

## Genetic Defects

Occasionally a case of a potential genetic defect is reported to the CSA. Genetic defects are not unique to the Simmental breed, as all animal populations carry some genetic defects. While extremely rare, these cases are also serious and should be addressed in a straightforward manner.

The CSA has a genetic defect policy that outlines how these cases should be dealt with. [Click here](#) to read the policy

Generally cases are worked through with an attending veterinarian. Where possible, DNA samples should be collected on the affected animal and its' parents, in order to confirm parentage. As well, your veterinarian can recommend and organize any appropriate pathology tests.

Most common genetic defects fall into the category of recessive, and the causative genes are extremely low frequency in the gene pool. Recessive genes may be transmitted for generations, with the defect never being expressed until a mating combines two recessive genes (See example).

		Parent 1	
		Normal (N)	defect (d)
Parent 2	Normal (N)	Normal (NN)	Carrier (Nd)
	defect (d)	Carrier (dN)	Affected (dd)

In many cases the cause of the defect may be environmental, rather than genetic. Examples of other causes may include teratogenic plants, mineral deficiencies or other forms of poisoning such as lead. For this reason, the CSA strongly encourages the use of appropriate testing where available.

Some of the defects reported in the Simmental population include:

Disorder	Description	Test
Tibial hemimelia (TH)	Shortened tibia (bone of lower hind leg)	DNA Test
Thrombopathia	Bleeding disorder	Pathology
RT 1/29, 14/20	Translocation	Chromosome test

Genetic defects are extremely rare, however the CSA does work to track and resolve any occurrences that are reported.