Ever wondered if world class athletes like Usain Bolt and Florence Griffith-Joyner (“Flo-Jo”) were just born sprinters? Or whether something in the genetic make-up of Michael Phelps and Ellie Simmonds is responsible for their lightning speed in the pool?

No doubt there is much hard work, perseverance and focus involved in making it to the very top of any sport, but can your DNA play a role too? Recent research would suggest so. Scientists have identified a variation in a gene called ACTN3 that is shared by almost all world-class athletes who require quick bursts of powerful force to compete in their sport.

The ACTN3 gene is located on chromosome 11

ACTN3 is the gene responsible for making a protein called alpha-actinin-3 that is found in skeletal muscle. Just about all body movement from nodding your head to running is caused by skeletal muscle contraction. This muscle is composed of two types of fibres: slow twitch and fast twitch. Whereas slow twitch muscle fibres are very efficient and can work continuously for long periods, it is your fast twitch fibres that are responsible for generating the short, explosive bursts of power needed in activities such as sprinting and weight lifting. The alpha-actinin-3 protein is found almost exclusively in fast twitch muscle fibres.

Why does it make a difference?

At a specific location within the ACTN3 gene there has arisen a single change in the DNA code, with a ‘C’ base being substituted for a ‘T’ base. This has resulted in two different versions of the ACTN3 gene and which version you carry determines how much of the alpha-actinin-3 protein you have present in your fast twitch muscle fibres.

The C base version, more commonly known as the R allele, results in a fully functioning ACTN3 gene that produces lots of the alpha-actinin-3 protein. The alternative T base version, known as the X allele, puts a premature stop to the growing protein chain resulting in a non-functional alpha-actinin-3 protein. This is not a problem for those of us that are XX, as we don’t actually need alpha-actinin-3 for normal muscle function at all. However, research has shown that its presence is associated with a ‘boost’ in muscle strength and sprint performance. As a result, sprint/ power athletes almost always have at least one copy of the R allele.
So if I want to be an elite sprinter or power athlete, I need to have the R version?
Essentially it would seem so! At the highest level of performance – Olympic level - the ACTN3 genotype appears to make a big difference with around 97% of elite sprint athletes having at least one copy of the R allele. However just having the ‘sprinter’ R version will not ensure you a place at the next Olympics as around 82% of the general population also have at least one! Of course the vast majority of people with the R allele don’t go on to become elite power athletes, and in fact most studies suggest that ACTN3 explains just 3% or less of the variation in muscle function in the general population. This might make all the difference at the top level, but it is unlikely to be discerning in the casual runner. In reality there is much more to becoming a great power athlete, which includes your training, diet, motivation and skill.

I am XX, does that mean I should forget a career in power sports?
Absolutely not, even in genetics there can be exceptions to the rule. Initial studies of world-class athletes did seem to support the conclusion that you could not reach the top levels of performance in power sports unless you had at least one copy of the R allele and were therefore able to make the alpha-actinin-3 protein.

However, after studying hundreds of athletes, researchers eventually identified one individual who bucked the trend: a champion long jumper with the XX genotype. Although his identity was kept confidential, we do know this person had a history of great achievement. He competed at international level by the age of 16 and went on to compete in several European and World championships as well as two Olympic games.

Did you know…?
The latest studies have identified over 240 different genetic variations that are linked to sporting performance, and this continues to increase every year!
This long jumper is the first and so far only world-class power athlete to be found who lacks the “gene for speed”, which demonstrates just how beneficial this gene is to sprint/ power performance. However it also demonstrates that you should never write off your chances on the basis of one small genetic factor!

**Are there any advantages to being XX?**
Your XX genotype may indeed give you a different kind of advantage. Some studies have shown that the frequency of the X version is significantly higher in endurance athletes, which suggests the X version may in fact provide an advantage for slow, efficient muscle performance. So, whilst there is a positive association between the presence of the R allele and greater fast twitch muscle performance, the X allele may alternatively predispose for better endurance performance.

Of course there are many sports where ACTN3 is unlikely to have any effect at all on your ability to perform. Skill-based sports such as snooker, shooting and sailing are just a few examples.

### What about sports where speed AND skill are important?

The effect of ACTN3 can differ markedly between different sports although if it has a power requirement such as sprinting or strength, generally having a copy of the R allele is beneficial.

Scientists recently determined the ACTN3 R and X alleles in 60 top-level professional footballers in Spain. In general they found that the frequency of the RR genotype was significantly higher in the footballers than in a sedentary control group although with notable exception - one individual who was XX was a three-time winner of the UEFA Champions League suggesting that skill and tactical prowess can be just as important as speed!

![Gel Images showing the three possible versions of the ACTN3 gene. Which combination of bands are present in your portrait tells us which genotype you have for this trait.](image)
### Genotype | Description
---|---
RR | **You have two copies of the ‘sprinter’ R allele.** Carriers of the R allele produce the protein alpha-actinin-3, which is associated with a ‘boost’ in muscle strength and performance, possibly due to an increase in the overall size and number of fast twitch muscle fibres. As a result, this is a genotype commonly found in elite sprint/power athletes, and there is even some limited evidence to suggest that there is a skew towards an excess of the RR genotype in this group. The ‘R’ stands for the amino acid arginine, which is encoded by this part of the DNA sequence. This genotype is found in approximately 30% of populations of European ancestry, 17% of populations of Asian ancestry and over 65% of populations of African ancestry.

RX | **You have one copy of the ‘sprinter’ R allele and one copy of the ‘endurance’ X allele.** Individuals with this genotype do produce the protein alpha-actinin-3, although possibly at lower levels than RR genotype individuals. This is still a common genotype seen in elite level sprint/power athletes as having at least one copy of the R allele ensures your body produces the alpha-actinin-3 protein that is needed for increased power performance. This genotype is found in approximately 52% of populations of European ancestry, 59% of populations of Asian ancestry and 30% of populations of African ancestry.

XX | **You have two copies of the ‘endurance’ X allele.** Carriers of the XX genotype do not produce any functional alpha-actinin-3 protein at all. Although a lack of this protein does not have any harmful effects, it appears to be a disadvantage to rapid, forceful muscle contraction such as is needed in speed/power sports. However the X allele may be beneficial to slow, efficient muscle performance such as that needed in endurance sports. The ‘X’ represents the fact that this base change results in a premature stop to the growing protein chain. This genotype is found in approximately 18% of populations of European ancestry, 24% of populations of Asian ancestry and just 5% of populations of African ancestry.

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*Did you know…?*

Interestingly, the percentage distribution of the XX genotype in Ethiopian and Kenyan elite endurance runners is only 11% and 1% respectively, suggesting something other than the ‘endurance’ X version of ACTN3 is responsible for their domination of the long distance running events.