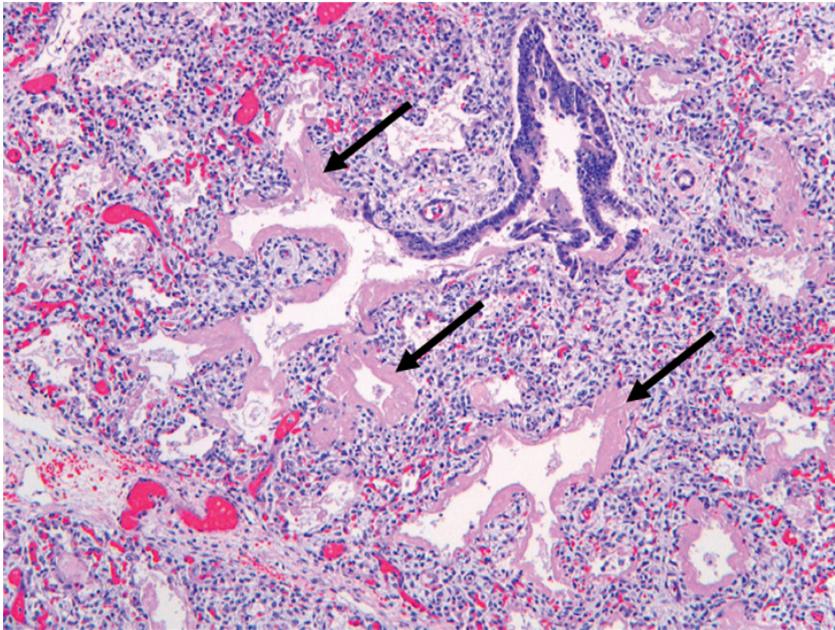


Examples of tasks of the All-Russian interdisciplinary Olympiad "Clinical Debut" with international participation

1. Name the morphological elements indicated by the arrows.  
Name the form of the pathology.



*Answers:*

*Hyaline membranes (in the lumen of the alveoli).*

*Respiratory distress syndrome (newborn).*

2. One of the manifestations of Lesch-Nyhan syndrome is the occurrence of self-harming behaviour, but it is not always present in the clinical picture. This condition is known to be associated with an enzyme defect.
  - a. Give the name of this enzyme
  - b. Explain what determines the presence or absence of self-harming behaviour

*Answers:*

*a. Enzyme name: hypoxanthine-guanine phosphoribosyltransferase.*

*b. The presence or absence of self-damaging behaviour depends on the degree of residual enzyme activity. If it is small (<1.5%) - self-damaging behaviour occurs*

3. Patient M., 12 years old, was admitted to hospital for repeated fainting episodes. EchoCG diagnosed cardiomyopathy - noncompact myocardium of the left ventricle. Proteinuria was noted. Further genetic test confirmed the presence of Val122Ile mutation (replacement of valine amino acid with isoleucine) in transthyretin molecules, which resulted in impaired folding of its molecules. As an alternative to therapeutic treatment, the patient's family was offered a liver transplant.
  - a. What is the name of the disease that causes left ventricular non-compaction (LVNC)? What function does the protein transthyretin fulfil?

- b. Describe the likely pathogenesis of cardiomyopathy, name clinical manifestations associated with this type of cardiomyopathy.
- c. What organ other than the heart is damaged in the patient? Describe its macro and micro pathology.

Answers:

- a. *ATTR amyloidosis (transthyretin amyloidosis).*  
*Transthyretin provides transport of thyroxine and retinol.*
- b. *Genetic abnormality leads to impaired TTR folding and amyloid deposition in the myocardium. As a result, the structure of muscle tissue, hence myocardial conduction and contractility, is disturbed. Subsequently, the inner layers of the left ventricular myocardium undergo fibroelastosis and become noncompact. The compact myocardium hypertrophies.*  
*Myocardial hypertrophy, decreased ejection fraction, periodic pressure drop due to impaired left ventricular contractility leads to syncope.*
- c. *Large white kidney (stage of proteinuria).*  
*Kidneys are enlarged in size, dense, their surface is pale grey or yellow-grey. On the section the cortex is wide, waxy, the medulla is grey-pink, greasy, cyanotic.*  
*Amyloid is in the mesangium and capillary loops of the tubules, under the endothelium of extraglomerular vessels, along the course of the basal membrane of tubules and reticular fibres of the stroma, stained dirty red colour with congo-red.*

4.

- a. IN DE TONI-DEBRE-FANCONI SYNDROME ARE DAMAGED:
  - proximal tubules
  - distal tubules
  - loop of Henle
  - collecting ducts
- b. WHICH OF THE FOLLOWING IS A PATHOLOGICAL MENINGEAL SYMPTOM:
  - Trousseau's symptom
  - Lust symptom
  - Lessage's symptom
  - Babinski's symptom
- c. WHAT TYPE OF BLEEDING DEVELOPS IN THROMBOCYTOPENIA:
  - angiomatous
  - haematoma
  - petechial spotting
  - vasculitic purpura

- d. GESTATIONAL DIABETES INCREASES PRODUCTION OF:
- chorionic somatomammotropin
  - placental growth hormone
  - chorionic gonadotropin
  - prolactin
- e. ST. VITUS' DANCE IS ONE OF THE CRITERIA FOR THIS DISEASE. NAME THE DISEASE.
- acute rheumatic fever
  - rheumatoid arthritis
  - congenital syphilis
  - attention deficit hyperactivity disorder

*Answers:*

- a. proximal tubules*
- b. Lessage's symptom*
- c. placental growth hormone*
- d. petechial spotting*
- e. acute rheumatic fever*