The Inheritance of Progressive Muscular Dystrophy in Japan

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Received January 18, 1968

Summary. In conclusion, there were no apparent differences between Japanese and foreign cases in clinical types, the frequency of each type, sex ratio, the age of onset, modes of the inheritance etc., except for a peculiar type of the disorder found in the South-Western Islands. However, sporadic cases were seem to be comparatively often encountered in each type especially in the Duchenne and limb-girdle type. Further, large families with as many victims as in foreign cases have not yet been reported in Japan.

I. Introduction

The inheritance of progressive muscular dystrophy is still under discussion. Here, I don't want to state the history of the above discussion in detail on the relationship between phenotypes and various modes of inheritance of the disease. Before 1940, almost all studies on both clinical and genetic aspects of this disorder had been done by German medical scientists except for some noteworthy studies done in other countries. After 1950, some investigators in the U.S.A. and Britain have been studying on this subject. In Germany, Becker published a monograph based on his extensive studies in 1953. He classified the disorder in 1964 as follows:

1. Shoulder-girdle type with dominant inheritance.
2. Pelvic-girdle type with X-chromosomal recessive type (Duchenne).
4. Pelvic-girdle type with autosomal recessive inheritance.
5. Pelvic-girdle type combined with other diseases.
6. Distal type.
7. Ocular myopathy.

Recently, Walton (1966) proposed the following classifications comparing those made by many investigators after 1950:

1. The Duchenne type muscular dystrophy. Sex-linked recessive variety.  
   a) Severe, b) Less severe of later onset.
4. Distal type.
5. Ocular myopathy.

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Between these two representative classifications, in general, we realized no fundamental contradictions.

The first systematic study discussing the relationship between clinical characters and inheritance was done by Niwa (1929). He described three modes of inheritance: autosomal dominant, autosomal recessive, and sex-linked recessive inheritance. Further, he recognized parallelism between the modes of inheritance and clinical types up to a certain degree. Thereafter, Murakami started a clinico-genetic study on this disorder in 1938 and published his results in 1957. At quite the same time, Miyoshi commenced a similar study. In 1958, Hanada published a paper on the genetics of this disorder on 55 patients of 41 families. Thereafter, Miyazaki (1963) also published a similar paper based on 187 cases of 141 families. There seem to be overlaps in cases between these two investigators and the major source of the cases were Kyushu (Fig. 5).

From 1964 to 1966, a scientific research group for the study of the nature and cause of myopathies was organized in Japan. This group had 16 members and the followings was the major subjects of cooperation:

1. Registration of victims with this disorder.
2. Examination of degrees of involvements.
3. Study on carriers.
4. Study on the therapy.
5. Measurement of the serum creatine phosphokinase (CPK) value, with special reference to the comparison of the results by Ebashi-Sugita's method with that of foreign works.

Parallel to the above cooperative subjects, genetic studies were done by Murakami, Miyoshi, Kondo, and Tsubaki, and Fukuyama. Here I will mention simply facts seen in my own materials; Studies carried out by other investigators such as Miyoshi, Fukuyama, Kondo, and Tsubaki, etc. will be supplementarily treated. In the above research group, classification of this disorder was chiefly influenced by Walton and in some parts by Becker. It was as follows:

1. Duchenne type. a) Sex-linked type of early onset. b) Benign type of later onset.
2. Limb-girdle type.
3. Facioscapulohumeral type.
4. Distal type.
5. Ocular myopathy.

II. Materials

1. Materials collected by Murakami from 1938 to 1957 (1st series of examinations): From 1938 to 1957, 94 cases from 59 families were studied by Murakami. Many of these families were located in the neighbouring areas of Aichi Prefecture, which is located on the Pacific coast in the middle part of the mainland of Japan (Fig. 4). Murakami visited all families and surviving victims. Further, as many family members as possible who were said to be healthy were examined so as not to miss minimally affected cases and associated anomalies. Pedigrees were prepared based on records of the National Family Registers. In Japan, the National Family Registration system was established in 1871 and everybody has an obligation to register his or her birth within 2 weeks. Further, in these families, Murakami examined all minor anomalies, not only in victims but also those found in healthy relatives. The result was