Swine Genetic Abnormalities – Pork Industry Handbook
Michigan State University Cooperative Extension Service

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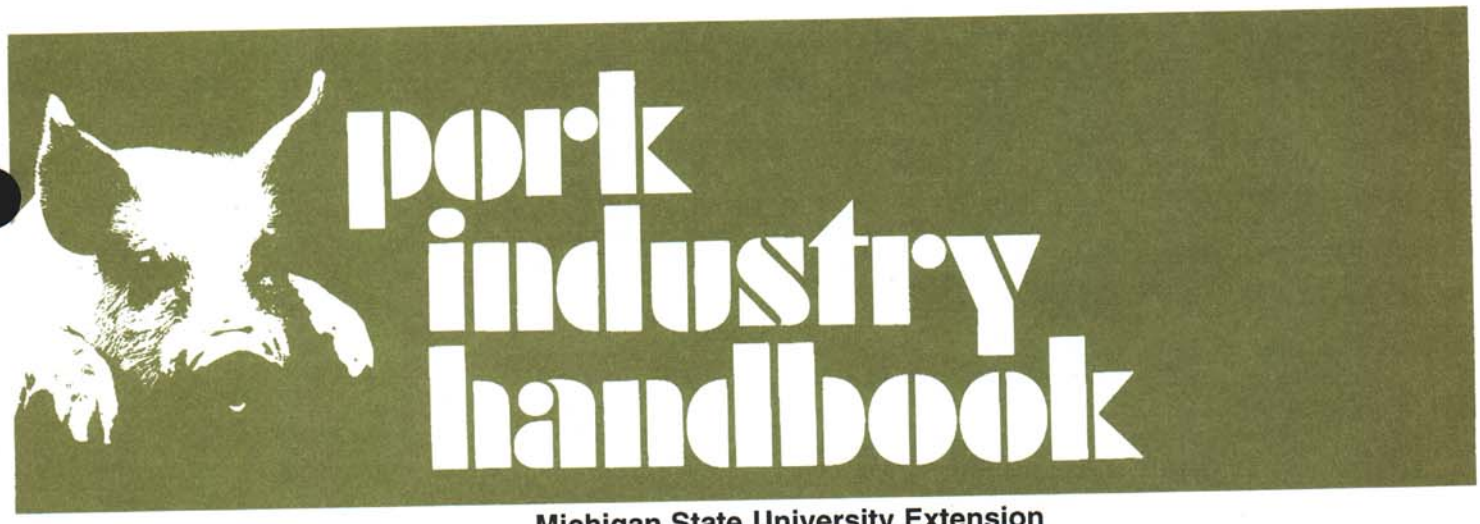

Michigan State University Extension

Swine Genetic Abnormalities

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Abnormalities are deviations from normal development and can involve any part of the pig, internal or external. These defects can impair the pig's ability to function or even cause death. Anatomical abnormalities or defects occur in at least 1% of newborn pigs. These defects may be caused by genetic or environmental factors. Although the frequency of these defects is low, they can be frequent enough in an individual herd to cause substantial economic loss.

Traditionally, producers have assumed all abnormalities were of genetic origin since the defects often are dramatic and congenital. Congenital defects only imply that they are visible at birth and not that they have a genetic cause. We now recognize that environmental factors such as viral infections, dietary deficiencies, and ingestion of certain drugs, chemicals, and pesticides during pregnancy may alter normal prenatal development of the pig. In fact, some cases of an abnormality may result from genetic factors while other cases of the same abnormality may result solely from environmental factors.

Determining the Causes of Abnormailities

When an abnormality occurs, the producer should objectively ascertain its cause. Answers to the following questions can assist in determining if the abnormality is of genetic origin.

1. Were the defective animals all of one breed or by one sire? Generally, an abnormality is so rare that it is unlikely to be of genetic origin if it occurs in more than one breed, in more than one sire progeny group when the sires are unrelated, or in a crossbred population but not in the contributing purebred populations.
2. If the condition is present in only one sire's offspring, is every litter affected? Study carefully the pedigrees of the dams of each litter. If most of the afflicted litters are from half-sister dams, be suspicious of a genetic cause. If it occurs in nearly every litter by a boar and the dams are unrelated, look for environmental influences; otherwise, dominant gene action is implied and the boar usually would exhibit the defect already.
3. Is the abnormality present in litters produced by matings of closely related animals (inbreeding)? Inbreeding results in increased frequency of expression of traits caused by recessive genes. Inbreeding with selection will reduce the frequency of recessive genes, but progress will be slow if the initial frequency of the recessive gene is low.
4. Were the afflicted litters under similar environmental, nutritional, and management conditions during gestation? A genetic cause is unlikely if the affected litters were born during the same time span and litters born during a different time span, sired by the same boar and from dams of similar breeding, were free of the condition.

Type of Genetic Mechanisms

Understanding the type of genetic mechanism responsible for a specific genetic abnormality will aid producers in developing methods to remove the problem from their herd. Causes for genetic disorders can be:

Chromosomal Aberrations. Chromosomes are threadlike bodies in the nucleus of a cell that carry genetic information. Chromosomes occur in pairs in body cells. A sperm or ovum contains only one of each pair of chromosomes. There are two types of chromosomes. One pair of chromosomes is known as the sex chromosomes because they are involved in determining the sex of an animal. In mammals the sex chromosomes are called X and Y, with the X chromosome being much larger than the Y chromosome. Females have two X chromosomes, and males have one X and one Y chromosome. All chromosomes other than sex chromosomes are called autosomal chromosomes.

1. **Number of chromosomes increased or decreased compared to normal (19 pairs in pigs).** The effects of increased or decreased number of chromosomes are usually so severe that early embryo death occurs. The exception is increased number of sex chromosomes, which usually results in infertility.
2. **Structural Alterations.** Structural alterations usually are the result of pieces of chromosomes breaking off and recombining in a non-normal manner during the process of sperm or egg formation. Such defects also tend to result in major abnormalities that often cause fetal death or early death of the newborn pigs. However, data from Europe show that some translocations (movement of pieces of one chromosome to another chromosome) are not fatal to some pigs and in boars may cause lower fertility. A sharp reduction in litter size in a group of sows mated to a specific boar may indicate that the boar possesses a translocation.

Simple Genetic Inheritance

The gene is the smallest unit of inheritance, and is a structural part of a chromosome. If genes at only one location on the pair of chromosomes are responsible for the disorder, it is considered simply inherited.

1. **Simple Recessive.** Recessive disorders are those caused when two copies of a gene (homozygous dd) are required for the trait to be expressed. Recessive genes are denoted with small letters. Since these are recessive, the gene is hidden in the heterozygote form Dd and may exist at a low frequency in the population. It is difficult to completely remove a recessive gene from a herd. Mating of related individuals increases the chance of uncovering such recessive disorders.
2. **Simple Dominant.** If only one copy of the gene (D_) is required to produce the defect, it is considered dominant. Dominant genes are denoted with capital letters. Generally only abnormalities that are considered mild are inherited as dominant since natural selection will tend to remove all DD (homozygous—two copies of same gene) or Dd (heterozygous—one copy of each gene) animals if the defect is severe.
3. **Sex-linked Recessive.** Sex-linked genes are found on only the X chromosome since the Y chromosome has few active genes. These genes also tend to be recessive, but since males have X and Y sex chromosomes they will exhibit the disorder at a higher frequency than females that have two X chromosomes. Only if dams are carriers can female offspring inherit two recessive genes (one from each parent) and express a sex-linked recessive trait.
4. **Sex-limited (recessive or dominant).** A sex-limited disorder means that it occurs only in one sex because it is related to sexual differences. For example, scrotal hernias are sex-limited to the male.

Multigenic Inheritance

Multigenic disorders are those controlled by genes at two or more locations on the chromosomes. Usually we estimate heritability (h^2) for these traits, which offers some estimate of how much genetic control exists for the trait. Heritability estimates range from 0.0 to 1.00 and indicate what proportion of all differences for a trait are caused by genetic differences.

Genetic Liability

Disorders that have a genetic liability or tendency are those which have a genetic basis but are not expressed unless a specific environmental situation or insult occurs. The heritability of liability may be high, but unless the environment is adverse, incidence of the defect is small. For example, an animal may inherit a tendency to have splaylegs but will not show the condition unless the floors are slick or unless a nutritional deficiency occurs.

Experimental Evidence

Unfortunately, most of the evidence concerning genetic defects in pigs is just from observation. Previous scientific reviews of genetic abnormalities do not consistently agree as to the mode of inheritance. Few planned experiments have been initiated to test the theories concerning inheritance. Therefore, comments concerning most of the following abnormalities are only best estimates.

Important Genetic Abnormalities

Porcine Stress Syndrome (PSS) This condition is characterized by a progressive increase in body temperature, muscle rigidity, and metabolic acidosis leading to sudden death of heavy muscled pigs. PSS also can lead to the production of pale, soft, and exudative (PSE) meat. PSS is inherited as an autosomal recessive. Pigs that are heterozygous or homozygous positive are superior to homozygous negative pigs in muscling. This superiority may cause the producer to select carriers or reactors when the opportunity arises, thus increasing the frequency of the gene for PSS. To reduce incidence of PSS and PSE, all PSS animals, their parents, and littermates should be eliminated from the breeding herd. PSS is caused by a defect in the gate of the Ca release channel that prevents it from closing, allowing Ca to continue to leak causing a high rate of metabolism leading to the prolonged acidosis and fatal collapse characteristic of this disorder. The presence or absence of this mutated gene is readily detected by the use of a DNA test. Only 5 ml to 10 ml of blood or tissue are required. At a diagnostic laboratory the DNA is extracted, amplified, and evaluated. There are two techniques available to classify the animals as normal (NN), heterozygote carrier (Nn), or stress reactor (nn). This test offers a highly accurate means of detecting each animal's genotype for PSS. Licenses are required for the use of this diagnostic test as it has been patented internationally.

Rendement Napole (RN) Carriers of this dominant gene show lower pH, paler color, lower protein extractability, lower water-holding capacity, and greater cooking loss in the loin. On the positive side, carriers have a lower shear force value, a stronger taste and smell, and greater acidity. This effect is reported primarily in Hampshire or Hampshire-cross pigs. This condition results from a single gene with two alleles that exhibit dominant-recessive inheritance. The dominant allele (RN-) is responsible for elevated glycogen stores in the muscle that results inincreased production of lactate and subsequent reduction in ultimate pH. The three genotypes currently can be classified by the estimation of glycolytic potential. The molecular basis has not yet been identified.

Scrotal Hernia

This sex-limited condition in males (Figure 1) is believed to result from a weakness of the musculature surrounding the inguinal canal, permitting the intestines to drop into the scrotum. It occurs much more frequently on the left side.

Figure 1. Scrotal hernia in pig.

Extreme care in castration generally will prevent great economic loss. At least two pairs of genes have been suggested as the mode of inheritance, but maternal and environmental influences are certain to be involved. In herds where frequency has been high, heritability of liability of scrotal hernia appears to be in excess of 0.5. If the incidence is high in a herd and if answers to the questions posed in the earlier section indicate a genetic cause, eliminating the affected individuals can reduce the incidence, their parents, and littermates from the breeding herd.

Umbilical Hernia

A weakened supportive musculature in the navel area resulting in intestines protruding through the belly wall is referred to as umbilical hernia, "belly rupture," or "belly bust" (Figure 2). Some afflicted individuals may die during growth due to strangulation of the intestine, but most reach market weight with no apparent adverse effects. Surgical correction is seldom recommended. Although genetic causes have been suggested, environmental causes such as navel infections are far more likely. This defect may have a genetic liability that is magnified by adverse environmental conditions, such as crowding to conserve heat during cold weather. Since genetic causes for this condition are not known, culling of related individuals is not recommended. Under no circumstances should a surgically corrected animal be used for breeding.

Atresia Ani

This condition is characterized by a pig being born without a rectal opening (Figure 3). Boar pigs die within a few days unless an opening is made surgically to permit him to void feces. Females with no anal opening can commonly defecate through the vulva (via an opening from the rectum into the vagina) and grow normally. There is little doubt that the condition has a genetic basis, but is also definitely not due to a single gene. If incidence is high and if answers to the questions posed in the earlier section indicate a genetic cause, the incidence can be reduced by culling all affected individuals, their parents, and their littermates from the breeding herd.

Cryptorchidism

Cryptorchids or ridglings are male pigs (Figure 4) with one or both testicles retained in the body cavity. Animals with both testicles retained are sterile. Sex-limited inheritance with at least two gene pairs seems possible. Parents and littermates of afflicted individuals should be culled from the breeding herd.

Hermaphrodites

Hermaphrodites are frequently observed among the Large White and Landrace breeds of Europe and with a frequency of 0.1 to 0.5% in Yorkshires and Landrace in the United States. Sex chromatin studies show most hermaphrodites (Figure 5) to be genetic females (XX genotype), but to possess portions of the male sex organs. USDA studies suggest female offspring of known carriers to segregate in a 3:1 unaffected to affected ratio and assumed sex-limited recessive inheritance as the probable cause. Altered chromosome structure has also been theorized, such as a portion of the Y chromosome being translocated to an X chromosome or a mutation in the X chromosome that has removed the depressing effect of maleness that is thought to exist on the normal X chromosome. In any case, this condition is generally under relatively simple genetic control. If incidence is high and if answers to the questions posed in earlier section

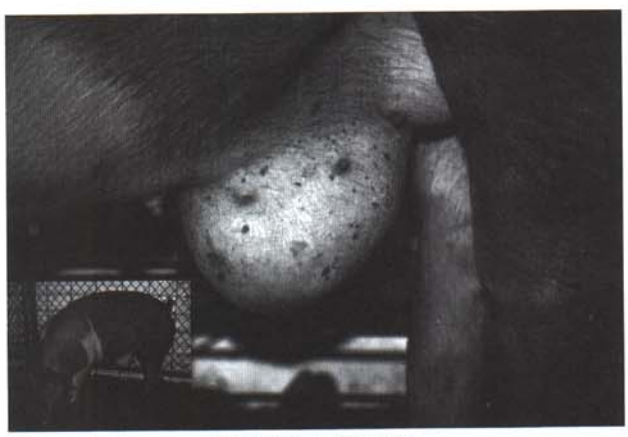

Figure 2. Large umbilical hernia in pig.

indicate a genetic cause, the incidence can be reduced by culling afflicted individuals, their parents, and their littermates.

Occasionally, female hermaphrodites will occur in very large litters. This may result from uterine crowding which causes a male and a female fetus to share a common blood supply due to joining of their placentas. This is similar to the freemartin syndrome in cattle. These hermaphrodites are not caused by genetics.

Nipple Abnormalities

Inverted nipples are the underlying abnormality of the greatest concern. This condition is characterized by failure of nipples to protrude from the udder surface.

The teat canal is held inward, forming a small crater so that normal milk flow is prevented. This abnormality has a genetic cause, but the number of pairs of genes involved is unknown. The heritability is estimated to be approximately 20 %. Caution must be exercised in evaluating underlines since nipples surrounded by a ring of loose skin are not inverted if the nipple tip

Table 1. Other anatomical defects and inherited disorder of swine.

Disorder	Description	Probable cause
Blood warts (Melanotic tumors)	Moles or skin tumors. Increase in size with age. Tumors heavily pigmented and contain hair. Injury causes depigmentation. Common in Durocs and Hampshires.	Inheritance unknown but multigenic inheritance has been postulated.
Brain hernia	Skull fails to close and brain protrudes. Generally lethal.	Simple recessive inheritance suggested.
Cleft palate	Palate does not close. Harelip results. Generally lethal.	Recessive lethal has been theorized but may result from multigenic genetic liability influenced by an environmental insult.
Gastric ulcers	Erosion of the epithelial lining of the stomach. Generally in the esophageal region.	Heritability estimates ranging from low to high have been reported. Pelleted and finely ground diets, high unsaturated fats and low selenium in the diet, copper toxicity, and psychosomatic factors have been found to cause that problem.
Hemophilia (bleeders)	Slow clotting time. Death results from slight wounds or from navel cord hemorrhage.	Known to be caused by mycotoxins in feed or vitamin K deficiency. One confirmed case of simple recessive inheritance.
Humpback	Crooked spine behind shoulder.	Likely to have genetic cause but inheritance is unknown.
Hydrocephalus	Fluid on the brain. Brain cavity much enlarged.	A lethal gene inherited as a simple recessive.
Lymphosarcoma (Leukemia, lymphoma)	Malignant tumors of the lymph nodes with increased lymphocite count. Stunted growth and death before 15 months of age.	Convincing evidence of an autosomal recessive.
Motor neuron disease	Distinctive locomotor disorder of nursery pigs, characterized by inability to coordinate muscle movements and slight paralysis.	Strong suggestion of autosomal dominant inheritance.
Oedema (myxoedema, dropsy, hydrops)	Abnormal accumulation of fluid in tissue and body cavities. Possibly associated with a thyroid defect.	Autosomal recessive disorder suggested.
Pseudo-vitamin D deficiency (rickets)	Indistinguishable from non-genetic lack of vitamin D resulting from deficiency of calcium or insufficient exposure to sunlight. The most noticeable effect is bowing of the limbs.	Inherited as an autosomal recessive.
Rectal prolapse	Protrusion of the terminal part of the rectum and anus.	Many environmental influences including coughing, piling,feed constituents, antibiotics, diarrhea have been implicated though genetic liability may exist.
Persistent frenulum	A close attachment of the prepuce to the body by a mucous membrane resulting in inadequate protrusion of the penis and inability to breed.	Inheritance unknown.
Screw tail (kinky tail)	Flexed, crooked, or screw tail caused by fusion of caudal vertebrae.	Multigenic recessive inheritance has been postulated.
Swirls (hair whorls)	Hair forms a cowlick or swirl on neck or back.	At least 2 pairs or recessive genes are involved.
Wattles (tassles, bells)	Fleshy, cartilaginous appendages covered with normal skin and suspended from the jaw.	Single-locus autosomal recessive inheritance.

is present. Nipples located near the sheath of boars are often falsely classified as inverted. Because of the uncertainty of the genetics of this trait, culling of parents and littermates is not recommended. Individuals with a high proportion of inverted nipples should not be placed in the breeding herd.

Blind nipples are those that simply have no visible nipple or canal. In most cases, these are considered to result from injury, particularly from irritation and scabbing caused by abrasive floors. Hence, blind nipples are not considered to have a genetic cause.

Tremors

Most cases of tremors in pigs are congenital (present at birth), but certain forms are often first expressed at later ages. The condition is also known as myoclonia congenita, trembles, shivers, shakers, or jumpy pig disease. The symptoms generally are present within a few hours of birth and are characterized by rhythmic twitching of the neck and legs with varying degrees of intensity. Animals with minor tremors are able to move and nurse, but those severely affected fail to function normally and are subject to starvation, chilling, and overlaying by the sow. The shaking becomes more intense with chilling and excitement. Survivors generally show less intense tremors with advancing age. Porcine circovirus has been isolated from a pig with congenital tremors and may be one of the causes of this condition. Tremors among pigs exhibiting morphological lesions have been classified into five types. Types A, and AII are

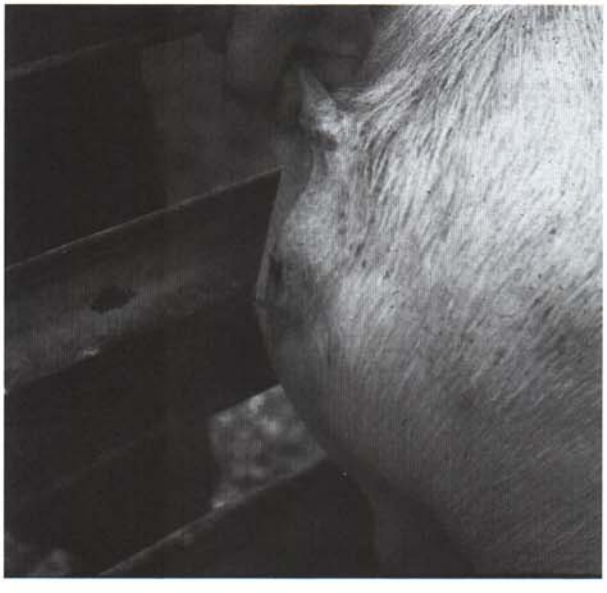

Figure 3. Atresia Ani in female.

caused by viruses and types AIII, AIV, and high frequency are considered to be under genetic control.

Type AI is caused by transplacental infection by particular strains of hog cholera virus. Necropsy reveals a reduction in size of both the cerebellum and the spinal cord. Since the United States is free of hog cholera, this type should not be encountered.

Type AII is caused by placental transfer of viruses other than hog cholera. Pseudorabies, parvo, and smedi viruses, among others, are known to cause this condition. Mortality is less than in the AI form. When tremors suddenly appear at a high

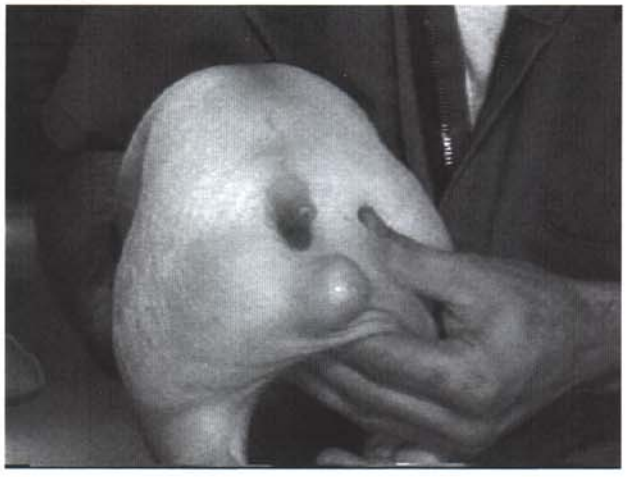

Figure 4. Cryptorchid (one testicle retained) in male pig.

frequency in a herd, it is generally type AII. Type AIII is a hereditary form. It has been reported to occur in only Swedish Landrace and to be inherited as a sex-linked recessive trait. Observations at Iowa State University found the condition to exist in many strains of Landrace and refute the sex-linked mode of inheritance in favor of a simple recessive type. Necropsy reveals a decrease from the normal amount of myelin in the central nervous system, resulting in a smaller than normal spinal cord.

Type AIV is thought to occur only in the BritishSaddleback breed. It is considered to be a simple autosomal recessive and is characterized by severe deficiency of myelin (material encasing some nerve fibers) in the central nervous system. The myelin is unstable and already breaking down when the piglet is born.

High-frequency tremors are characterized by muscular weakness and a very intense tremor of the legs when standing and walking but not when at rest in a lying position. The intensity of the tremor and muscular weakness progressively increases with age resulting in greater instability while standing. High-frequency tremors have autosomal dominant inheritance.

If incidence of tremors is high, and if answers to the questions posed in the earlier section indicate that type AIII (or type AIV) is involved, the incidence can be reduced by culling affected individuals, their parents, and their littermates.

Neonatal Diarrhea

Neonatal diarrhea in piglets is often caused by strains of *Escherichia coli* bacteria. Various strains of *Escherichia coli* have different cell-surface antigens that combine with glycoprotein receptors on the wall of the piglet's intestine, enabling the bacteria to attach themselves to the intestines. Once attached, the bacteria proliferate, releasing enterotoxins and thus producing diarrhea, which can lead to high mortality. Certain piglets lack the intestinal receptor for the K88 strain of *Escherichia coli.* These pigs are therefore resistant to K88 bacteria and to diarrhea caused by K88 strains. Lack of the K88 receptor is a single-locus autosomal recessive trait. It also seems likely that resistance to the K99 and F4 strains of *Escherichia coli* will also turn out to be determined by a single gene.

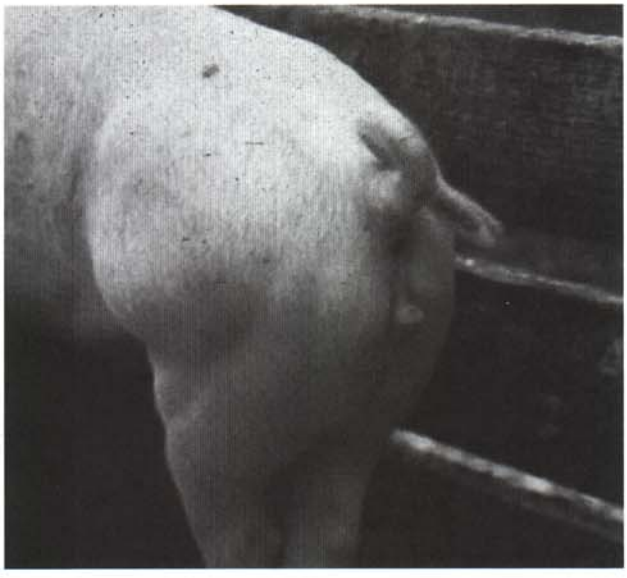

Figure 5. Hermaphrodite pig with large clitoris.

Leg Defects

Many abnormalities of the legs have been noted in pigs. Several of the most commonly observed congenital condi-tions are as follows:

Splayleg. This condition, also known as spraddle legs, is probably the most common of the leg disorders (Figure 6). The rear legs are most often affected but the forelegs are sometimes involved. Histological examination often reveals incomplete development of muscle fibers in the front and rear legs. These muscles are responsible for pulling the hind legs and forelegs toward the body. The cause of the defect is unknown, but evidence points to multiple factors including genetic liability to muscle weakness, viral infection, nutritional deficiency, and slippery floors. Heritability of liability appears to be above 0.40 in some breeds. If the incidence is high and if answers to the questions posed in the earlier section indicate a genetic cause, the incidence could be decreased by culling affected individuals, their parents, and their littermates.

Small inside toes. This condition is believed to be of genetic origin, but the mode of inheritance is unknown. Duration of service in the herd is normally reduced in animals possessing this defect. Only culling of affected individuals is recommended.

Bent legs. This lethal abnormality generally affects only the forelegs, although in isolated cases the rear legs are involved. The legs are bent back at right angles and are stiff. This disorder is recessively inherited, but can be caused by non-genetic factors such as ingestion of Jimson weed or tobacco by pregnant sows. If incidence is high and if answers to the questions posed in the earlier section indicate a genetic cause, the incidence can be reduced by culling the parents and littermates of the affected individuals.

Polydactyly. Extra toes and/or dewclaws are quite common in pigs. This genetic condition is of little economic value. Culling of affected individuals is recommended.

Syndactyly. This disorder, also known as mulefoot, is recognized by the presence of only one toe per foot rather than two and is inherited as a single dominant gene. It has been nearly eliminated from the U.S. pig population. Only culling of affected individuals is recommended.

Thickened forelegs. Connective tissue replaces muscle in the forelegs of afflicted pigs resulting in legs that appear thick and swollen. This should not be confused with callouses that result from skin irritation. A simple recessive mode of inheritance has been postulated, but supporting data are limited. If incidence is high and answers to the questions posed in the earlier section indicate a genetic cause, incidence can be reduced by culling affected pigs, their parents, and their littermates.

Other Disorders

Table 1 summarizes several other anatomical defects of swine. Those listed are of lower frequency, of less economic importance, or less clearly understood than those conditions described in the text. A comprehensive catalog of inherited disorders and abnormalities is available electronically on the Internet. It is accessible as Online Mendelian Inheritance in Animals (OMIA) at: http://www.angis.su.oz.au/ and at http://probe.nalusda.gov:8300/animal/omia.html.

Figure 6. Splayleg or spraddle leg pig.

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