Small genes, BIG IMPACT:

Why single genes matter in dairy

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The dairy industry is one of the leaders in using advanced technologies to select cattle based on performance and functional traits such as milk yield, fertility, udder structure, feet and leg properties, longevity and others. These traits remain the foundation of successful dairy production and are typically influenced by many different genes working together (additively, with each gene having a small effect). However, in some cases, single genes can have a profound impact on the expression of certain traits.

One of the most important advances in modern livestock breeding is the ability to accurately identify each animal's unique DNA profile (the code determining the genetic influence on differences among individuals). This helps breeders understand valuable traits, identify possible health risks, and predict more accurately what an animal may pass on to future generations. It also makes it possible to detect important single-gene mutations within a herd.

Single genes

A single genetic mutation (a random permanent change in an animal's DNA sequence that arises by itself or through outside influences and is carried over to the next generation) can influence whether a cow produces milk more (or less) efficiently, yields higher milk protein percentages, tolerates heat better, or carries a hereditary defect.

Every animal carries a genetic "instruction manual" in each of its cells – its DNA – which influences how it looks, grows and functions. Most production traits are controlled by many genes, each contributing a small effect. These are known as polygenic traits. Single genes, however, work differently. A mutation in one gene can have a major – and sometimes immediate – effect on the expression of a trait.

Occasionally, genes can mutate, leading to an alteration in the expression of a very specific trait, resulting in differences from the normal appearance or performance of animals. These observations or recordings are known as phenotypes. Each animal inherits two copies of every gene – one from each parent. These copies may be identical (homozygous) or different (heterozygous).

In many cases, harmful mutations are recessive, meaning that one form of the gene is dominant (usually the favourable or "normal" variant) and masks the effect of the other. In these cases:

- Animals with two normal copies (BB) are unaffected
- Animals with one normal and one mutated copy (Bb) are carriers and usually appear normal
- Animals with two mutated copies (bb) are affected and show signs of the disorder

This concept is important because detrimental recessive mutations can "hide" within a herd – and even a breed – as carriers appear normal but still pass the gene on to the next generation.

Traits controlled by single-gene mutations are generally not influenced by environmental effects and result in a clear affected or unaffected phenotype. Understanding the genetic status of single genes in a breeding population allows breeders to minimise the risk of affected progeny by making informed mating decisions. Genomic testing enables breeders to test specifically for genes known to cause inherited disorders. The Online Mendelian Inheritance in Animals database (**www.omia.org**) is a valuable reference for such genes.



Which single genes should dairy farmers find relevant?

Milk-related traits

Milk is a complex blend of proteins, fats and sugars. These components are shaped by genetic instructions that vary slightly between cows, and these differences matter.

» **Milk fat:** ABCG2 gene variants lower milk fat and protein percentage but increase milk volume. This may suit producers focusing on volume. In contrast, DGAT1 mutations increase fat percentage but reduce milk volume, which may benefit systems focused on cheese and butter.

» **Milk protein:**

- Beta-casein (A1/A2): A1 is associated with higher fat and protein percentages, while A2 is linked to higher milk volume. A2 is generally considered the more favourable variant.
- Kappa-casein (A/B): A increases protein percentage, while B decreases it. In cheese systems, the BB variant is often desirable.
- Beta-lactoglobulin (A/B): A is associated with increased milk volume, while B improves milk composition (fat and protein).

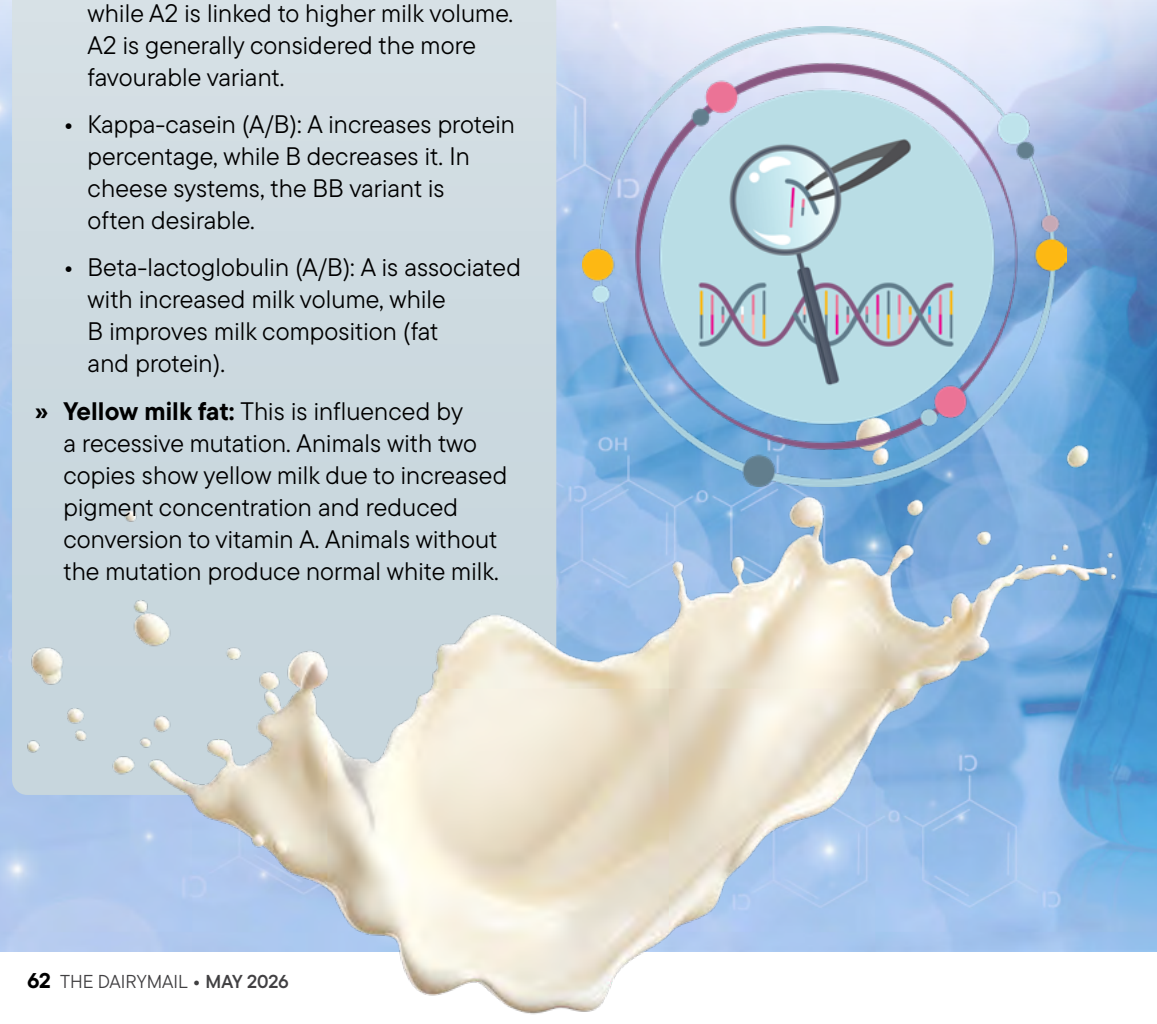
» **Yellow milk fat:** This is influenced by a recessive mutation. Animals with two copies show yellow milk due to increased pigment concentration and reduced conversion to vitamin A. Animals without the mutation produce normal white milk.

Physical appearance

» **Horn status (horned/polled):** SA Stud Book currently reports on the Celtic (Pc) and Friesian (Pf) polled variants, both dominant over the horned (H) gene. If either variant is present, the animal will be polled. Only animals with an HH status will develop horns.

Natural polledness improves animal welfare by eliminating the need for dehorning. Additional variants (Guarani and Mongolian) are currently being validated.

Scurs may also occur – these are loose, horn-like growths not attached to the skull and are controlled by a different gene.



Detrimental (harmful) mutations

Some mutations reduce survivability, while others affect reproduction. These often result in non-functional or poorly functioning proteins, disrupting normal biological processes. Due to historical selection pressure and inbreeding, several genetic disorders exist in dairy breeds.

Fortunately, these can be managed and avoided through careful breeding decisions. Genomic testing by SA Stud Book allows screening for known detrimental mutations. While some are not present locally, others occur in South African populations. All listed below are recessive unless otherwise stated:

- Arachnomelia
- Bovine Leukocyte Adhesion Deficiency (BLAD)
- Bovine Lymphocyte Intestinal Retention Deficit (BLIRD)
- Bovine Spinal Dysmyelination (BSD)
- Retinitis Pigmentosa (REPTIG)
- Jersey Neuropathy with Splayed Forelimbs (JNS)
- Bulldog Dwarfism (BD)
- Citrullinaemia
- Complex Vertebral Malformation (CVM)
- Deficiency of Uridine Monophosphate Synthase (DUMPS)
- Idiopathic Male Sub-Fertility (IDMS)
- Haplotypes (JH, HH, AH, MH, BH groups)
- Spinal Muscular Atrophy (SMA)
- Weaver Syndrome
- Brachyspina

Why it matters

Although sometimes seen as less important than polygenic traits, single genes can have a significant impact on dairy production. By testing for and understanding these mutations, breeders gain an additional tool to manage risk, reduce economic losses, protect animal welfare, and make more informed mating decisions.

Animals carrying recessive mutations may appear normal but can silently pass on unwanted genes. Genomic testing allows these risks to be identified before problems arise.



One test, many advantages

A genomic test not only reveals the carrier status for single genes but also offers several additional advantages. Accurate parentage can be confirmed and even determined in cases where parent information is unknown.

While pedigree data helps estimate inbreeding, missing information may lead to underestimation. Genomic testing provides a more accurate measure of true inbreeding levels (homozygosity) and genetic relationships between animals. This helps prevent excessive inbreeding in future generations.

SA Stud Book also uses genomic data to determine breed composition, which is particularly valuable for crossbreeding systems.

The greatest advantage remains the ability to use genetic information to predict an animal's value as a parent for the next generation. For this to be most effective, testing must be done locally so that breeding values are relevant to South African conditions. This is achieved through SA Stud Book evaluations.

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