



Facioscapulohumeral Muscular Dystrophy: A Guide to Molecular Diagnosis and Genetic Counseling

Najib Kissani

Department of Biological Sciences, Federal University of Goiás, Brazil

Introduction

Facioscapulohumeral muscular dystrophy (FSHD) is a rare, autosomal dominant, degenerative muscle disease. It is characterized by progressive weakness and wasting of the muscles of the face, neck, and upper arms. The disease is caused by a deletion of a portion of the 4q35 region of chromosome 4, which contains the D4Z4 repeat. The number of D4Z4 repeats is inversely related to the severity of the disease. In affected individuals, the number of D4Z4 repeats is typically less than 10, while in unaffected individuals, it is typically greater than 10. The D4Z4 repeat is located on the short arm of chromosome 4, at the 4q35 locus. The D4Z4 repeat is a type of variable number of tandem repeats (VNTR). The D4Z4 repeat is present in all human populations, but its number varies between individuals. The D4Z4 repeat is located on the short arm of chromosome 4, at the 4q35 locus. The D4Z4 repeat is a type of variable number of tandem repeats (VNTR). The D4Z4 repeat is present in all human populations, but its number varies between individuals.

Genetics of FSHD

FSHD is caused by a deletion of a portion of the 4q35 region of chromosome 4, which contains the D4Z4 repeat. The number of D4Z4 repeats is inversely related to the severity of the disease. In affected individuals, the number of D4Z4 repeats is typically less than 10, while in unaffected individuals, it is typically greater than 10. The D4Z4 repeat is located on the short arm of chromosome 4, at the 4q35 locus. The D4Z4 repeat is a type of variable number of tandem repeats (VNTR). The D4Z4 repeat is present in all human populations, but its number varies between individuals.

FSHD1: Clinical Features and Diagnosis

FSHD1 is the most common form of FSHD. It is characterized by progressive weakness and wasting of the muscles of the face, neck, and upper arms. The disease is caused by a deletion of a portion of the 4q35 region of chromosome 4, which contains the D4Z4 repeat. The number of D4Z4 repeats is inversely related to the severity of the disease. In affected individuals, the number of D4Z4 repeats is typically less than 10, while in unaffected individuals, it is typically greater than 10. The D4Z4 repeat is located on the short arm of chromosome 4, at the 4q35 locus. The D4Z4 repeat is a type of variable number of tandem repeats (VNTR). The D4Z4 repeat is present in all human populations, but its number varies between individuals.

