



# Using Genomics Part II: Risk Management of Genetic Defects

## The Oklahoma Cooperative Extension Service Bringing the University to You!

The Cooperative Extension Service is the largest, most successful informal educational organization in the world. It is a nationwide system funded and guided by a partnership of federal, state, and local governments that delivers information to help people help themselves through the land-grant university system.

Extension carries out programs in the broad categories of agriculture, natural resources and environment; family and consumer sciences; 4-H and other youth; and community resource development. Extension staff members live and work among the people they serve to help stimulate and educate Americans to plan ahead and cope with their problems.

Some characteristics of the Cooperative Extension system are:

- The federal, state, and local governments cooperatively share in its financial support and program direction.
- It is administered by the land-grant university as designated by the state legislature through an Extension director.
- Extension programs are nonpolitical, objective, and research-based information.
- It provides practical, problem-oriented education

for people of all ages. It is designated to take the knowledge of the university to those persons who do not or cannot participate in the formal classroom instruction of the university.

- It utilizes research from university, government, and other sources to help people make their own decisions.
- More than a million volunteers help multiply the impact of the Extension professional staff.
- It dispenses no funds to the public.
- It is not a regulatory agency, but it does inform people of regulations and of their options in meeting them.
- Local programs are developed and carried out in full recognition of national problems and goals.
- The Extension staff educates people through personal contacts, meetings, demonstrations, and the mass media.
- Extension has the built-in flexibility to adjust its programs and subject matter to meet new needs. Activities shift from year to year as citizen groups and Extension workers close to the problems advise changes.

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In recent years an abundance of genetic defects in a variety of breeds have resulted in substantial associated economic losses. In this fact sheet, we will discuss how these defects are inherited, how to determine the probability of producing an affected calf, and strategies to decrease issues related to genetic defects in your herd.

### Inheritance

The bovine genome is made up of 29 pairs of autosomal chromosomes plus the sex chromosomes. Genetic defects are a result of alleles that cause a lethal condition or that severely handicap the performance of an individual. Simply speaking, at any location in the genome where there is a mutation, there are two alleles, which can be thought of as alternate forms of a gene. One allele comes from the sire and the other from the dam. There are many mutations within the genome which cause no known effects. However, the ones which are of interest are either ones which cause small effects on phenotype (The animals' performance or how it looks), typically for performance traits, and those which cause large detrimental effects on phenotype or are lethal. Most genetic defects in beef cattle are a result of recessive autosomal mutations. The term recessive autosomal reflects the mode of inheritance for these defects meaning the mutation is on one of the 29 autosomal chromosomes (not a sex chromosome), and it is not expressed as a phenotype unless the animal receives two copies of the damaged allele (one on each of the two chromosomes inherited from its parents). The inheritance of these genetic defects works exactly like the inheritance of horns or red coat color, which are also autosomal recessive conditions and which require two copies of the red (or horned) allele before a difference is seen in phenotype. Because these traits are recessive, possessing only one copy of each of these alleles (a defect, red color, or horns) is not enough to change phenotype, because it is masked by a dominant allele (normal condition, black color, or polled). These animals are often called carriers, because they carry a recessive condition, but do not express it.

In any mammalian genome (the size of most mammalian genomes is three billion base pairs), it is almost assured that there will be multiple recessive lethal genetic defects present within the DNA sequence. However, because they are recessive, there must be two copies in the genome (one from each parent) in order to see affected progeny. In practice, the pairing of two disease alleles rarely occurs when animals are not inbred, because each animal likely carries mutations that cause different diseases. One of the best strategies to avoid incidence of genetic defects is to avoid mating animals to

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their relatives, or to employ a planned crossbreeding system (but keeping carrier status in mind when mating animals that have common breed composition).

### Determination of Risk

When the mode of inheritance is known, we can calculate the probability of producing an affected calf by knowing the carrier status of the parents. Because the normal allele is dominant, there is no way to visually determine which animals are carriers and which are not. The only way to make that determination is through genetic testing or through known parentage of an affected calf.

If the animal tests as normal for a particular genetic defect, it has two normal alleles (represented by NN) and if the animal is tested and determined a carrier, it has one normal allele and one disease allele (represented by Nn). If an animal has ever produced an affected progeny (has the defect shown by phenotype, represented by nn), they can automatically be determined a carrier (Nn) and do not need to undergo genetic testing. If an inherited disease/defect is lethal, all adult animals are either normal or carriers (NN or Nn). They cannot have two copies of the disease allele (nn) and live.

When the carrier status of the parents is known, we can calculate the probability of producing normal, carrier, or affected progeny using what is called a Punnet square. It is simply a square with four quadrants and works like a multiplication table. The alleles for each possible parental gamete (sperm and egg) are placed around the outside and the letters are matched on the interior squares and the percentages are multiplied together to determine the probabilities of each status in the progeny. To illustrate this point, consider the following examples:

**Example 1. Punnet square for the mating of two normal (NN) animals.**

		Male	
		N 50%	N 50%
Female	N 50%	NN 25%	NN 25%
	N 50%	NN 25%	NN 25%

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