METHODOICAL INSTRUCTIONS
FOR THE INDEPENDENT WORK OF STUDENTS
FOR PREPARATION TO PRACTICAL CLASSES
AND DURING PRACTICAL CLASSES

<table>
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<tr>
<th>Academic subject</th>
<th>Neurology</th>
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<tr>
<td>The module № 2</td>
<td>Special neurology</td>
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<tr>
<td>Topic</td>
<td>Hereditary degenerative diseases of the neuromuscular system.</td>
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<tr>
<td>Year of study</td>
<td>IV</td>
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<tr>
<td>Faculty</td>
<td>Foreign Students Training (Medicine)</td>
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Poltava 20__
1. Substantiation of the topic.
When studying this topic, students must consider the following issues: clinical features, inheritance, modern treatment of hereditary muscular and neuromuscular diseases. Hereditary disorders of the nervous system do not belong to a number of the most common. However, most of them have their onset in childhood and young age; they limit the work capacity and lead to disability – this determines practical importance of this pathology for physicians of all specialties, especially for neurologists. Modern theories of pathogenesis and treatment are expanding general medical knowledge of physicians – neurologists, psychiatrists, pediatricians, neurosurgeons, thoracic surgeons, orthopedists.

2. The specific objectives:

<table>
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<tr>
<th>The specific objectives</th>
<th>Initial level of knowledge</th>
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<tr>
<td><strong>To know:</strong></td>
<td><strong>To be able to:</strong></td>
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<tr>
<td>Select data from medical history, which indicate the family, the hereditary nature of</td>
<td>1. Collect family history</td>
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<td>the disease.</td>
<td>Chart the genealogy</td>
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<tr>
<td>2. Identify the most informative features of the nervous system in the objective</td>
<td>2. Conduct a survey of the neurological status (topical diagnosis of diseases of the</td>
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<td>examination of the patient (neurological) and in electrophysiology laboratory studies</td>
<td>nervous system), to evaluate the results of laboratory tests, electrophysiological and</td>
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<td>(biochemical blood tests, electromyography, radiography, reovasography, CT, MRI).</td>
<td>radiological</td>
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<td>3. Make topical diagnosis.</td>
<td>3. Recognize the symptoms of nervous system (topical diagnosis of diseases of the nervous</td>
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<td>4. Use educational and reference books for the differential diagnosis of hereditary</td>
<td>system)</td>
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<td>diseases of the nervous system with tumors, neuroinfections, polyneuropathies.</td>
<td>4. Integrate morphological substrate of pathologies of the nervous system</td>
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<td>5. Formulate the clinical diagnosis (pathological, topical, syndromological, functional).</td>
<td>5. Correctly make the clinical diagnosis</td>
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<td>6. Use educational and reference books for the selection and justification of therapy.</td>
<td>6. Use means of etiological, pathogenetic and symptomatic therapy, medication, physical</td>
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<td>7. Apply deontological skills in treatment of patients.</td>
<td>therapy</td>
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### 3. Basic level of training

| 1. Anatomy | Know the basic groups of skeletal muscles, describe the structure of the segmental apparatus of the spinal cord |
| 2. Histology | Know the histological characteristics of muscle tissue |
| 3. Pathophysiology | Determine the mechanism of degenerative process |
| 4. Physiology | Describe the functioning of the neuromuscular synapse |
| | Give the classification of the mediators and describe the mechanism of their action |
| | Evaluate the data of electromyography |
| 5. Pharmacology | Describe the pharmacological properties of drugs: biostimulators, anticholinesterase, anabolic drugs, vitamins |
| 6. Biology | Describe the main types of inheritance |

### 4. The tasks for self-directed work when preparing for the class.

#### 4.1. The list of basic terms, parameters, characteristics that students must learn when preparing for the lesson:

*Give a written definition of the following terms:* chorea, myotonia, dystrophia myoplegia, myotonia.

#### 4.2. Theoretical questions for the lesson:

2. Clinical signs and forms of myopathy.
3. The main features of Charcot-Marie-Tooth neural amyotrophy.
4. Clinical signs of spinal amyotrophy.
5. Features of myotonic syndrome.
6. Features of paroxysmal myoplegia.

#### 4.3. Practical tasks that are performed in class:

1. Evaluation and methods of determining the atrophy of muscles, including face, tongue, fibrillar twitching.
2. Examination of deep and superficial reflexes.
3. Evaluation of muscle strength (on a 5-point scale), and the volume of voluntary movements in the arms and legs. Evaluation of strength of the masticatory muscles.
4. Examination of muscle clonus.
5. Content of the topic

<table>
<thead>
<tr>
<th>No.</th>
<th>Questions</th>
<th>Answer models</th>
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<tr>
<td>1.</td>
<td>The principle of classification of hereditary diseases</td>
<td>Syndromological principle and by the level of damage: neuromuscular diseases</td>
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<td>2.</td>
<td>The main clinical signs and clinical forms of myopathies</td>
<td>Symptoms: progressive muscle atrophy, pseudohypertrophy, strengthening of the lumbar lordosis, “goose” gait, alar scapula, masklike face, stair-like rising, quantitative changes of electroexcitability. The main clinical forms: Erb juvenile form, Duchenne pseudohypertrophic form, brachial-scapular-facial, etc.</td>
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<td>3.</td>
<td>The main features of Charcot-Marie-Tooth neural amyotrophy</td>
<td>Atrophy of muscles in the lower legs, areflexia, foot deformities, foot in the form of &quot;inverted bottle&quot;, steppage gait, neurological pain, hypoesthesia in the form of &quot;socks&quot;, in the study of electroexcitability - the reaction of degeneration</td>
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<td>4.</td>
<td>Clinical signs of spinal amyotrophy.</td>
<td>Progressive peripheral paresis, fascicular twitching, the reaction of degeneration.</td>
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<td>5.</td>
<td>What are the features of myotonic syndrome</td>
<td>Myotonic phenomenon - the difficulty of muscle relaxation, increased mechanical excitability.</td>
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<td>6.</td>
<td>Features of paroxysmal myoplegia.</td>
<td>Attacks of paralysis in the extremities, areflexia, in the study of electroexcitability - &quot;cadaveric reaction&quot;.</td>
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Situational tasks

**Task No.1.** The clinic admitted two girls, sisters: one of them is aged 15, another one – 14. At the age of 12, both began to develop weakness in the arms and legs, their gait changed, the disease gradually progressed. Relatives are healthy, their younger brother, 9 years old, is healthy. Neurological status: cranial nerves are without pathology. Weakness in the proximal parts of the hands and feet, patient can rise their hands only by a jerk. When hands are outstretched, winged scapulae are clearly defined. Advanced lumbar lordosis. Hypotrophy of the back muscles. Skeletal muscles are enlarged in volume, dense by palpation. Reflexes of average liveliness. When trying to get out of the floor, both girls are gradually resting with their hands on their knees, thighs, pelvis, abdomen, and then straighten out. Goose gait. No disruption of sensitivity was detected.
Electromyography of trunk muscles and proximal limbs: decreased amplitude, low maximum voltage. Make the diagnosis, determine the clinical form, draw a genealogy chart, establish the type of inheritance, and prescribe treatment.

**Task No.2.** A boy, aged 8, delays in mental development from peers, hardly walks and does not run at all. Up to the age of 4, he developed normally, and then gradually developed weakness in the legs, stopped jumping, running, began to get up from the floor with difficulty. The family had 4 boys, all of them suffered from similar illnesses and died at the age of 12-15 years, there are two older girls, they are healthy. Parents and relatives of the mother (father, mother, sister and brother) are healthy. On examination: muscle atrophy of the proximal parts of the arms and legs, muscles of the trunk, pelvic and shoulder girdles. Marked pseudo-hypertrophy of the calf muscles. Active movements in the legs are limited, strength is significantly reduced. Tendon reflexes from the arms and legs are absent. EMG: changes indicate primary muscle damage. The activity of fructose diphosphate collodia in serum is increased by 100 times. Make the diagnosis, determine the clinical form, type of inheritance, draw a genealogy chart, to prescribe treatment.

**Task No.3.** A patient, aged 29, was admitted to the hospital with complaints of headache, toothache due to the sharp closure of the jaws while sleeping, disrupted movements in the legs – it is difficult to clench and unclench the fingers. The patient considers herself ill for 5 years old. In the past, she had a spontaneous abortion. On examination: hypomimia, slight ptosis of the eyelids, atrophy and weakness of masticatory muscles, weakened closing of the eyes, bared teeth, stretched lips. The voice is silent, the speech is slow. Expressed atrophy of the sternocleidomastoid muscles. Hypotrophy of the limb muscles, especially the legs and forearms. Sharply expressed myotonic syndrome, compressed fingers are slowly and hardly unclenched. On mechanical irritation (a stroke of a hammer) of the deltoid muscle and thenar, there is a stable roller and pit symptom. Reflexes of the limbs are reduced. Marble skin and cold extremities, fingers are thin. Biochemical tests of blood and urine are without pathological abnormalities. From the family history, the following has been found: the elder brother also suffers from such a disease, two more brothers and sisters are healthy. Relatives: the father died at a young age, he had 5 brothers and sisters, three brothers died young. The sons of two brothers suffer from similar diseases. Make the diagnosis, prescribe treatment, draw a genealogy chart.

**Task No.4.** A patient, aged 25, delays in the physical development from his peers since early childhood (hardly able to run, unable to jump). At the age of 13, the muscle atrophy of the shoulder girdle began to develop, and the calf muscles increased in volume. After the age of 18, the disease has almost no progress. Weakness of the muscles of the proximal limbs is observed. The grandfather and a son of a cousin (maternally) suffer from a similar disease. On examination: weakness of the facial muscles. Hypertrophy of the calf muscles is expressed. Moderate atrophy of the muscles of the back and shoulder girdle. When standing up from the chair, the patient leans on the thighs. Muscular strength is moderately decreased in the proximal parts of the extremities, tendon reflexes are sharply reduced. Goose gait. Sensitivity is not disrupted. Intellectual abilities are
preserved. Biochemical studies indicate a high level of serum enzymes, moderate aminoaciduria. Make the diagnosis, to draw a genealogy, to prescribe treatment.

Task No.5. A patient, aged 47, complains of weakness of the muscles of hands and difficulty with chewing. The disease has developed gradually over the course of 10-12 years, the condition worsened significantly about six months ago after tooth extraction. On examination: hypomimia, eyelids are not completely closed, atrophy and weakness of masticatory muscles and orbicular muscle of the mouth. When chewing, the patient facilitates the movements of the lower jaw with his hands. Atrophy of the muscles of the neck and shoulder joints, winged scapulae. Sharply limited movements in the shoulder joints. Abduction of arms is possible only up to 30°. Advanced lumbar lordosis, muscular strength in the legs is satisfactory. Tendon reflexes are low. The gait is slightly rocking. EMG: changes are typical for muscular damage. Make the diagnosis, prescribe treatment.

Task No.6. A patient, aged 25, complains of the difficulty with the first movements in the hands, muscles of the face. This is especially evident in chewing, unclenching fists, moving from the seated to standing position, walking down the stairs and ladder. At repetition of movements, this muscle spasm subsides. He has been ill since the age of 10. The patient noticed changes in the gait, later began to feel spasm in the muscles of the hands and with chewing. He adapted to this defect, but once was nearly knocked down by a trolleybus, since he could not jump away immediately. The same illness afflicts his elder brother. The 10-year-old brother's son complains of spasm in the muscles at the beginning of movements. On examination: athletic structure of the body, muscles are tight. When squeezing the jaws, the patient cannot open the mouth immediately, but after 3-4 repetitions, the opening of the mouth becomes free. With handshake and clenching of his hands in the fist, he cannot immediately open his fingers. After several repetitions, the movements become free. Mechanical behavior; agitation of the muscles. With a stroke of a hammer on thenar, there is a steady tension of the muscles with the contrast of the I finger, with a stroke on the tongue, there is a pit symptom. Make the diagnosis, draw a genealogy chart, recommend treatment.

Task No.7. A patient, aged 32, was hospitalized completely immobile with preservation of movements only in the muscles of the face and chest. Such a situation arose suddenly, in the morning, when he woke up. The previous evening, he visited friends, was eating a lot and drinking alcohol. In the past, there were 2 similar attacks – at the age of 18 and 29, the movements were restored in 2-3 days. On examination: BP: 100/55, total hyperhidrosis, pulse: 92 per minute. Complete absence of movements and reflexes in the legs and hands, muscular hypotension. Sensitivity is not disrupted. Electroexcitability of muscles is absent. A sharp decrease in the level of potassium in serum was detected. By the evening, the state began to improve, the movements in the hands, then in the forearms, appeared. Make the diagnosis, prescribe treatment.

Task No.8. A patient, aged 36, complains of weakness in the legs, difficulty with walking. He has been ill for 15 years, weakness in the legs is slowly progressing, feet hanging while walking, periodic numbness and pain in the shins. Within the last 2 years, weight loss in the hands has started to develop. On
examination: hypotrophy of the muscles of the hands, hands are somewhat reminiscent of "claw-hand", but the strength is satisfactory. Atrophy of the muscles of the legs, legs resembling an inverted bottle, hanging feet. Deformation of the feet – a high arch of the foot. Carporadial, knee, and achilles reflexes are absent. On palpation, there is mild pain in the course of large tibia nerves. Hypesthesia in the distal legs with a decrease in vibration sensitivity. Steppage gait. Heredity: the father suffered from the same illness, there are three brothers and sisters in the family, two brothers have a changed gait and slightly hanging feet. The patient is married, has a son and a daughter, son with a high arch of the foot. Make the diagnosis, draw a genealogy chart, prescribe treatment.

**Task No.9.** A man, aged 17, was referred for examination from the military enlistment office. He complains of abrupt weakness in the muscles of the shoulder girdle, which has imperceptibly appeared at the age of 12. The patient did not seek medical consultation. Family members are healthy. On examination: cranial nerves without pathology. Hypotrophy of the muscles of the shoulder girdle, winged scapulae, reduced muscle strength in the proximal parts of arms. Expressed fascicular twitching in the muscles of the arms and back. Tendon reflexes of arms are absent; the knee and achilles reflexes are preserved. Abdominal reflexes are absent. The gait is not changed. In the study of electroexcitability of muscles of the upper extremities, the reaction of degeneration is revealed. Make the diagnosis, prescribe treatment.

**Task No.10.** A patient, aged 27, complains of weakness in the legs, abrupt difficulty with walking. The disease has developed gradually over the period of 10 years, growing stiffness in the legs was increased all the time. The father suffered from the same illness, he could not walk for several years, died 3 years ago. Neurological status: cranial nerves are without pathology. Movement and muscle strength in their hands are sufficient. Active motions are very limited, sharply expressed spastic hypertension. Knee and achilles reflexes are high, clonus of the patella and feet, on both sides – the symptoms of Babinsky, Oppenheim, Scheffer, Gordon. The patient walks with a sharp pronounced spasticity (spastico-paraparetic gait). The sensitivity and function of the pelvic organs are not disrupted. Liquor: Pressure – 150 mm. H2O, protein – 0.33 g / l, cytosis – 2 lymphocytes per 1 mcL, RW – negative. Make the diagnosis, prescribe treatment.

**Task No.11.** The mother of a newborn, aged 3 months, complains that the infant is sluggish, hardly sucking, at feeding cannot suck more than 50 ml of milk, it is constantly sleeping, there are frequent regurgitation and hiccup. The infant virtually does not move the limbs, is very calm, cries weakly. The woman had already had a child, who had defects in the development of the bone skeleton, did not sit at all and died at the age of 3 years. Neurological status: the infant is sluggish, does not hold the head, the head is enlarged, blood vessels on the head are dilated, pulsating fontanel, reduced sucking and search reflexes, pharyngeal reflex is absent. Fibrillations are visible on the tongue. Make a preliminary diagnosis.

**Task No.12.** A young boy, aged 16, during the physical education lesson failed to complete 10 knee-bending exercises. After interviewing the mother, it was
found that for several years the boy virtually does not perform any tasks at home because he gets very tired. When she asked him to screw in the light bulb, he could not hold his arms up and the bulb fell. However, this fact did not cause suspicion to the mother, because nobody in the family is able to perform such tasks: the father and 2 elder sons, as well as paternal grandfather and his brother are very weak in their hands. Examination revealed facial amimia, muscle atrophy of the shoulder girdle, winged scapulae, large lips, which protrude forward. Pseudo-hypertrophy of the leg muscles, decreased muscle tone, reduced tendon reflexes on the hands. Make the diagnosis, prescribe treatment.

Task No.13. A young man, aged 15, during the check-up at the military enlistment office, showed signs of muscular fatigue: he was unable to perform knee-bending exercises, stopped several times while walking, goose gait. During the examination by ophthalmologist, color-blindness was detected. From the history, it was found that fatigue has been troubling him for several years, at physical education lessons he cannot meet the required norms, but it does not cause major problems in everyday activities. The father also has the goose gait. Neurological status: symmetric atrophies of the thighs and pelvic girdle, pseudohypertrophy of the calf muscles are detected. Muscle tone is slightly decreased in the proximal legs, knee reflexes are decreased. No pathological signs and sensitive disorders were detected. Electromyography revealed quantitative changes in electricoexcitability. In the biochemical analysis of blood: increased transferase and creatine phosphokinase. Make the diagnosis, prescribe treatment.

Task No.14. A woman with a daughter, aged 18, consulted a doctor with complaints of attacks, during which the girl completely loses the ability to move. Consciousness is retained, she can move the head and eyes, she can talk and breathe. The attack is accompanied by profuse sweat, salivation, decrease in blood pressure to 80/50 mm Hg, and lasts for 3-4 hours. After the attack, she experiences significant weakness. The patient is unable to do anything for a few days. The first attack occurred in childhood after a cross at a physical education lesson. It was transient and was associated with physical overexertion. The patient entered the university a year ago. When preparing for examinations, an attack occurred, which was repeated during the second session. The conducted examination did not reveal deviations from the norm. Make a preliminary diagnosis and give recommendations for further patient management.

Task No.15. A young man, aged 15, of athletic physique, failed to perform simple knee-bending exercises when undergoing check-up by the commission at the military enlistment office. From anamnesis, it was found out that since childhood, the young man was distinguished for his strength. He is part of the arm-wrestling team, and practically has no defeats during the competition. However, during the physical education classes he cannot go in the ranks and run. At the neurological examination, the following was revealed: diffuse hypertrophy of muscles, tonic spasms of the facial muscles, with a stroke of a hammer in the area of the thenar, a steady motion of the thumb to the little finger occurs; when a hammer hits the muscle, there is a local steady tension in the form of a roller
symptom. Reflex, sensitivity and coordination functions have not been changed. Robot-like movements when walking. Make the diagnosis and prescribe treatment.

Tasks for self-check:

1. What trace element plays a key role in the emergence of an attack of paroxysmal myoplegia?
   A) K
   B) Ca
   C) Cl
   D) Na
   E) Mg

2. Inheritance of Erb-Roth muscular dystrophy occurs by:
   A) autosomal recessive
   B) autosomal dominant
   C) coupled with Y-chromosome
   D) X-linked
   E) correct answers are A and D

3. What is the pathognomonic symptom for Thomsen's myotonia?
   A) correct answers are C and B
   B) plates symptom
   C) speech sign
   D) symptom of thumb
   E) correct answers are D and B

4. What is one of the first signs of Charcot-Marie-Tooth neural amyotrophy?
   A) abnormal fatigue in the distal extremities
   B) progressive muscular atrophy
   C) sensory ataxia
   D) sensitivity disorders behind polyneural type
   E) intellectual disability

5. What is the main manifestation of myotonia?
   A) difficulty in relaxing muscles
   B) muscles pseudohypertrophy
   C) muscle weight loss
   D) bouts of paralysis in the arms and legs
   E) muscle fatigue

6. At what age does the early form of spinal Werdnig-Hoffmann amyotrophy occur in children?
   A) at 6 months of life
   B) at about 1 year of life
   C) at 4 months of life
   D) at 2 months of age
   E) at birth

7. When does the attack occur more frequently in periodic paralysis?
   A) all answers are correct
B) at mealtime  
C) during exercise  
D) during sleep or immediately after it  
E) with stress  
8. What changes on electromyography will we observe during the attack of periodic paralysis?  
A) complete lack of response  
B) pathological waves  
C) decrease in wave amplitude  
D) the appearance of additional waves  
E) the random distribution of the waves  
9. In most cases, the lesion in Adolph Strümpell familial spastic paralysis is found at:  
A) brain stem and thoracic spinal cord  
B) cortex  
C) internal capsule  
D) lumbar spinal cord  
E) cauda equina  
10. What is the appearance of a patient with Thomsen's myotonia?  
A) athletic  
B) thin with atrophic muscles  
C) muscles pseudohypertrophy in the form of "inverted bottle"  
D) he walks, highly lift foots - "steppage" walking type  
E) no change in appearance  
11. What is most characteristic for the clinical presentation of progressive Erb-Roth muscular dystrophy?  
A) atrophy of proximal muscle groups  
B) atrophy of distal muscle groups  
C) fiber twitching in the muscles of the upper limbs  
D) fiber twitching in the muscles of the lower extremities  
E) myopathic face  
12. Select the drug during the attack of hypercalcemia form of periodic paralysis.  
A) intravenous administration of 10% calcium chloride solution  
B) introduction panangin  
C) intravenous administration of 40% glucose solution  
D) introduction of 25% solution of magnesium sulfate  
E) intravenous administration of 10% solution of potassium chloride  
13. Where is the topical level of myopathy lesion?  
A) muscles  
B) neuromuscular synapse  
C) peripheral nerve  
D) front horn of the spinal cord  
E) nerve plexus  
14. Increase of which component of blood serum occurs at the primary muscular dystrophy?
A) CK  
B) ALT  
C) ATP  
D) GABA  
E) AST

15. What method of study will you choose to diagnose forms of periodic paralysis?  
A) the study of blood electrolytes  
B) overall analysis blood  
C) radiography  
D) reovasography  
E) electromyography

References

Basic:

Additional

Web resources:
1. www.umsa.edu.ua/  
2. http://www.likar.info/neurology/article-64140-  

The methodical guidance has been compiled by H.Ya. Sylenko

The methodical guidance has been considered and approved at the meeting of the Department of Nervous Diseases with Neurosurgery and Medical Genetics

___________________________________________________________________

and subsequently revised (amended)
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