

Examples of tasks for the Olympiad “Virtuoso of Pediatrics” for 5-6 courses of study

Competition “What? Where? When?”

There is a rare mutation that usually do not cause any particular inconvenience to its carrier, although in some situations they make it difficult for them to travel. For some lawbreakers, however, having it (mutation) would only be an advantage. This mutation leads to the absence of...

Answer: fingerprints

Competition of polymaths «All or nothing»

1. What unites all this pictures?



Answer: Nephritis (kidney disease and the name of the stone)

2. Match **the gene** and **the disease** associated with the mutation in these genes

Disease	Number	Gene
A. Proximal spinal muscular atrophy 5q	2	1. NPC1
B. Medium-chain acyl-coenzyme A dehydrogenase (MCAD)	4	2. SMN1
C. Cystic fibrosis	10	3. PAH
D. Gaucher disease	5	4. ACADM
E. Niemann-Pick disease Type C	1	5. GBA
F. Cystinosis	8	6. COL1A
G. Lysosomal acid lipase deficiency	9	7. CBS
H. Classic form of phenylketonuria	3	8. CTNS
I. Classic form of homocystinuria	7	9. LIPA
J. Osteogenesis imperfecta	6	10. CFTR

3. Name all the components of the classic Hutchinson’s triad. What disease describes this triad?

Answer: interstitial keratitis, malformed teeth (Hutchinson incisors), and eighth nerve deafness.

Congenital syphilis

4. Today, the American College of Rheumatology proposes to call **THIS DISEASE** differently, due to the fact that the German pathologist who described it was a member of the the National Socialist German Workers' Party. Name the past and current name of this disease.

Answer: Wegener's granulomatosis, granulomatosis with polyangiitis

Competition of clinicians «Five Questions»

This case is about a famous classical music composer who lived from 1810 to 1849 in Europe. Many historians, studying his biography, tried to understand: why such a talented person died at such a young age? People began to become interested in the health of the great musician only after his death, and still, it is a subject of a debate.

From birth, he was fragile and often suffered from colds. In his youth he had frequent lung problems, diarrhea and weight loss. At the age of 16 years, he had symptoms such as cough, headache, and enlarged cervical lymph nodes for 6 months. Similar symptoms were observed when he was 20 years old. He could never grow whiskers, mustache, beard - his hair was thin and brittle. He had intimate relationships with several women, but never had children.

During the flu epidemic in Paris, the musician had a high fever and hemoptysis. His doctor recommended a warm climate for him. Throughout his life, he was constantly bothered by shortness of breath and increased fatigue. He often had to stay in bed for a long time after playing the piano for a long time.

At the age of 30, his health had noticeably deteriorated. He lost even more weight and began to weigh 45 kg with a height of 170 cm. According to the descriptions of others, he was pale, thin and looked bad. The caricature drawn by his friend clearly shows that he has developed a barrel chest shape. And some biographers wrote that the maestro brilliantly fingered the piano keys with his drum fingers.

Condition of brilliant musician and great composer in the last years of his life had noticeably worsened. Cough, shortness of breath, and intermittent fever were accompanied by frequent hemoptysis, bloody vomiting and swelling of the legs. Several doctors consulted him and gave him different opinions and recommendations, but his condition was regarded as serious and it was difficult to provide adequate medical care.

The musician's father also suffered from respiratory tract infections, but died in 73 years old. Mom enjoyed good health and died at the age of 77. The musician had three sisters: the eldest died at the age of 77, the middle one also suffered greatly from respiratory tract infections and, according to available data, died of a lung disease at the age of 47. The younger sister's health was poor. She periodically

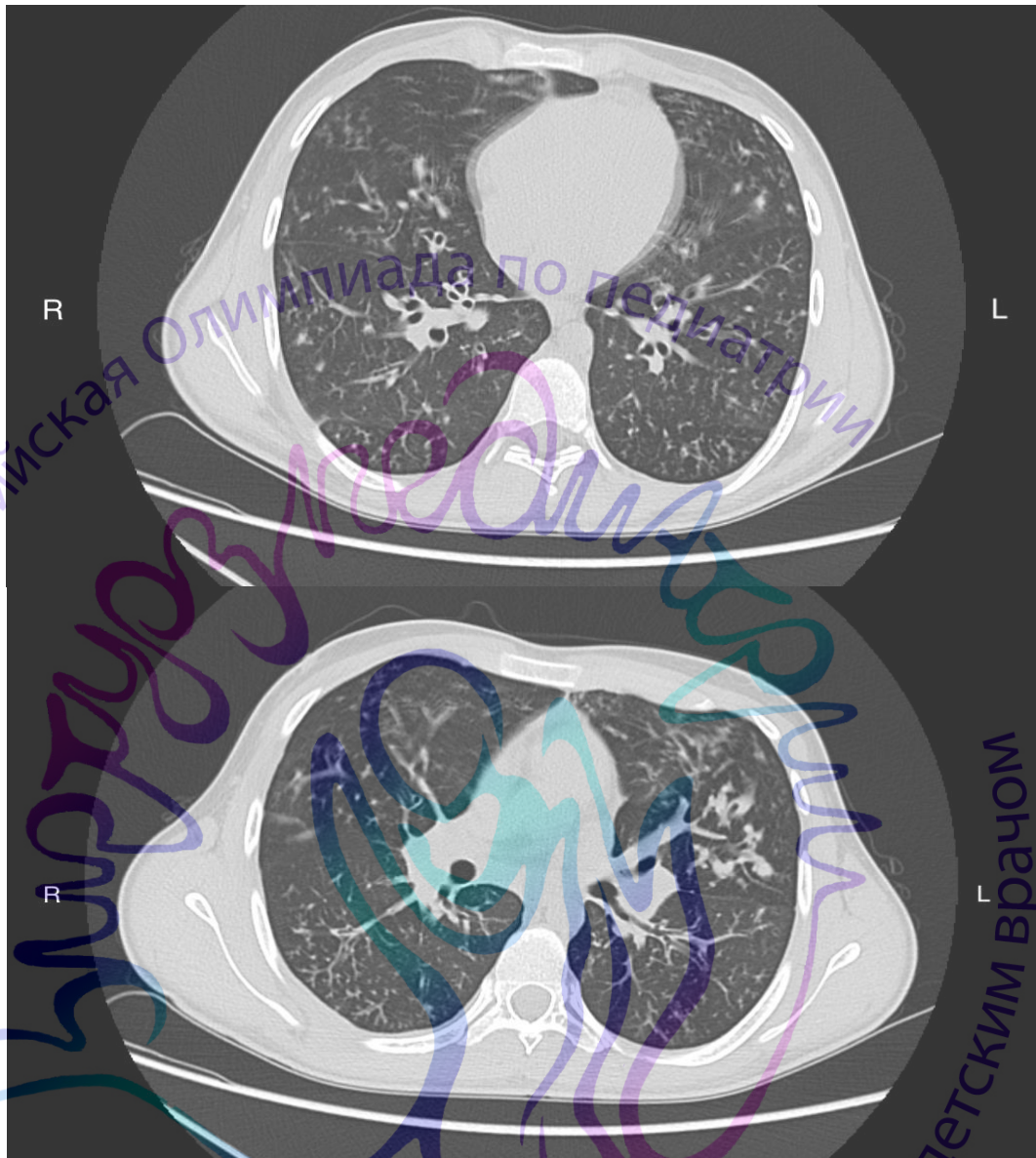
suffered from respiratory tract infections, and also experienced shortness of breath, hematemesis, and weight loss. She died of massive bleeding from the upper gastrointestinal tract when she was only 14 years old.

It is now known that some of this man's descendants also showed similar symptoms of the disease.

1. Do you recognise the person's name?
2. What symptoms could be signs of illness in this man?
3. What disease can be discussed in this case?
4. What diseases need to be differentiated from this one in past years and today?
5. What is the cause of death of the younger sister with these symptoms?
6. Explain the etiology and pathogenesis of this disease.
7. What treatment could the musician currently get and what would be the present prognosis for him?
8. Assess the ABS analysis of a patient with this disease. What is the name of this complication? What is its pathogenesis and principles of treatment?

	Result	Units	Reference values
t	37	°C	
Oxygen of the supplied mixture	21	%	
pH	7.627		7.35- 7.45
Partical pressure of carbon dioxide (PCO2)	64.1	mmHg	35-48
Partical pressure of oxygen (PO2)	44,5	mmHg	83 - 108
Hb	125	g/l	140-180
Ht	38,6	%	
Oxygen saturation	80,7	%	95-99
Oxyhemoglobin	79,2	%	
Carboxyhemoglobin	1	%	0-0.8
Deoxyhemoglobin	19	%	
Methemoglobin	0.8	%	0.2-0.6
K ⁺	2	mmol/l	3,5-5
Na ⁺	118	mmol/l	136-146
Ca ²⁺	0.95	mmol/l	1.15-1.29
Cl ⁻	50	mmol/l	98-106
Glucose	10.6	mmol/l	3.9-5.8
Lactate	2,5	mmol/l	0.5-1.6
Bilirubin	5	µmol/l	
osmolality	247,5	mmol/kg	
cO2	14	mmol/l	
Partical pressure O2 50% SO2	26,41	mmHg	
BE	39.7	mmol/l	
HCO3	65,6	mmol/l	

9. What pathological changes are present on the CT scan of the chest of the patient with this disease? Name them.



Answers

1. Frederic Chopin
2. Frequent colds, nutritional disorders (diarrhoea, weight loss), signs of respiratory failure (dyspnea, drum-finger symptom, barrel-shaped chest) and liver failure (legs oedema, bloody vomiting - esophageal varices).
3. Cystic fibrosis, pulmonary- gastrointestinal form.
4. Pulmonary tuberculosis, Chronic obstructive pulmonary disease (COPD), Bronchial asthma, Inflammatory bowel disease, Cirrhosis.
5. We can think of cystic fibrosis with a complication in the form of cirrhosis with liver failure and bleeding from esophageal varices.
6. Mutation in the cystic fibrosis gene (CFTR) leads to dysregulation of ion channels and membrane transport of exocrine glands. The balance of liquid

secretion and absorption of Na^+ and Cl^- ions is disturbed, as a result of which, the volume of liquid decreases and viscosity of liquid secretion of all exocrine glands increases, which leads to impairment of their functions.

7. Physical activities (special breathing exercises and drainage massage), mucolytic, antimicrobial, anti-inflammatory therapy, enzyme replacement therapy, hepatoprotective therapy, high-calorie diet and vitamin therapy. Today the prognosis is more optimistic, genetically engineered treatment have emerged.
8. Pseudo-Bartter syndrome. Metabolic alkalosis (hyponatremia, hypokalemia, hypochloremia). Pathogenesis: patients with cystic fibrosis have increased salt loss with sweat, leading to sodium and chlorine deficiency. Hyponatremia leads to RAAS activation, hyperaldosteronemia and sodium reabsorption in distal renal tubules in exchange for increased secretion of hydrogen and potassium ions. The result is metabolic alkalosis, potassium, sodium, and chlorine deficiency. Treatment: oral or infusion therapy with potassium chloride, sodium chloride drugs, potassium-sparing diuretics.
9. Bilateral cylindrical bronchiectasis, infiltration of peribronchial tissue, intensification and deformation of the pulmonary pattern on both sides.

