

Table 1

Genetic defects in cattle

Condition	Description	Inheritance	Breeds affected ¹
Achondroplasia (bulldog dwarfism)	Homozygous calf may be aborted at 6 to 8 months gestation; has compressed skull, nose divided by furrows, and shortened upper jaw for a bulldog facial appearance; heterozygous calf is small and heavy-muscled	Incomplete dominant	
Alopecia	Lethal abnormality very similar to hypotrichosis; takes laboratory analysis to distinguish; calves have kinky, curly hair that is soon lost in patches around the head, neck, and shoulders; skin changes and anemia occur in all cases; death before 7 months of age due to anemia	Simple recessive	Polled Hereford
Ankylosis	Abnormal union of any joints in calf; cleft palate frequently occurs	Recessive	
Arthrogyposis (palate-pastern syndrome; rigid joints); Arthrogyposis Multiplex (AM; curly calf syndrome)	Pastern tendons are contracted; joints of all four legs fixed symmetrically; upper part of mouth not properly fused together (cleft palate); calves usually die shortly after birth; AM in Angus includes twisted malformation of spine and fixed leg joints	Simple recessive; many environmentally caused forms	Angus (AM), Red Angus (AM), Charolais, many other breeds
Brachynathia inferior (parrot mouth)	Cattle have a short lower jaw; under- or overshot jaw with varying degrees of expression when polygenic	Simple recessive; polygenic (more common)	Angus, Simmental
Cryptorchidism	Retention of one (unilateral) or both (bilateral) testicles in body cavity instead of descending into scrotum	Sex-limited trait; polygenic	Many breeds
Dermoid (feather eyes)	Skin-like tissue masses occur on eye or eyelid; cattle may become partially or completely blind	Polygenic	
Double muscling	Muscle enlargement with large grooves between muscle systems; particularly evident in hind leg; cattle usually grow slowly; fat deposition (internal and external) is reduced; typically fine boned; considerable variation in expression	Simple recessive; dominant in Piedmontese	Belgian Blue, Piedmontese, Angus, Red Angus
Dwarfism	At least three types in cattle; snorter dwarfism: most common; short, blocky appearance; deformed bone growth in nasal passages causing difficult breathing; long head dwarfism: small size but normal nasal passages; compress dwarfism: extremely compressed body conformation with one compress alleles and one normal gene; dwarf and death at or soon after birth with two compress alleles	Simple recessive (snorter dwarfism; long head dwarfism); incomplete dominance (compress dwarfism); environmental causes	Angus, Hereford, Brahman, Dexter
Fawn Calf Syndrome	Abnormal crouched posture at birth, resembling a deer fawn, with the feet placed more to the rear than normal, hocks pulled up and back and the spine slightly arched; flat down on pasterns during first days of life; tend to be taller and more slender with poor foot conformation; poor muscle development; difficulty in some with movement and suckling; can result in calf death but most can walk, suckle, and survive; poor growth performance; early onset of degenerative arthritis	Simple recessive	Angus
Hypotrichosis (hairlessness)	Partial to almost complete lack of hair; hair grows in and falls out, so appearance varies over time; non-lethal	Simple recessive; low frequency	Hereford
Hypotrichosis (rat-tail)	Form of congenital hypotrichosis; colored hair anywhere on body is short, curly, malformed, and sometimes sparse; abnormal tail switch; often confused with premature birth; slower post-weaning weight gains	Interaction between two loci where at least one gene is for black color and must be heterozygous at the other locus involved	Matings of Simmental with Angus; Hereford can carry
Ideopathic Epilepsy (IE)	Neurological disorder causing seizures	Simple recessive	Hereford
Mannosidosis	Lethal nervous disease; failure to thrive; progressive incoordination; aggressive disposition when disturbed; death typical before sexual maturity reached	Simple recessive	Angus, Murray Grey, Galloway, Salers
Neuraxial edema (maple syrup urine disease)	Calf is normal size at birth but may not be able to stand or lift head; sudden touch or loud noise may cause vigorous extension of the legs and neck; muscle spasms of neck and legs may last for 1 to 2 minutes; death typical within 5 days	Simple recessive	Hereford, Shorthorn
Neuropathic Hydrocephalus (NH; water head)	Fluid accumulation in brain ventricles (internal: water head) or cranium (external); bulging forehead; calves usually die shortly after birth	Simple recessive; environmental causes	Angus, Red Angus (external), Hereford (internal), many other breeds
Oculocutaneous Hypopigmentation; Heterochromia Irides (white eye)	Hair coat is bleached color; iris is pale blue around pupil with tan periphery	Simple recessive	Angus
Osteopetrosis (marble bone disease)	Bone tissue fills marrow cavity of the long bones; brittle bones that break easily; calves have short lower jaws, protruding tongues, and impacted molar teeth; calves usually born dead 2 to 4 weeks preterm	Simple recessive	Angus, Red Angus, Simmental, Holstein
Polydactyly (extra toes)	One or both front feet or sometimes all four feet have outer dew claw develop into extra toe	Polygenic	Simmental, Holstein
Progressive bovine myeloencephaly (weaver calf)	Calves develop a weaving gait at 6–8 months of age and get progressively worse until death at 12–20 months of age	Simple recessive	
Prolonged gestation	Fetus fails to trigger parturition; calving must be induced or the calf removed; calf is often extremely large and often dies	Simple recessive	
Protoporphyrria (photosensitivity)	Sensitivity to sunlight; development of scabs and open sores upon sunlight exposure; liver function affected; seizures possible	Simple recessive	Limousin
Pulmonary Hypoplasia with Anasarca (PHA)	Fluid collection in skin and body cavities at birth; lethal to calves; usually causes dystocia because of added weight; lungs underdeveloped	Simple recessive	Shorthorn, Maine-Anjou, Chianina, Simmental
Syndactyly (mule foot)	One or more hooves are solid in structure rather than cloven; front feet most often affected; can occur in all four feet; cattle cannot tolerate hot temperatures; high mortality rate in calves	Simple recessive	Chianina, Angus, Simmental, Holstein
Tibial Hemimelia (TH)	Calves born with twisted legs with fused joints, large abdominal hernias, and skull deformity	Simple recessive	Shorthorn, Maine-Anjou, Chianina, Simmental, Galloway
Translocations	Part of a chromosome breaks off and attaches to another chromosome; carriers have reduced conception rates and increased abortion rates	Simple recessive	1/29 in Simmental, Charolais, and Blonde D'Aquitaine; 14/20 or 14/21 in most Continental breeds

¹Specific defects have been noted in the breeds listed. However, these defects could occur in other breeds.