



+ Affected chromosome 4
 - Normal chromosome 4

What are sporadic cases of FSHD?

Sporadic FSHD cases are those resulting from a new mutation. Studies report from 10% to as high as 33% of FSHD cases as sporadic (de novo mutation). Approximately 20% of reported sporadic cases are those inherited from a seemingly unaffected parent who is a “germline mosaic” meaning that only the mother’s or father’s germ cells (the egg or sperm) is affected. When a germline mosaic is involved, the parent appears unaffected but the children are at risk.

In the remaining 80% of sporadic cases, a new spontaneous mutation results in a chromosome 4 deletion that causes FSHD. When the 4q35 deletion fragment appears in a sporadic FSHD case, it is transmitted in an autosomal dominant (only one parent needs to be affected) manner to succeeding generations. The probability, then, of passing the disease to an offspring is 50%.

How many people have FSHD?

It is difficult to calculate the exact incidence of FSHD. It may be under reported, but an accepted estimate of its occurrence in the general population is one in 20,000. A recent publication reports FSHD as the most prevalent dystrophy: one in 14,286. FSHD occurs in all racial groups and with equal frequency in both sexes.

