Facioscapulohumeral disease

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LIST OF ABBREVIATIONS

ATP-ase	adenosine triphosphatase
A-V	atrio-ventricular
BSAPP	brief small abundant polyphasic potentials
CK	creatine kinase
ECG	electrocardiogram
EMG	electromyogram
FSH	facioscapulohumeral
FSHD	facioscapulohumeral disease
FSHS	facioscapulohumeral syndrome
HE	haematoxilin and eosin
LD	lactic dehydrogenase
MRC	medical research council
NADH-TR	NADH-tetrazolium reductase
PAP	persistent atrial paralysis
PK	pyruvate kinase
PMA	peroneal muscular atrophy
SGOT	serum glutamic oxaloacetic transaminase
SGPT	serum glutamic pyruvic transaminase
SMA	spinal muscular atrophy
SP	scapuloperoneal
SPD	scapuloperoneal muscular dystrophy

Introduction

The purpose of this study is to discuss several aspects of facioscapulohumeral disease, also called "autosomal dominant facioscapulohumeral muscular dystrophy" or "Landouzy-Dejerine type of muscular dystrophy" or "Landouzy-Dejerine's disease". We consider this disorder well defined and recognizable, justifying the term facioscapulohumeral disease, abbreviated FSHD.

We studied the literature, as well as a personal series of 107 cases of FSHD. Chapter 1 reviews the major historical reports pertinent to the recognition of FSHD as an independent entity. Chapter 2 describes current knowledge on FSHD: a summary is presented at the end of this chapter. Chapter 3 discusses the differential diagnosis of FSHD. A great deal has been written on this subject, most of it causing more confusion than clarification. As it was considered necessary to argue why some reports were so obfuscating, this chapter has become quite lengthy: for those who feel that this subject should not claim so much attention, a summary is proffered. Chapter 4 deals with the results of the clinical examination of 107 patients with FSHD and Chapter 5 with the laboratory studies in some of these patients. The kindreds were ascertained through probands that had been studied at the "Muscular Research Center" (head Prof. Dr. J. Bethlem) of the University of Amsterdam and at the Neuromuscular Clinic (head Dr. A.R. Wintzen) of the Department of Neurology (chairman Prof. Dr. G.W. Bruyn) of the University of Leiden. The kindreds were examined as extensively as possible. In three instances family examination was incomplete or did not reveal autosomal dominant inheritance. These cases are discussed briefly in Chapter 6. In the last chapter some of our results are compared with concepts dominating in the literature.

In order to provide a framework for the discussion of this autosomal dominant disorder, we prefer to summarize FSHD as follows: the presenting complaints are mostly those of weakness of the shoulder girdle muscles. The clinical signs at the time of

presentation include weakness and atrophy of the shoulder girdle muscles with early involvement of the facial muscles in most cases. The disease subsequently spreads to the upper arm muscles, justifying the adjective facioscapulohumeral (Landouzy and Dejerine, 1884, 1885, 1886) and to the peroneal muscles. Weakness of the abdominal muscles may occur early. Pelvic girdle weakness is a fairly late sign in most cases. The muscle involvement is often asymmetrical. The intrafamilial and interfamilial expression of the disease is quite variable. The clinical course and rate of progression of the disease may also vary considerably from case to case. A large number of affected individuals may be asymptomatic (abortive cases). The age of onset may range from infancy to late adulthood. The penetrance of the gene is almost complete. There are no solid grounds to assume to existence of an autosomal recessive disorder resembling FSHD. The problem of the isolated case in which the examination of the family is negative, has no simple answer: there can be low expressivity of the gene in the ancestry, non-paternity, a mutation, or a different disease altogether. Although FSHD is considered a myopathy, both electromyography (EMG) and muscle biopsy may reveal features suggesting a neurogenic lesion.