

# ACTN3

**Alpha-actinin-3**, also known as **alpha-actinin skeletal muscle isoform 3** or **F-actin cross-linking protein**, is a **protein** that in humans is encoded by the *ACTN3* gene.<sup>[1][2]</sup>

Alpha-actinin is an **actin-binding** protein with multiple roles in different cell types. This gene expression is limited to skeletal muscle. It is localized to the **Z-disc** and analogous dense bodies, where it helps to anchor the myofibrillar actin filaments.<sup>[1]</sup>

## 1 Fast versus slow twitch muscle fibers

Skeletal muscle is composed of long cylindrical cells called muscle fibers. There are two types of muscle fibers, slow twitch or muscle contraction (type I) and fast twitch (type II). Slow twitch fibers are more efficient in using oxygen to generate energy, while fast twitch fibers are less efficient. However, fast twitch fibers fire more rapidly and generate more force. Fast twitch fibers and slow twitch fibers are also called white muscle fibers and red muscles fibers, respectively.

## 2 ACTN3 in muscle fiber

Each muscle fiber is composed of long tubes called myofibrils which in turn are composed of filaments. There are two types of filaments: **actin** (thin filaments) and **myosin** (thick filaments) which are arranged in parallel. A muscle contraction involves these filaments sliding past each other.

Actin filaments are stabilized by actin binding proteins known as actinins of which there are two main types, type 2 and type 3. Each of these is encoded by a specific gene, ACTN2 and ACTN3 respectively.

ACTN2 is expressed in all skeletal muscle fibers whereas ACTN3 is expressed only in fast twitch fibers.

## 3 rs1815739 mutation

A mutation (rs1815739; R577X) has been identified in the ACTN3 gene which results in a deficiency of alpha-actinin 3 in a significant proportion of the population.<sup>[3][4]</sup> Based on ethnicity the deficiency is found in 20-50% of

people. Generally, Africans have the lowest incidence of the mutation while Asians have the highest. Scientists speculate that variations in this gene evolved to accommodate the energy expenditure requirements of people in various parts of the world.<sup>[3]:155-156</sup>

Studies have linked the fiber twitch type with ACTN3, i.e. fast twitch fiber abundant individuals carry the non-mutant gene version. Also, studies in elite athletes have shown that the ACTN3 gene may influence athletic performance. While the non-mutant version of the gene is associated with sprint performance, the mutant version is associated with endurance.<sup>[5][6][7][8][9]</sup>

## 4 Interactions

ACTN3 has been shown to interact with Actinin, alpha 2.<sup>[10]</sup>

## 5 See also

- Actinin

## 6 References

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## 7 Further reading

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