ტებში ფქოდ-ით, საკონტროლოსთან შედარებით, შემცირდა 2,72-ჯერ, ახალგაზრდა ასაკის პაციენტებთან შედარებით კი – 1,88-ჯერ. GOLD I –ში ოსტეოკალცინის შემცველობის შემცირება აღინიშნა 10 (66,67%) პაციენტში, GOLD II –ში - 22-ში (89,0%),GOLD III–ში – 17-ში (85,0%), GOLD IV–ში - ექვსივე (100%) პაციენტში.

ვიტამინი D-ს კონცენტრაცია შემცირებული იყო ყველა პაციენტში ფქოდ-ით, ხოლო ვიტამინი D-ს მძიმე დეფიციტი დიაგნოსტირდა 45 წლამდე ასაკის პაციენტთა 23,08%-ში, ხანდაზმული ასაკის პაციენტების 70,59%-ში და მოხუცებულობითი ასაკის ყველა პაციენტში.

GOLD IV–ში ვიტამინი D-ს დონე შემცირებული იყო 1,75-ჯერ, GOLD I-თან შედარებით. ვიტამინი D-ს მძიმე დეფიციტი დიაგნოსტირდა: GOLD I –ში - 7 (46,67%) პაციენტში, GOLD II –ში - 10 (40,0%), GOLD III–ში – 13 (65,0%), GOLD IV–ში – ყველა პაციენტში.

მიღებული შედეგები მიუთითებს, რომ ასაკის მატებასთან და ფქოდ-ის დამძიმებასთან ერთად აღინიშნება ძვლოვანი ქსოვილის ფორმირების მარკერების წარმოქმნის დათრგუნვა. ეს პროცესები მიმდინარეობს ვიტამინ D-ს დეფიციტის ფონზე. ასეთი დისბალანსი ქმნის ხელსაყრელ პირობებს ოსტეოპოროზის განვითარებისათვის. იმის გათვალისწინებით, რომ ამ დარღვევების პირველი ნიშნები, კერძოდ - ვიტამინ D-ს და ოსტეოკალცინის დონის შემცირება, დიაგნოსტირდება უკვე GOLD I –ში, შეიძლება გაკეთდეს დასკვნა, რომ ფქოდ წარმოადგენს წამყვან ფაქტორს.

CLINICAL CHARACTERISTICS OF ALS IN GEORGIAN PATIENTS

[1]Kekenadze M., [2]Kvirkvelia N., [1]Beridze M., [3]Vashadze Sh., [1]Kvaratskhelia E.

[1]Tbilisi State Medical University; [2]P.Sarajishvili Institute of Neurology; [3]Batumi Shota Rustaveli State University, Georgia

"Does it take place through simple propagation, extending gradually across the neuroglia?" [1] This is what French Neurologist -J.M Charcot has been questioning regarding amyotrophic lateral sclerosis progression in his lectures on the diseases of the nervous system in 1877. It's been 145 years since, even though many questions have been answered, the cause of amyotrophic lateral sclerosis (ALS) remains today unknown for most of the patients with the disease. the aim of this article is to describe the clinical characteristics of Georgian ALS patients. ALS onset and progression vary greatly among individuals, so these data provide additional insight into the phenotypic differences within a national population. An understanding of symptoms of ALS onset may help clinicians make a quicker diagnosis, which could lead to earlier therapeutic interventions.

Material and methods. Overall 47 patients with ALS were investigated, among them 24 male (51.06%), 23 female (48.9%), aged 25-84 (Table 1) we documented clinical manifestations of the disease in those patients, age at diagnosis, Patient survey of clinical symptoms was taken using Mayo Clinic Lab. Neurology patient form , Cognitive changes assessed via Addenbrooke Cognitive Examination scale (ACE III), and frontal behavioral inventory, diagnosis of FTD was based on Strong criteria of FTD. Patient functional status was assessed with ALSFRS-R. Diagnosis of ALS was based on the new Gold coast criteria -incorporating progressive motor impairment documented by history or repeated clinical assessment, preceded by normal motor function, and presence of upper and lower motor neuron dysfunction in at least 1 body region, (with upper and lower motor neuron dysfunction noted in the same body region if only one body region is involved) or lower motor neuron dysfunction in at least 2 body regions, most importantly excluding other diseases.[7] All patients underwent nerve conduction studies and needle electromyography , In patients with signs of dementia or suspicion of other diseases MRI of Head and Spine was done. Those with unexplained sensory signs or symptoms, abnormal nerve conduction studies, weakness in the distribution of individual motor nerves, or any abnormality on cervical or head MRI suggestive of an alternate diagnosis such as spinal stenosis or cervical myelopathy, multiple sclerosis were not included in the study. Patients diagnosed with conditions such as spinal muscular atrophy, Kennedy syndrome, monomelic amyotrophy, Hirayama syndrome, or multifocal motor neuropathy were excluded from the study.

Results and discussion. *Age of onset*. Recent studies have shown that the mean age of ALS onset is between 51 and 66 years [4]. When compared with patients from Asian countries , ALS usually strikes patients in Europe later in life. The greater age at ALS onset in Europe may be partly explained by the use of population-based studies [4,5] the mean age at ALS onset in Georgian was found to be 58.30 years, patients were aged 26 to 84 years, 63.8% of the patients were 50–69 years old (Table 1) ALS begins with nonspecific symptoms that can mimic those of other neuromuscular diseases. ALS diagnosis can therefore be delayed if a misdiagnosis occurs in the early stages. Due to the lack of valid diagnostic biomarkers, diagnostic delay is marked. ALS is diagnosed clinically through progressive symptoms, which takes time to demonstrate. According to recent studies, diagnostic delays typically range from 9 to 24 months in different populations [6,8,9]. According to our data, the average time to diagnose ALS was 6 to 15 months from onset of symptoms, key factors for diagnostic delay were referrals to specialists rather than neurologists, and consequent misdiagnosis resulting in unnecessary procedures, patients with a bulbar onset were diagnosed earlier than those with spinal onset.

Site of onset. The "Site of Onset" is defined as the part of the body where the patient first experienced symptoms of ALS, most of the time which is muscle weakness. Body parts are divided into three categories: 1) limb 2) bulbar and 3) truncal - involving neck, back or abdominal areas, breathing muscles. Classical -typical ALS involves simultaneous UMN and LMN signs, leading to gradual muscle weakness that moves from one body part to another in a propagating manner . Most commonly, it begins in one of three body regions (face, arm, or leg), it rarely begins in the muscles of the trunk or respiratory system. It might seem that the ALS phenotype is similar between populations, but there is a subtle difference in the clinical presentation across European registries.[3] Atypical phenotypes of ALS currently recognized include:

1. Progressive bulbar palsy (PBP)
2. Primary lateral sclerosis (PLS)
3. Progressive muscular atrophy (PMA)

- The progressive bulbar palsy affects only the muscles supplied by bulbar motor nuclei and the corticobulbar pathways. Both upper and lower motor neuron deficits are discernible.
- Progressive muscular atrophy is diagnosed when there is lower motor neuron involvement in one limb or region and clinical or electrophysiological evidence of involvement in an adjacent limb or region. When the syndrome affects at least two body regions, ALS can be diagnosed without UMN signs according to Gold Coast Criteria.
- Primary lateral sclerosis is characterized with upper motor neuron deficits without LMN involvement. It is possible to diagnose ALS if there is clinical or electrophysiological evidence that the lower motor neurons in at least one limb or body region are involved.[7]
- According to the site of onset and consequent propagation in body regions ALS is classified as follows :
- Flail arm syndrome (Vulpian-Bernhardt syndrome) brachial amyotrophic diplegia
- Flail leg syndrome (Marie-Patrikios) pseudopolyneuritic variant
- Bulbar onset
- Mills Variant (hemiplegic).

We assessed the clinical characteristics of ALS patients and classified them accordingly. Site of onset and ALS variants among the 47 patients is presented in Table 2. 97.8% of patients had progressive muscle weakness before being diagnosed with ALS. Only in 2.1% (n-1) of patients symptoms started with UMN sign- Spasticity and has been diagnosed with Primary lateral Sclerosis, after two years of symptom onset LMN signs have appeared, and subsequently resembling typical ALS.

Almost 95% of participants had a "typical"ALS , defined as UMN and LMN signs in one region or more, and Only 2.1% - PLS , 4.3%- PMA demonstrating LMN deficits in two or more body regions with clinical examination or Electrophysiological studies. A significantly higher proportion of patients had limb onset weakness, in particular upper limb onset variant of ALS (UL-ALS), lower limb onset ALS (LL-ALS) coming to close second. In majority of patients propaganaton manner was as follows: Ipsilateral Limb (UL or LL) to Contralateral Limb According to our study pseudopolyneuritic variant (Flail leg) and brachial amyotrophic diplegia (Flail arm) variant of ALS were not identified. For this we suggest a reason to be long term follow up of involved patients, and therefore more chance of spreading disease to different regions of the body. All of our patients with LL or UL onset later on developed deficits in bulbar, cervical, truncal or lumbar body regions, which categorizes them as "typical" ALS. Usually bulbar onset ALS progresses most rapidly and is characterized by the lowest survival rate (*2 years post diagnosis), and a markedly reduced quality of life[13,14] Our results indicate that approximately 1/4 of investigated patients had B-ALS (bulbar onset ALS). time from symptom onset and diagnosis was shorter than in LL-ALS and UL-ALS. Following symptoms were observed in B-ALS patients: gradual onset of difficulty with speech (dysarthria) and swallowing (dysphagia); breathing, excessive salivation, tongue atrophy and fasciculations, dysphonia -hoarseness of voice. One patient who presented with above mentioned symptoms in addition with brachial muscles atrophy and fasciculations demonstrated significant decrement after Repetitive Nerve stimulation (RNS) on Electrophysiological studies, subsequently specific antibodies were tested for differential diagnosis of Myasthenia Gravis (MG) ,which led to diagnosis of anti-MuSK (Muscle-specific tyrosine kinase) MG. An important lesson from this case is to rule out anti-MuSK MG, a disease which can talentedly mimic Bulbar onset ALS.

Table 1. Age at diagnosis and Gender ratio in ALS patients

Characteristic	**n-47**	**%**
Age at diagnosis		
18-39	4	8.5%
40-49	4	8.5%
50-59	11	23.4%
60-69	19	40.4%
70-79	6	12.7%
Gender		
Male	24	51%
Female	23	49%

Table 2. ALS Phenotypes in Georgian patients

Flail Leg Phenotypic Variant	UMN	LMN	n	%
Based on neuronal level				
Typical ALS	+	+	44	93.6%
PLS	++	-	1	2.1%
PMA	-	++	2	4.3%
PBP	+	+	0	0
Based on Somatic Region and site of onset				
Bulbar ALS			10	21.3%
UL-ALS			18	38.3%
LL-ALS			19	40.4%
Mill's (hemiplegic) variant			0	0
Flail Arm			0	0
Flail Leg			0	0

ALS, amyotrophic lateral sclerosis; UMN, upper motor neuron; LMN, lower motor neuron; + typical to variable degree; ++ primary feature, - Not a feature, PLS- Primary Lateral sclerosis, PMA -Progressive muscular atrophy, PBP- Progressive Bulbar Palsy, UL-ALS- upper limb onset ALS, LL-ALS - Lower Limb onset ALS

Table 3. ALS Plus syndromes

Extrapyramidal Signs	n=47	%
	6	12,7
Autonomic Dysfunction	10	21,3
Cerebellar Dysfunction	4	8,5
No-ALS Plus	27	57,5

Frontotemporal dementia in ALS patients. Over the last two decades overlap between FTD and ALS became more evident. A view that ALS and FTD share a common biology and exist on the same pathological spectrum is worned out. Clinically, it is now recognized that up to 50% of ALS patients have some degree of cog- nitive or behavioral impairment[15,16]. The findings confirm that, together with ALS-FTD, these disorders can be considered progressive disorders that are part of a multisystem degenerative disorder. In addition, the single most common genetic cause of either ALS or FTD is mutation in C9ORF72, which is known to cause both diseases or a combination of both [17,18].

We have screened ALS patients for FTD, used addenbrooke's cognitive examination (ACE -III) to assess neuropsychological status and to identify cognitive impairment. The ACE encompassed tests of five cognitive domains: attention/orientation, memory, language, verbal fluency, and visuospatial skills. It is scored out of 100, with a higher score denoting better cognitive function. Cut-off score used is 83. The presence of frontotemporal atrophy, defined by MRI imaging, was a sensitive indicator of a frontotemporal lobar degeneration in ALS. Caregivers completed Frontal Behavioral Inventory- FBI -24 item inventory to assess behavior and personality changes associated with behavioral variant of frontotemporal dementia via caregiver report. While we were diagnosing ALS-FTD, we kept into account possible pitfalls in diagnosis due to comorbid conditions like: 1. other neurological conditions such as cerebrovascular disease 2. systemic conditions such as hypothyroidism or diabetes, 3. pharmacological conditions such as substance/drugs abuse or 4. psychiatric conditions such as depression, anxiety, psychosis [19]. Patients demonstrating above mentioned symptoms were excluded from study.

Frontotemporal dementia (FTD) was diagnosed in 6 patients (12.7%) whose symptoms were characterized by cognitive and behavioral dysfunction associated with changes in personal and social conduct. Within FTD patients memory and visuospatial abnormalities remained intact, which was a factor of differentiating FTD from different types of Dementia.Verbal Fluency, language was affected mostly, FTD patients demonstrating lack of fluent speech, Language difficulties. In accordance to FBI filed by caregiver - Patients diagnosed with FTD demonstrated 1.disinhibition (the patients behaved in a socially inappropriate manner, is overactive and distracted, and often exhibited binge eating behaviors), 2. apathy (The patient displayed apathy, inertia and loss of volition, distractibility) and 3. stereotypic behaviors (the patient performed stereotypic ritualistic behaviors). Neuroimaging -Head MRI -showed different levels of bilateral Frontotemporal atrophy in FTD patients. FTD developed later on after developing MND signs in most of the cases, only in one patient signs of frontotemporal dementia appeared two years before MND signs, characterized by inappropriate behavior, disinhibition , aggression and distractibility, our data proves that ALS and FTD are diseases of one continuum, with overlapping pathogenesis.

ALS- PLUS syndromes. Amyotrophic lateral sclerosis (ALS)–Plus syndromes is considered as ALS with additional features such extrapyramidal signs, autonomic dysfunction, cerebellar degeneration, those atypical clinical manifestations we categorized into groups including: extrapyramidal features (Tremor, masked face, startled appearance, bradykinesia, rigidity, dystonia, and/or retropulsion), cerebellar features (ataxia and/or dysmetria) and autonomic dysfunction (excessive sweating, GI disturbances , Urinary disturbances).

ALS-Plus syndrome was found in 20 (42.5%) patients , demonstrating extrapyramidal disorders- 6 (12.7%), autonomic functioning disturbances -10 (21.3.8%) and cerebellar dysfunction 4 (8.5%), ALS-Plus syndrome was not found in 27(57.5%) patients diagnosed with ALS. Symptoms dominating atypical clinical features of ALS were: GI disturbances, in particular constipation, resting tremor and excessive sweating. To investigate additional symptoms of ALS we used Mayo clinic Laboratory Neurology Patient form, where the administrator of the form should indicate the presence of symptoms.

Results of our study confirm the notion of ALS being further more than only a disease of motor neurons . It is safe to say, disease has pathological features of degenerative multisystem disorder with predominance of motor neuron involvement. Implying that ALS-plus symptoms should be screened vigorously by neurologists and managed appropriately.

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SUMMARY

CLINICAL CHARACTERISTICS OF ALS IN GEORGIAN PATIENTS

[1]Kekenadze M., [2]Kvirkvelia N., [1]Beridze M., [3]Vashadze Sh., [1]Kvaratskhelia E.

[1]Tbilisi State Medical University; [2]P.Sarajishvili Institute of Neurology; [3]Batumi Shota Rustaveli State University, Georgia

Amyotrophic Lateral sclerosis (ALS) is a fatal progressive neurodegenerative disease that affects the upper and lower motoneurons. .The disease is characterized by a plethora of neurological symptoms. There is a lot of information in the medical literature about ALS phenotypes, but the clinical diversity of ALS has not been studied in the Caucasus region and a unified clinical picture has not been conclusively established. In this regard, it is very important to study the symptoms among patients with ALS in Georgia. From 2018 to 2021, we examined 47 patients with ALS living in Georgia from different parts of the country, 23 - female, 24 - male, diagnosed based on clinical picture, electromyographic studies (AWAJI) and who met the EL ESCORIAL -Revised criteria. Also clinical symptom studies were conducted using the Mayo Clinic Laboratory Neurological Questionnaire. Cognitive changes were assessed using Adden-

brooke's Cognitive Examination scale (ACE III) and the Frontal Behavioral Questionnaire, the patient's quality of life was assessed by ALSFRS-R.

Patients were 26 to 84 years old, the age of onset of the disease was 58-60 years in men, 55-57 years in women. The bulbar type was observed in 21.3%, the upper limb type in 38.3% and the lower limb type in 40.4%. Frontotemporal dementia (FTD), diagnosed in 6 patients (12.7%). No reliable correlation was found between the forms of ALS and FTD.

The results of the study showed that ALS is a multisystem disease and is not limited to damage to motoneurons. It is safe to say that ALS has characteristics of polysystemic degeneration, with the predominance of motorneuron damage. Therefore, we consider it advisable to screen all patients with ALS for additional symptoms with a focus on the examination of cognitive function, which ensures the proper management of the disease in the future.

Keywords: amyotrophic lateral sclerosis, neurodegeneration.

РЕЗЮМЕ

КЛИНИЧЕСКИЕ ХАРАКТЕРИСТИКИ БОЛЬНЫХ БОКОВЫМ АМИОТРОФИЧЕСКИМ СКЛЕРОЗОМ В ГРУЗИИ

[1]Кекенадзе М.Д., [2]Квирквелия Н.Б., [1]Беридзе М.З., [3]Вашадзе Ш.Г., [1]Кварацхелия Э.Б.

[1]Тбилисский государственный медицинский университет; [2]Институт неврологии им. П. Сараджишвили; [3]Батумский государственный университет им. Шота Руставели, Грузия

Боковой амиотрофический склероз (БАС) - прогрессирующее нейродегенеративное заболевание со смертельным исходом, поражающее верхние и нижние мотонейроны. Заболевание характеризуется множеством неврологических симптомов. В медицинской литературе имеется достаточно информации о фенотипах БАС, однако в Кавказском регионе клиническое разнообразие БАС не изучалось и единая клиническая картина окончательно не установлена. В связи с этим весьма значимо изучить симптомы у пациентов с БАС в Грузии. С 2018 по 2021 гг. обследовано 47 пациентов с БАС, проживающих в Грузии из разных ее частей: 23 женщины, 24 мужчин, диагноз которых поставлен на основании клинической картины, электромиографических исследований и которые соответствовали пересмотренным критериям GOLD COAST. Проведены исследования клинических симптомов с использованием неврологического опросника лаборатории Mayo Clinic. Когнитивные изменения оценивались с использованием шкалы когнитивного обследования Адденбрука (ACE III) и фронтального поведенческого опросника, качество жизни пациентов оценивалось с помощью ALSFRS-R.

Возраст пациентов составил 26-84 лет, начало заболевания у мужчин 58-60 лет, у женщин 55-57 лет. Бульбарный тип наблюдался у 21,3%, тип верхней конечности - у 38,3%, тип нижней конечности - у 40,4%. Лобно-височная деменция (ЛВД) диагностирована у 6 (12,7%) пациентов. Достоверной корреляции между формами БАС и ЛВД не обнаружено.

Результаты исследования показали, что БАС является мультисистемным заболеванием и не ограничивается повреждением мотонейронов. Можно с уверенностью сказать, что БАС имеет характеристики полисистемной дегенерации с преобладанием повреждения мотонейронов. Авторы считают целесообразным обследовать всех пациентов с БАС на дополнительные симптомы с акцентом на исследование когнитивных функций, что обеспечит соответствующее лечение заболевания в будущем.

რეზიუმე

საქართველოში გვერდითი ამიოტროფული სკლეროზით დაავადებულ პაციენტთა კლინიკური მახასიათებლები

[1]მ.კეკენაძე, [2]ნ.კვირკველია, [1]მ.ბერიძე, [3]შ.ვაშაძე, [1]ე.კვარაცხელია

[1]თბილისის სახელმწიფო სამედიცინო უნივერსიტეტი; [2]პ.სარაჯიშვილის სახ. ნევროლოგიის ინსტიტუტი; [3]ბათუმის შოთა რუსთაველის სახელმწიფო უნივერსიტეტი, საქართველო

გვერდითი ამიოტროფული სკლეროზი (გას) არის ფატალური პროგრესული ნეიროდეგენერაციული დაავადება, რომელიც აზიანებს ზედა და ქვედა მოტონეირონებს. დაავადება გამოირჩევა მრავალფენოვანი ნევროლოგიური სიმპტომატიკით. სამედიცინო ლიტერატურაში არსებობს საკმაო ინფორმაცია გას-ის ფენოტიპების შესახებ, მაგრამ გას-ის კლინიკური მრავალფეროვნება არ არის შესწავლილი კავკასიის რეგიონში და არ არის საბოლოოდ ჩამოყალიბებული ერთიანი კლინიკური სურათი. ამ მხრივ მეტად აქტუალურია საქართველოში გას-ით დაავადებულ პაციენტებში სიმპტომების შესწავლა. 2018 წლიდან 2021 წლამდე გამოკვლეულია საქართველოში მცხოვრები გას-ით დაავადებული 47 პაციენტი ქვეყნის სხვადასხვა კუთხიდან: 23 ქალი და 24 მამაკაცი; დიაგნოზი ემყარებოდა კლინიკურ, ელექტრომიოგრაფიულ გამოკვლევებს და აკმაყოფილებდა GOLD COAST კრიტერიუმებს. ასევე კლინიკური სიმპტომების შესწავლა ჩატარდა მაიოს კლინიკის ლაბორატორიის ნევროლოგიური კითხვარის გამოყენებით. კოგნიტიური ცვლილებები შეფასდა ადენბრუკის კოგნიტიური სკალით (ACE III) და ფრონტალური ქცევითი კითხვარით, პაციენტების ცხოვრების ხარისხი - ALSFRS-R-ით.

პაციენტები იყვნენ 26-დან 84 წლამდე, დაავადების დაწყების ასაკი მამაკაცებში იყო 58-60 წელი, ქალებში - 55-57 წელი. ბულბური ფორმა დაფიქსირდა 21.3%-ში, ზედა კიდურის ფორმა - 38.3%-ში და ქვედა კიდურის ფორმა - 40.4%-ში. ფრონტოტემპორალური დემენცია (ფტდ) დიაგნოზირებულია 6 (12,7%) პაციენტში. ფტდ-სა და გას-ის ფორმებს შორის სარწმუნო კორელაცია არ გამოვლინდა.

კვლევის შედეგებმა აჩვენა, რომ გას მულტისისტემური დაავადებაა და არ შემოიფარგლება მხოლოდ მოტონეირონების დაზიანებით. თამამად შეიძლება ითქვას, რომ გას-ს აქვს პოლისისტემური დეგენერაციისათვის დამახასიათებელი თავისებურებები, მოტონეირონების დაზიანების უპირატესობით. აქედან გამომდინარე, მიზანშეწონილად მიგვაჩნია, რომ ყველა გას-ით დაავადებულ პაციენტს ჩაუტარდეს გას-ის დამატებითი სიმპტომების სკრინინგი, აქცენტით კოგნიტიური ფუნქციის გამოკვლევაზე, რაც განაპირობებს შემდგომში დაავადების სწორ მენეჯმენტს.