DOI 10.26724/2079-8334-2020-2-72-32-35
UDC 617.52-007

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PARRY-ROMBERG SYNDROME: DIFFICULTIES OF DIAGNOSIS AND IMPROVEMENT OF TREATMENT

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The article described a case of a rare disease – progressive facial hemiatrophy in a patient of the Neurological Department of Poltava Regional Clinical Hospital. We reviewed a literature on the problem, analyzed the etiological factors, pathogenesis and clinical characteristics of the disease. There was defined a range of necessary examination methods of patients, which included examination by a neurologist, rheumatologist, endocrinologist, as well as the use of additional instrumental and laboratory research methods. Comprehensive treatment tactics include the use of trophotropic, vascular drugs, anabolic hormones, potassium supplements, massage, physical therapy.

Key words: facial hemiatrophy, Parry-Romberg syndrome, neurodental syndrome.

The work is a fragment of the research project "Clinical and pathogenetic optimization of diagnosis, prognosis, treatment and prevention of complicated disorders of the central nervous system, as well as neurological disorders in somatic pathology", state registration No. 0116U004190.

Parry-Romberg syndrome is a progressive facial hemiatrophy, neurodental syndrome. The disease incidence is 1 case per 2,000 people [1, 4]. According to the information provided by the National Organization for Rare Diseases (NORD), Parry-Romberg syndrome affects about 1,000,000 people worldwide [5]. The first mention of this disease is found in the study of English physician Caleb Hillier Parry, published in 1825. The disease was later described in detail by the German neurologist Moritz Heinrich Romberg, who included this syndrome in the category of trophoneurosis [4].

Most often, Parry-Romberg syndrome occurs at the age of 10-20 years and very rarely in people over 30 years. The prevalence is higher in women than in men in a ratio of approximately 3:0.2 [3]. The left side is affected more often than the right. Most cases are considered sporadic. At the same time, familial cases are described, presumably with autosomal recessive and autosomal dominant types of inheritance and incomplete penetrance. The etiology and pathogenesis of the disease are still unknown, there is suspicion about the multifactorial genesis. A number of authors [3] consider progressive facial hemiatrophy as a variant of scleroderma, but in contrast to systemic sclerosis, the process of atrophy is limited to unilateral skin lesions, subcutaneous tissue and is not accompanied by other manifestations of collagenosis.

In the occurrence of Parry-Romberg syndrome are important lesions of the hypothalamus, pathology of the sympathetic division of the autonomic nervous system, lesions of the superior sympathetic ganglion, trigeminal nerve system, focal lesions of the cerebral cortex. The connection of facial hemiatrophy with cerebrovascular disease in the brain stem is described [4]. The onset of the disease is sometimes preceded by injuries to the face and skull, infectious diseases. In some cases, Parry-Romberg syndrome is manifested as a syndrome of syringomyelia, syphilis, brain tumors, cerebral echinococcosis. There is an assumption that patients have congenital functional changes of the autonomic nervous system at the level of suprasegmental stem–diencephalic divisions, and these factors play the role of a "trigger" mechanism [3].

The purpose of the study was to highlight the current view of a rare disease – Parry-Romberg syndrome, the diagnosis of which presents significant difficulties, and to present our own observations with atypical clinical symptoms, as well as determining the range of necessary examination methods in such patients and improving the treatment of this disease. The relevance of this study is determined by the unexplored pathogenesis and etiology of the disease, the occurence of a serious cosmetic defect, the lack of differential diagnostic criteria and adequate treatment regimens.

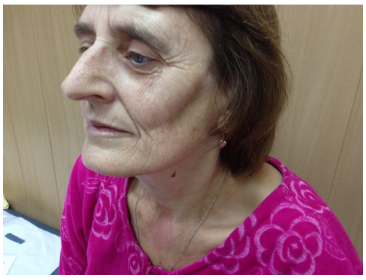
Materials and methods. The following examination methods were used to diagnose the Parry-Romberg syndrome: clinical (objective and neurological examination of the patient, consultation with a rheumatologist). Laboratory methods: complete blood count, clinical urine test, biochemical blood test (including creatine phosphokinase), electrolytes (to exclude the acute infectious process, metabolic disorders), thyroid hormones (to exclude thyroid gland disorders), rheumatoid factor tests – to clarify the presence of systemic dysplasia of connective tissue. Instrumental examinations: ECG, REG (to determine cerebrovascular disorders), electroneuromyography (ENMG) – to assess the primary lesion of a muscle or nerve structure. Neuroimaging methods (MRI of the brain – to exclude the expansive intracranial process, acute cerebrovascular accident, neurosyphilis, post-traumatic or parasitic lesions).

Results of the study and their discussion. Patient K., 59 years old, was hospitalized in the Neurological Department of Poltava Regional Clinical Hospital. Was admitted with the next complaints: facial asymmetry (left side deformity). History of the present illness: facial asymmetry occured 2 years ago for no apparent reason. First patient noticed a thinning of the left cheek muscles, then – the forehead and later – the neck. Past medical history: grew and developed according to gender and age. Patient denied any previously suffered diseases, injuries, operations, infections. Heredity was not burdened. There was no bad habits. Clinician-observed: the patient's condition was satisfactory, the skin was normal color, vesicular respiration in the lungs, without wheezing, rhythmic heart sounds, heart rate – 74 per minute, blood pressure – 140/80 mm Hg. Body temperature was 36.6 °C. The abdomen was soft, with painless palpation. Healthy bladder and bowel habits.

Neurological status: consciousness was clear, oriented to person, place and time. There were no meningeal signs. Cranial nerves: pupils D=S, palpebral fissure – mild ptosis of the upper eyelid on the left, without nystagmus, smoothed left nasolabial fold, atrophy of the left buccinator muscle, masticatory muscles, frontalis muscles on the left, subcutaneous tissue on the left (Fig. 1a, c). The motor function of the facial muscles was preserved. There was mild hypotrophy of the left sternocleidomastoid muscle (Fig. 1b). The skin of the face and neck on the affected side was without foci of depigmentation. Deviation of the tongue to the left, mild atrophy of the muscles of the left side of the tongue, twitching of the tongue. The tone of the limb muscles was normal, the strength is sufficient, there were no pathological reflexes. Reflexes of the upper and lower extremities were of medium vivacity, without asymmetry. There was no atrophy of the limb muscles. Coordination tests were performed satisfactorily. In Romberg's standing position was stable. Violations of superficial and deep sensitivity were not detected. Pelvic functions were not impaired.

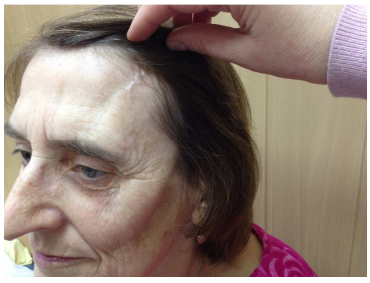
A

Fig. 1a. Patient K., anterior view



B

Fig. 1b. Patient K., lateral view



C

Fig. 1c. Patient K., frontalis muscles atrophy

Examination results: complete blood count, clinical urine test, biochemical blood test, electrolytes, thyroid hormones, rheumatoid factor tests – within normal limits. ECG – without pathology. The rheumatologist excluded connective tissue pathology.

REG: pulse blood volume in the left carotid system was increased, in the vertebrobasilar arterial system – reduced, in the right carotid system – normal. In all arteries there were signs of increased arterial tone. In the vertebrobasilar system there were signs of complications in venous outflow, in the carotid system venous outflow was normal.

MRI of the brain (December 22, 2017): In the left frontal lobe near the anterior horn of the left lateral ventricle, in the left side of thalamus and in the left parietal lobe area near the posterior horn of the left lateral ventricle, the foci of MRI signal hyperintensities on T2WI, FLAIR, iso-/low signal intensity on T1WI, irregularly shaped, with clear uneven contours up to 5-6 mm in diameter were determined, the MRI signal on diffusion was not changed (probably, foci of dyscirculation). Conclusion: foci of altered MRI signal of the left hemisphere of the brain (probably of vascular origin). (Fig. 2, 3).

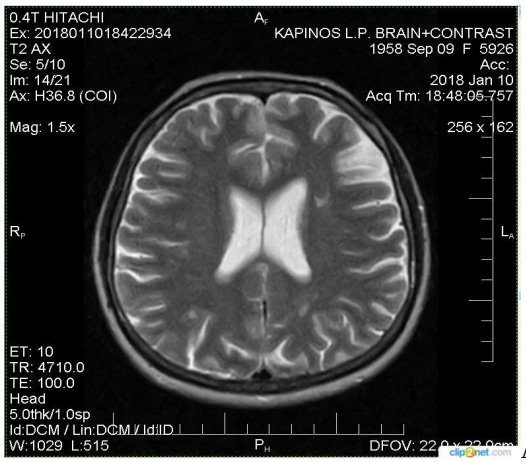
On MRI of the cervical spine (December 14, 2017): Common degenerative disc disease of the cervical spine, stage I-II of osteochondrosis, posterior central protrusion C3-C4 of the intervertebral disc up to 2 mm. Osteophytes were up to 4 mm of C6 and C7 vertebral bodies.

ENMG: the obtained data corresponded to the primary myopathy of the facial muscles and the muscles of the left side of shoulder girdle.

Based on the history of the present illness, clinical evidence and examination results, the diagnosis was made: progressive left-sided hemifacial atrophy.

The patient was prescribed treatment: actovegin, tocopheryl acetate, piracetam, neurotropin, neuromidin, darsonvalization of the head and neck area. It was recommended to take outpatient courses of vascular and metabolic therapy, neuroprotective agents, vitamins, physiotherapy, and massage 1-2 times a year.

Usually first develops atrophy of a particular facial area (orbital cavity, cheeks, lower jaw), atrophy of the skin and its depigmentation or hyperpigmentation, then atrophy of the subcutaneous tissue, muscles, bones. Muscle motor function was usually little disturbed. There were no objective sensitivity disorders, Horner-Bernard syndrome can be observed. Sometimes atrophy affected the neck, arm and even half of the body on the same and opposite sides. Parry-Romberg syndrome could be accompanied by alopecia, atrophy of the tongue, various occlusal disorders, refractive pathology, heterochromia of the iris.



A

Fig. 2. T2WI MRI of the brain of patient K. (January 10, 2018)

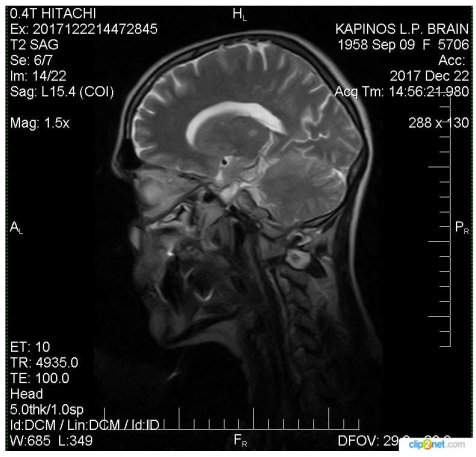


Fig. 3. T2WI MRI of the brain of patient K.. (December 22, 2017)

Facial hemiatrophy syndrome in lesions of the segmental autonomic system most often occured due to pathology of the superior sympathetic ganglion or lateral horns of the spinal cord CVII-DII or white ramus communicans of this level. It was characterized by minor soft tissues atrophy of half side of the face, mainly cheeks, hyperpigmentation, very slow progression, frequent combination with sympathetic pain, changes in the intensity of the iris color, moderate dilation of the pupil [1].

In Parry-Romberg syndrome of the stem level, the causes were usually syringomyelia, stem encephalitis, brain tumor and other pathological processes. In syringomyelia, autonomic dysfunction in the affected side of the face was manifested by complete or incomplete Horner-Bernard syndrome, cyanosis or skin paleness, the presence of wrinkles and uneven distribution of hair, as well as pain on palpation of the neurovascular bundle running along the anterior surface of the neck. Often patients had diffuse hypotrophy of the soft tissues of the cheek, dissociated sensitivity in the arm and torso, mild bulbar disorders, horizontal nystagmus. It was characterized by the absence of a gross cosmetic defect [1].

Peripheral Parry-Romberg syndrome could be of iatrogenic origin and occured, in particular, due to procaine-alcohol blockade in patients with trigeminal neuralgia. In these cases, trophic disorders occured in the area of innervation of the affected communicans. They were manifested by moderate soft tissue atrophy in the area of the cheek, eyelid, lower jaw, the tip of the nose and its alae, as well as the masticatory and temporal muscles. There also were swelling of the cheeks, hair loss, focal hyperpigmentation in the fronto-temporal lobe or abundant moles on the cheek. The disease can begin with trigeminal neuralgia, then there is atrophy in any limited area. The atrophic process can spread to the corresponding side of the neck and torso, affecting the vocal cords and half of the larynx. In severe forms of the disease there is a thinning and sagging of the chin bone, jaw reduction, tooth loss. Ophthalmic symptoms occur in 15% of patients. Typical: loss of eyelashes, eyebrows, enophthalmos, ophthalmoparesis (due to atrophy of orbital tissue and extraocular muscles on the affected side), narrowing of the eyelid, keratitis [3]. The disease progresses slowly over 3-5 years, after which, as a rule, the condition stabilizes.

At affection of a cerebral cortex besides a facial hemiatrophy, neurologic symptoms of lesions of brain structures were defined (focal Jackson's and generalized epileptic seizures, upper motor neuron lesion, cerebellar disorders). At carrying out EEG, the focal epileptic activity in a homolateral hemisphere could be found. On the affected areas, there may be graying and loss of hair on the head, eyebrows and eyelashes, there is no sweating. Muscle motor function was little disturbed.

The diagnosis is based on the progressive development of tissue atrophy of one side of the body or only the face with the presence of autonomic and trophic disorders [4]. There is no radical treatment for Parry-Romberg syndrome. Therapeutic tactics include the use of trophotropic drugs, anabolic hormones, potassium supplements, anticonvulsants and dehydrators, massage, physical therapy.

In case of irreversible paralysis of the facial muscles, surgical treatment is indicated: static and kinetic suspension of the lowered tissues, musculoplasty, canthoplasty – plastic surgery for the case of the eyelid narrowing, that is, its elongation and expansion [1]. In our clinical case, there were found dyscirculatory foci in the left and left frontal and parietal lobes due to hemodynamic disturbances in the vertebrobasilar arterial system, which were the probable cause of Parry-Romberg syndrome in the patient. Thus, the above-described case of progressive facial atrophy is characterized by a typical for this disease gradual onset, slow progress with the development of facial asymmetry, atrophy of facial muscles, tongue, subcutaneous fat on the left with preserved motor muscle function.

Conclusion

Facial hemiatrophy or Parry-Romberg syndrome can be either an independent disease or a syndrome of the prior disease (scleroderma, syringomyelia, encephalitis, lesions of the sympathetic ganglion, etc.). It is advisable to comprehensively examine patients, which includes examination by a neurologist, rheumatologist, endocrinologist, as well as the use of additional instrumental and laboratory diagnostic methods (electroneuromyography, rheoencephalography, electroencephalography, MRI of the brain and cervical spinal cord). The disease requires the application of non-specific therapy, including vasoactive, metabolic drugs, antihypoxants, neuroprotectors, anticholinesterase drugs. With regard to the patient's life, the prognosis is favorable, with regard to recovery – unfavorable, but early diagnosis, regular combined treatment is necessary to achieve a positive effect.

Prospects for further research are: it is planned to continue monitoring the course of the disease in the patient, re-examination and additional courses of maintenance therapy.

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Реферати

СИНДРОМ ПАРРІ-РОМБЕРГА: СКЛАДНОСТІ ДІАГНОСТИКИ ТА ВДОСКОНАЛЕННЯ МЕТОДИКИ ЛІКУВАННЯ

Гладка В.М., Пурденко Т.Й., Таряник К.А.

Представлений опис випадку рідкісного захворювання – прогресуючої атрофії обличчя у пацієнтки неврологічного відділення Полтавської обласної клінічної лікарні. Проведений огляд літератури щодо проблеми, проаналізовані етіологічні чинники, патогенез та клінічні характеристики захворювання. Визначене коло необхідних методів обстеження хворих, яке включає огляд невролога, ревматолога, ендокринолога, а також використання додаткових інструментальних і лабораторних методів дослідження. Комплексна лікувальна тактика передбачає використання трофотропних, судинних засобів, анаболічних гормонів, препаратів калію, масаж, ЛФК.

Ключові слова: геміатрофія обличча, синдром Паррі-Ромберга, нейростоматологічний синдром.

Стаття надійшла 18.07.2019 р.

СИНДРОМ ПАРРИ-РОМБЕРГА: СЛОЖНОСТИ ДИАГНОСТИКИ И УСОВЕРШЕНСТВОВАНИЕ МЕТОДИКИ ЛЕЧЕНИЯ

Гладка В.М., Пурденко Т.И., Таряник К.А.

Представлено описание случая редкого заболевания – прогрессирующей атрофии лица у пациентки неврологического отделения Полтавской областной клинической больницы. Проведен обзор литературы по проблеме, проанализированы этиологические факторы, патогенез и клинические характеристики заболевания. Определен круг необходимых методов обследования больных, включая осмотр невролога, ревматолога, эндокринолога, а также использование дополнительных инструментальных и лабораторных методов исследования. Комплексная лечебная тактика предполагает использование трофотропных, сосудистых средств, анаболических гормонов, препаратов калия, массаж, ЛФК.

Ключевые слова: гемиатрофия лица, синдром Парри-Ромберга, нейростоматологический синдром.

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