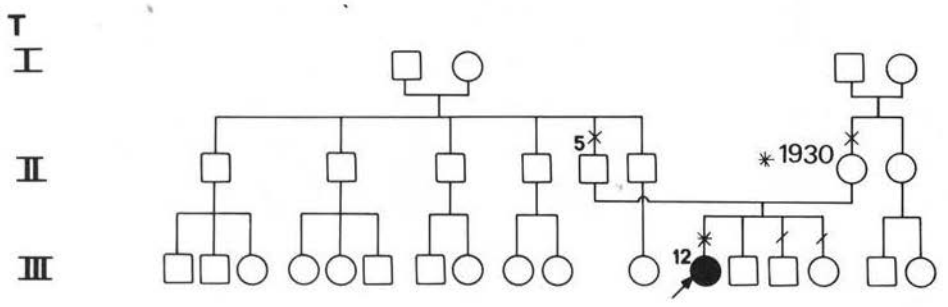
Chapter 6

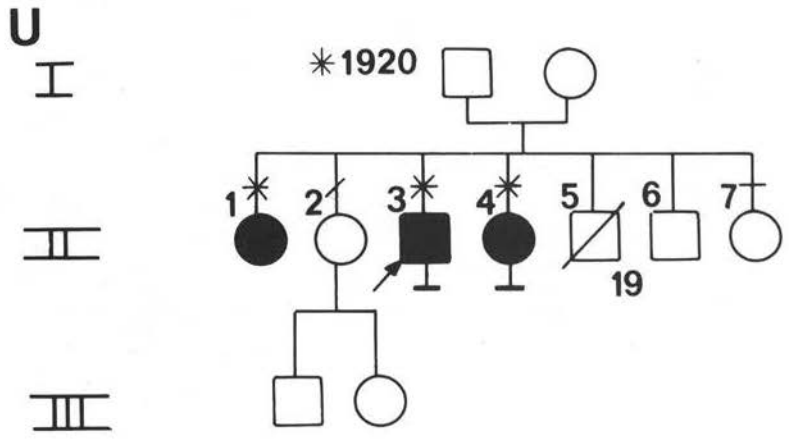
Cases resembling facioscapulohumeral disease

It has been argued that establishing an autosomal dominant pattern of inheritance is essential for the diagnosis of FSHD. So far, no solid proof has been put forward for an autosomal recessive disorder closely resembling FSHD. Sporadic cases have been observed but it is impossible to prove that they are not new mutants of FSHD. The opposite is equally difficult to ascertain at the time of first examination. Genetic advice should be given with caution in these cases. We observed several patients who were proof to their parents of the autosomal dominant nature of the disorder. In several families (kindreds G, K, R and S) the autosomal dominant pattern of inheritance had not been established before our study, nor was it suggested by the family history.

While studying the families of the probands diagnosed on the basis of clinical criteria to suffer from FSHD, in three families we were unable to establish an autosomal dominant pattern. In two families adequate family data were lacking. Extensive reporting of these cases is not useful; they will be mentioned briefly below.



In one family (kindred T) both parents were examined in detail: they demonstrated no abnormalities. Paternity investigation was not undertaken. A brother (T III 14) and a sister (T III 15) of the proband were examined briefly; neither of these showed signs of muscle weakness or atrophy. Genealogical examination, going back 180 years, did not demonstrate or even suggest a relationship with the FSHD families reported in this study. The mother is sure that facial weakness was present in her daughter since the age of nine months. At the age of 12, the patient (T III 12) noticed difficulties writing on a blackboard. Her shoulder girdle weakness steadily progressed. When examined at the age of 24, she had a marked facial weakness and she was just able to swing her arms up on a shelf above shoulder level. She also had a mild paresis of the abdominal muscles, foot extensor weakness and asymptomatic ankle contractures. Fasciculations and myotonia were not observed. Sensory examination was normal at the age of 15. EMG and biopsy of the quadriceps femoris muscle had revealed no abnormalities. Serum CK activity at that time was raised twice the upper limit of normal. A definite diagnosis could not be made in this patient. Facial onset and the clinical picture, as it subsequently developed, is quite compatible with FSHD.



In another family (kindred U), three sibs were affected. The parents and U II 6 had no complaints; they were not examined as they lived abroad. U II 1 had facial weakness before the age of five. Running became difficult at elementary school. Climbing stairs was possible until 21 years. She fractured her right tibia at the age of 32: after 11 weeks of immobilisation she was unable to walk. At the time of examination (34 years old) she had a marked facial paresis with good strength of the masseter muscles and no atrophy or fasciculations of the tongue. There was an asymmetric paresis of the shoulder and arm muscles, proximally more marked than distally. The legs were severely paretic with the exception of the gastrocnemius muscles. Sensory examination was normal. All myotatic reflexes were absent, with the exception of the ankle jerks; no pathological reflexes could be elicited.

Her brother (U II 3) developed a similar clinical picture, starting with facial weakness at the age of five. Progressive shoulder girdle weakness and footextensor weakness was noted since the age of eight. U II 4 was reported to have gait disturbances at the age of three. She remembered having needed speech therapy at the age of four, together with her brother. She could walk until the age of 15 and finally developed a picture very similar to that of her sister. In both U II 1 and U II 3, EMG and muscle biopsy revealed myopathic changes. Two other sibs (U II 2 and U II 7) were examined and demonstrated no abnormalities. A brother (U II 5), who was a marine, had died in an accident. Genealogical examination did not suggest any relationship with the FSHD families reported in this study.

The clinical picture in these three patients is similar to FSHD. The early onset of facial weakness and the rapidly progressive course would fit Brooke's description of the infantile form of FSHD. It is frequently observed in those instances that one of the parents has slight facial weakness; and, as facial weakness may go unnoticed to the patient, FSHD is still a possible diagnosis in this family.

In another sporadic case, family examination revealed the patient to be an illegitimate child. Nothing was known about the natural father, who could not be examined. Physical examination

of the mother revealed no abnormalities.

All the cases discussed above demonstrate once more, that examination of the families is crucial in all instances where autosomal dominant inheritance is not apparent. In the true sporadic case in this material (T III 12) the myopathic nature of the disorder has not been documented sufficiently.