What is the (facio) scapuloperoneal muscular dystrophy? Is it an independent form or a variant of FSHD? Short extracts from the works of some authors given below specify their position concerning nosological place of (facio) scapuloperoneal muscular dystrophy.

Oransky [1] considered that it is a peculiar autosomal dominant form of muscular dystrophy with predominantly early involvement of the shoulder – girdle and peroneal group muscles and in some cases with a mild weakness of the face and trunk muscles.

Davidenkov and Kulkova [2] “Scapulo-peroneal amyotrophy is probably an independent disease which significantly closer to the Landouzy-Dejerine amyotrophy” and later… “It is possible to suppose that actually scapula-peroneal amyotrophy and Landouzy-Dejerine amyotrophy are connected with the action of the same basic gene but different phenotypes may be due to a modified action of the changing familal gene pool.”

Seitz [3] related scapuloperoneal myogenic syndrome to Erb’s Dystrophy musculorum progressiva.

Davidenkov and Dogel [4] “…Scapuloperoneal amyotrophy is a well outlined, peculiar form which obviously belongs to a combined group of myopathies and approaches mostly to the amyotrophy of Landouzy-Dejerine, although it has some features which differ substantially both from the classical amyotrophy of Landouzy-Dejerine and from the other variants of myopathy.”

Becker [5] “Autosomal dominant myogenic scapulo-peroneal syndrome probably is an independent form of muscular dystrophy.”

Ricker and Mertens [6] “The myogenic autosomal dominant scapulo-peroneal syndrome is a variant of FSHD.”

Zellweger and McCormick [7] “Should the familial disorder reported by Oransky be considered as scapuloperoneal dystrophy, one would need to postulate a new type of autosomal dominant muscular dystrophy which would be classified between the scapulohumeral variant (Erb) of type I (Landouzy-Dejerine) and autosomal dominant, distal type of Velander.”

Serratrice et al. [8] wrote that a scapula-peroneal muscular atrophy “It is probably an unusual form of progressive muscle atrophy of facio-scapulo-humeral type with peroneal involvement – or myopathies (facio) scapuloperoneal”

Hausmanova-Petrušević [9] “Scapuloperoneal syndrome (its myopathic variant) appears to be a variety of the FSHD.”

Kazakov [10], Kazakov et al. [11,12] “…the FSHD type can be considered as an independent form. The best name for it is “facio-scapulo-limb (FSL) muscular dystrophy adding the eponym “Duchenne-Landouzy-Dejerine”. FSL myodystrophy is clinically heterogeneous and two varieties in the form of muscular dystrophy included: the gradually descending variety and the more frequently a descending one, characterized by a “jump” from the upper part of the body to the peroneal groups of the shin muscles (the descending variety with a “jump”). Genealogical analysis of 62 families suggests the existence of a clinical and genetic heterogeneity in FSL myodystrophy”.

Thomas et al. [13] “The clinical features of the present series (a scapuloperoneal myopathy), however, clearly indicate that they represent a distinct group.”

Carroll [14] “…Although the preceding paragraphs form the basis of the name FSHD, other muscles are often involved… Also sometimes what it noted early in the disease is weakness of the anterior tibial muscles Walton and Gardner-Medwin [15,16] In our experience, this has been a frequent occurrence. … Thus, if a patient were encountered with scapular muscle and anterior tibial weakness along with very little facial weakness, then the term scapuloperoneal dystrophy would just as well apply.”

(Serratrice et al. [17]; Kazakov et al. [11]) “Autosomal dominant scapuloperoneal myopathy,… The possibility that some recorded cases have “incomplete” or abortive forms of facioscapulohumeral muscular dystrophy is suggested by the large kindred reported by Kazakov et al. in which facial muscles was a late and mild feature in many cases.”

Serratrice et al. [17] “…1 scapula-peroneal dystrophies of a predominantly hereditary nature (31 cases) The most frequently observed form (very probably of a dystrophic nature) appear to be only a topographical variety of Landouzy-Dejerine’s facio-scapulo-humeral myopathy, particularly as the facial region is often involved. The following forms…: pure facio-scapulo-humeral, facio-scapulo-humeral with peroneal involvement, facio-scapulo-peroneal, and pure scapula-peroneal.”

Padberg [18] “…Ricker and Mertens did one year earlier (1968) namely, that a scapuloperoneal syndrome might be a stage in the development of FSHD. A similar conclusion was drawn by Kazakov et al. [11] about the famous family K. This family was reported initially by Oransky in 1927, …Kazakov et al. [11] reexamined this family… In his opinion this family suffered from FSHD” and late “…autosomal dominant SP myopathy with facial weakness in indistinct from FSHD” (My note: this citation is not exact. I wrote that in the famous kindred K. there is a FSLD descending with a “jump” type, but not a classical FSHD (or FSLD gradually descending type).

Rossi et al. [19] “We agree with Kazakov et al. [11] that involvement of the peroneal muscles could be a stage in the development of Landouzy-Dejerine dystrophy rather than characterizing a separate entity as Becker proposed [5]”. (My note: this citation is not exact).

Brooke [20] “Scapuloperoneal dystrophy is probably a variety of facioscapulohumeral dystrophy…. Bilateral foot-drop may be present (on FSHD). Occasionally this is one of the initial symptoms and it is then difficult to know whether to call the disorder facioscapulohumeral dystrophy or scapuloperoneal dystrophy.”

Mcusick [21] “Kazakov et al. provided a follow-up the kindred reported by Davidenkow (1939). The disorder in many ways resembled Landouzy-Dejerine facioscapulohumeral muscular dystrophy.”

*Corresponding author: Valery Kazakov, Department of Neurology, Pavlov State Medical University, L. Tolstoy Str. 6/8, 197022 St. Petersburg, Russia, E-mail: valerykazakov@mail.ru

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Swash and Schwartz [22] "The extensor digitorum brevis muscles may be spared both in FSHD and in scapulo-peroneal muscular dystrophy and the two disorders are probably closed related, if they are not variations of the same disorder."

In classification compiled by [23] the scapuloperoneal muscular dystrophy was presented as nosological entity: «c) Scapuloperoneal muscular dystrophy, autosomal dominant». Munsat and Serratrice [24] "The scapuloperoneal dystrophies of autosomal dominant inheritance or sporadic… suggested a relationship between FSHD and scapuloperoneal syndrome. There appears to be a continuum with 4 clinical forms: pure FSHD; FSHD with peroneal weakness; scapuloperoneal dystrophy with facial weakness (in these cases, humeral muscles are frequently spared and ankle contractures are constant) and pure scapuloperoneal dystrophy."

Brouwer [25] "For instance, Oransky [1] described three families with autosomal dominant scapuloperoneal syndrome. … Kazakov et al. reexamined the Oransky [1] family and finally concluded that this family suffered from FSHD". ([My note: this citation is not exact]).

Jardine et al. [26] "The existence of scapuloperoneal myopathy, without contractures or cardiomyopathy, as a genetically separate condition from FSHD is uncertain…. Scapular onset muscular dystrophy without facial involvement inherited as an autosomal dominant condition, may be allelic with FSHD."

In classification compiled by Walton et al. [27] the scapuloperoneal muscular dystrophy was presented as nosological entity: «c) Scapuloperoneal muscular dystrophy, autosomal dominant (not proven to differ from facioscapulohumeral muscular dystrophy; note possible neurogenic type)"

Kazakov [28] "… the FSHD is a heterogeneous form of muscular dystrophy. The name facio-scapulo-limb muscular dystrophy (FSLD) was offered for this disease. On the historical and clinical study FSLD may be divided into two nosological entities, namely: Facioscapulolimb muscular dystrophy, type 1 (FSLD1), a gradually descending type with initial facio-scapulo-humerum phenotype; autosomal dominant (Duchenne de Boulogne) and Facioscapulolimb muscular dystrophy, type 2 (FSLD2), a descending with a "jump" with initial FSP phenotype; autosomal dominant (Erb, Landouzy, Dejerine)."

Griggs et al. [29] "Kazakov et al. personally examined patients belonging to four generations of the family originally reported by Oransky …and the clinical picture was indistinguishable from FSHD dystrophy". ([My note: this citation is not exact]).

Tawil et al. [30] "Consequently, some authors suggest that a least the myopathic form of FSPs is a stage in the development of FSHD" (Kazakov et al.). ([My note: this citation is not exact]).

Rowland [31] "Locus heterogeneity… helped to explain many diseases …such as the separation of scapuloperoneal muscular dystrophy from facioscapulohumeral dystrophy."

Wilhelmson et al. [32] "If there were facial weakness in a person with autosomal dominant scapuloperoneal dystrophy, it would be indistinguishable from FSHMD." (My note: this citation is not exact). Fisher and Upadhyaya [33] "Muscle disorders related to FSHD include scapuloperoneal and scapulohumeral dystrophies which are associated with minimal or absent facial weakness and may represent milder forms of the disease since there is evidence for involvement of the same genetic locus."

Bushby [34] "within the broad definition of FSHD there can be extreme variation… In some patients, muscle involvement remains restricted to the facial, scapular, and proximal upper limb musculature. In others, anterior tibial and peroneal muscles may be involved subsequently, leading to foot-drop as a relatively early sign. Alternatively, the proximal muscles of the lower limb may be involved before the distal muscles …"

Padberg [35] "Subsequent nosography introduced the term scapuloperoneal [4] which is also the most concise summary of the early picture of FSHD patients with minimal facial weakness [1]. It took some time to recognize that most of the families reported to suffer from scapuloperoneal myopathy had FSHD [1]". ([My note: this citation is not exact]).

Orrell and Griggs [36] "The status of scapuloperoneal syndrome is not fully defined. Clinical features overlap with FSHD… The availability of molecular diagnostic testing of FSHD has led to the reclassification of some patients previously diagnosed with scapuloperoneal syndrome as having FSHD."

Attarian et al. [37] "We propose that SP represents the mild end of the spectrum of FSHD with DFS ranged between 28 to 35 kb."

The Neuromuscular Home Page (Washington University) (2003, 2012) [38] suppose “The scapuloperoneal weakness (scapular winging, foot dorsiflexion weakness and abdominal muscle weakness) observe on mild or atypical FSHMD1A, chromosome 4q35, dominant with small deletion >35 kb DNA fragment".

Padberg [39]"However, it is likely that the adjective FSH later precluded recognition of the disease in some families with lower limb involvement [1,13] while mild facial weakness in other families prompted the use of the term scapuloperoneal muscular dystrophy [2] My note: this citation is not exact (see below)). In any case, a discussion was necessary to get early foot-extensor weakness accepted as part of the natural course of FSHD [18]."

In other articles [Zeevaert et al. [40], Pou-Serradell [41]] was written again about the wide clinical spectrum of 4q35 linked FSHD and the difficulty to diagnose unusual familial cases with facial-sparing and scapuloperoneal muscle afferent related to 4q35 chromosomal deletion.

The question is: What is the autosomal dominant scapuloperoneal muscular dystrophy with minimal affection of facial muscles linked with 4q35 chromosomal deletion?

In the article published in Eur. Neurol [42] which by Padberg [18], Brouwer [25], Griggs et al. [29], Tawil et al. [30] and Wilhelmson et al. [32] did not cited correctly enough (see above), I wrote in conclusion :"The study of this form of muscular dystrophy (facio-scapulo-limb or facioscapulohumeral) carried out by us, revealed the existence of two independent varieties: a gradually descending variety and more frequent a descending one with a "jump" of the atrophies from facial and shoulder girdle muscles to the peroneal group of the shin muscles (the descending variety with a "jump"). To our mind, it is to the latter variety that the muscular dystrophy in the K. kindred should be related (p. 358)".

In other article published in Clinical Genetics [43] which by Rossi et al. [19], Walton and Gardner-Medwin [15] and McKusick [21] did not total cited correctly enough (see above), I wrote: "From the clinical and genetic data available at the present time, is seems that the muscular dystrophy in the K. kindred is one of the varieties (namely, a descending type with a "jump") of the facio-scapulo-limb (or facioscapulohumeral) muscular dystrophy (p.41)".
Our present clinical and MRI data, as well as our earlier investigations (1969-2009), allows suggesting that FSP muscular dystrophy is probably an independent form with “hard” (stereotypical) static and dynamic pattern of muscle involvement and a mild course of the disease. The term “FSLD2 descending with a “jump” with initial FSP phenotype (Erb, Landouzy and Dejerine type)" would be more correct. The FSP or (F) SP phenotype constitutes merely a stage in the development of FSLD2.

I suppose that classical AD FSPMD (or FSLD2, a descending with a “jump” with initial FSP phenotype) is different (accoring specific static and dynamic pattern of muscle involvement as well as a mild degree affection of mimic muscles and mild course of the disease) from the classical AD FSHD which was called by me as a FSLD1, a gradually descending with initial FSH phenotype (Duchenne de Boulogne type) [28] and may be these diseases are connected with the various 4q35 chromosomal mutations [28,42-68].

I would like to present the photos of two different patients who are both called in the World literature as a classical and typical FSHD [48,49].

This is my proband K.N. aged 63 (Figure 2) from famous family K. with typical FSLD2 descending with a “jump” type [42]. At the age of 18.5 he had a pure mild scapuloperoneal phenotype [1] which at the age of 30 (Figure 1) transferred in (facio)-scapulo-peroneal-(humeral) phenotype [2], and then at the age of 63 (Figure 2) it transferred in final severe (facio)-scapulo-peroneal-humero (biceps brachii, but not triceps) -femoro (posterior thigh muscles, but not quadriceps) –gluteal (gluteus maximus muscles, but not gluteus medius) phenotype with increased the lumbar lordosis due to the severe weakness of abdominal and gluteus maximus muscles and slight atrophy of the right half of upper lip. At the age of 68 he could walk independently and climb the stairs (on 6 floors) with the aid of a stick and raling and he could walk with the help of a stick on long distances. In his a first cousin once removed aged 39 and a second cousin once removed aged 16 the 4q35 DFS 33/30 kb were found, and in other a first cousin once removed aged 34 the 4q35 DFS was 24/21 (double digestion). They all were presymptomatics.

The Figure 3 was taken from Wikipedia, the Free Encyclopedia, 4 November 2011 (en.wikipedia.org). In my opinion this 27-year-old female have a typical FSLD1 gradually descending type with initial FSH phenotype with gradually extension of weakness and atrophy from the face and shoulder girdle to the upper arm, trunk, pelvic girdle and thigh muscles. She has increased lumbar lordosis due to the severe weakness of erector trunci muscle predominantly together with abdominal and pelvic girdle muscles.

Conclusion

It is necessary to remark that in the present time there is an opinion that FSHD is genetically heterogeneous, but clinically a homogeneous disease. "In recent years …allowed the identification of the two forms of FSHD, the classical autosomal dominant FSHD type 1, and FSHD type 2 characterized by identical clinical phenotype but associated with a different (epi) genetic defect" and after “… FSHD1 and FSHD2 share a common pathophysiological pathway since they present an identical clinical phenotype and FSHD2 genetic defect, and that the FSHD2 gene may act as a modified disease in FSHD1 families” [69].

On my opinion FSHD1 on clinical picture and molecular genetics may be corresponded to FSLD2 (a descending variety with a "jump") due to deletion of D4Z4 on chromosome 4q35. This form was described at the first time by Erb, Landouzy and Dejerine under different names in 1882-1885 years [28].

Another form of FSHD which was called as a FSHD2 due to hypomethylation of D4Z4 on chromosome 4 and 10 [69,70] and may be corresponded to FSLD1 (a gradually descending type) which was described at the first time by Duchenne in 1855 under the name "progressive hereditary muscular atrophy with fatty degeneration beginning with a face, trunk and limbs in adults or in youth or in children" [28]. That is why with historical point of view the FSHD1 more correctly to name as a FSHD2 but a FSHD2 – as a FSHD1. However, with molecular genetics point of view (if to accept in attention the time of molecular genetics description of these forms) the names FSHD1 and FSHD2 are correct.

In 1969-1971, I analyzed the pattern of muscle involvement (67 isolated muscles and their parts) in different stages of the disease of 200 patients with FSLD (or FSHD): 145 cases were taken from the
World literature and 55 cases were under personal observation [10,11]. Between these 200 patients of 78 (31 personal cases and 47 from literature, 59 hereditary and 19 sporadic) had developed FSLD2 a descending with a jump type with the FSP or FSP (H) phenotypes (38 patients) and with the final FSPFGH (facio-scapulo-peroneal-femoro (posterior thigh muscles)-gluteo (gluteus maximus)-humeral (biceps brachii) or the FSPHFG phenotypes (40 patients).

Besides this, between 200 analyzed FSLD (FSHD) cases of 60 (47 hereditary and 11 sporadic cases included Duchenne's cases, hereditary 8 and sporadic 3 taken from the World literature and only 2 my personal sporadic cases) had developed FSLD1 a gradually descending type [10,11]. Between these 60 cases the 31 were with the FSHGF (facio-scapulo-humero-gluto-femoral) phenotypes and the 29 cases were with the final FSHGFP (facio-scapulo-humero-gluteo-femoro-peroneal) phenotypes. In last group the pelvic girdle and thigh muscles were more severe affection then peroneal group excluded 6 cases in which the peroneal group muscles were the same severe affection [71-75].

It was found that only one of the two type of the disease (the gradually descending type or the descending one with a jump) occurred in each family. The distribution of the muscle affections in the members of the same family did not usually overlap the limits of the type of the development of the disease.

As well the existence of FSLD1 are confirm the fact that in some Handbooks on Muscle Diseases or Neurology the FSHD was described as a gradually descending muscular dystrophy with affection of pelvic girdle, trunk and hip muscles after involvement of the face, scapular and humeral muscles (and others).

Thus, in my opinion the FSLD1 and FSLD2 are clinical and historically well documented i.e. FSLD (or FSHD) is genetically as well as clinically a heterogeneous disease [10-12,28,42-46,49-71].

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