CONGENITAL SPINAL AND VERTEBRAL MALFORMATIONS

BASICS

OVERVIEW
• Abnormal development of spinal structures, which are apparent at birth or within the first weeks of life
• “Congenital” refers to something that is present at birth; “spinal” refers to the spine; “vertebral” refers to the vertebrae
• The spine is composed of multiple bones (vertebrae) with disks (intervertebral disks) located in between adjacent bones; the disks act as shock absorbers and allow movement of the spine; the vertebrae are named according to their location—cervical vertebrae are located in the neck and are numbered as cervical vertebrae one through seven or C₁–C₇; thoracic vertebrae are located from the area of the shoulders to the end of the ribs and are numbered as thoracic vertebrae one through thirteen or T₁–T₁₃; lumbar vertebrae start at the end of the ribs and continue to the pelvis and are numbered as lumbar vertebrae one through seven or L₁–L₇; the remaining vertebrae are the sacral and coccygeal (tail) vertebrae
• “Vertebra” (plural, vertebrae) is the medical term for backbone

GENETICS
• A genetic background, with unknown mode of inheritance, is suspected in most congenital (present at birth) diseases of the spine
• Defective development of the sacral and coccygeal (tail) vertebrae (known as “sacrococcygeal dysgenesis”)—autosomal dominant
• Incomplete development of one side of a thoracic vertebra (known as “thoracic hemivertebra”) of German shorthaired pointers—autosomal recessive

SIGNALMENT/DESCRIPTION of ANIMAL
Species
• Dogs and cats
Breed Predilections
• Malformation of the bone at the back of the skull (known as the “occipital bone”), the first cervical vertebra (C₁; known as the “atlas”), and the second cervical vertebra (C₂; known as the “axis”)—most common in small-breed dogs
• Hemivertebra (incomplete development of one side of a vertebra), transitional vertebra (abnormal development of the backbone at the junction between two vertebral types, such as at the junction of the lumbar and sacral vertebrae), block vertebra (where two or more adjacent backbones are fused together), and butterfly vertebra (type of hemivertebra in which incomplete development of one side of the backbone causes a defect that looks like a butterfly on X-rays)—most common in short-nosed, flat-faced (known as “brachycephalic”), “screw-tailed” breeds (such as the French bulldog, English bulldog, pug, Boston terrier)
• Defective development of the sacral and coccygeal (tail) vertebrae (sacrococcygeal dysgenesis)—Manx cats
• Defective development of the spine leading to exposure of the covering of the spinal cord (known as “meninges”) or spinal cord (known as “spina bifida”)—bulldogs, Manx cats, and other screw-tailed breeds
• Abnormal development of the spinal cord (known as “myelodysplasia”)—Weimaraners
• Congenital (present at birth) narrowing of the spinal canal (known as “spinal stenosis”)—Doberman pinchers; breeds with “normal” short, bowed legs (known as “chondrodystrophic breeds”)

Mean Age Range
• Congenital (present at birth) malformations of the spine and/or backbones may cause clinical signs during the rapid growth of the animal (such as 5 to 9 months of age)
• Congenital (present at birth) spinal cord abnormalities cause clinical disease from the time of birth

SIGNS/OBSERVED CHANGES in the ANIMAL
• Distortion of the back or spine—lordosis (abnormal curvature that causes the spine to arch downwards, so called “sway back”), kyphosis (abnormal curvature that causes the spine to arch upwards); and scoliosis (abnormal curvature of the spine to one side) in cases of malformed backbones or vertebrae
• Incoordinated or “drunken” appearing gait or movement (known as “ataxia”) and weakness or partial paralysis (known as “paresis”) associated with squeezing or pressure on the spinal cord and trauma
• Signs vary with location of the spinal cord segment(s) involved

CAUSES
• Breed-related inherited defects are suspected for most congenital (present at birth) spinal abnormalities, although interactions between several genes and environmental factors (such as nutritional deficiencies) likely are involved

RISK FACTORS
• Compounds that cause abnormal development of the embryo (known as “teratogenic compounds”)
• Toxins
• Nutritional deficiencies
• Stress

TREATMENT

HEALTH CARE
• Depends on severity of nervous system deficits and related signs
• Outpatient—if animal is able to walk (known as being “ambulatory”)
• Inpatient—if animal is not able to walk (known as being “nonambulatory”) or requires emergency surgical treatment (such as for partial dislocation of the joint between the first and second cervical vertebra [condition known as “atlantoaxial subluxation”])
• Restricted activity combined with physical therapy—may help neurologically disabled patients in their postoperative period; a cart may be necessary for severely affected patients
• Management of urination may be essential for cases in which disorders of urination accompany the spinal abnormality

ACTIVITY
• Restricted, especially if partial dislocation of the vertebrae or backbones (known as “vertebral subluxation”) is present

SURGERY
• In general, surgery to relieve pressure on the spinal cord is required when congenital (present at birth) malformation(s) cause narrowing of the spinal canal and put pressure on the spinal cord; in cases of long-term (chronic) or widespread (diffuse) pressure on the spinal cord, therapeutic responses following surgery are minimal
• Partial dislocation of the joint between the first and second cervical vertebra (atlantoaxial subluxation)—surgery to relieve pressure on the spinal cord combined with stabilization of the atlantoaxial joint with pins or screws is the treatment of choice
• Defective development of the spine leading to exposure of the covering of the spinal cord (known as “meninges”) or spinal cord (spina bifida)—protrusion of the membranes around the spinal cord (meninges) through an opening in the spine (protruding membranes known as “meningoceles”) can be closed surgically to prevent leakage of cerebrospinal fluid and infections; surgery usually is not attempted when the spinal cord itself is involved

MEDICATIONS
Medications presented in this section are intended to provide general information about possible treatment. The treatment for a particular condition may evolve as medical advances are made; therefore, the medications should not be considered as all inclusive.

• Steroids may be used in some cases; response to steroid treatment has been variable

FOLLOW-UP CARE

PATIENT MONITORING
• Frequent (such as every 4 to 6 months) nervous system examinations—often required to monitor the progression of clinical signs
• X-rays—repeat as needed

PREVENTIONS AND AVOIDANCE
• Avoid breeding affected animals

POSSIBLE COMPLICATIONS
• Depend on the type and severity of nervous system signs
• Partial dislocation of the joint between the first and second cervical vertebra (atlantoaxial subluxation)—sudden (acute) death may occur
• Partial dislocation of the vertebrae or backbones (vertebral subluxation)—sudden (acute) paralysis can be seen with further trauma and pressure on the spinal cord
• Surgical implant (such as bone plates or pins) failure may be observed after surgery to relieve pressure on the spinal cord and to stabilize the spine

EXPECTED COURSE AND PROGNOSIS
• Prognosis varies depending on the type of malformation, degree of pressure on the spinal cord or injury, and surgical techniques to relieve pressure on the spinal cord and to stabilize the spine
• Congenital (present at birth) malformation of vertebrae without pressure on the spinal cord—prognosis is good
• Partial dislocation of the joint between the first and second cervical vertebra (atlantoaxial subluxation) following surgery to relieve pressure on the spinal cord and/or stabilization of the backbones—prognosis is fair to good
• Surgery to relieve pressure on the spinal cord—prognosis is fair
• Defective development of the spine leading to exposure of the covering of the spinal cord (known as “meninges”) or spinal cord (spina bifida) associated with spinal cord malformation; long-term (chronic) nervous system disease despite surgical treatment; and disease of the nerves that connect the spinal cord and muscles (known as “lower motor neuron disease”) with lack of control of urination (known as “incontinence”)—prognosis is poor
• Medical treatment usually is insufficient to alleviate moderate to severe nervous system signs caused by pressure on the spinal cord secondary to congenital (present at birth) malformation(s) of the vertebrae
• Many dogs and cats with nervous system signs that are not treated are euthanized

KEY POINTS
• Many congenital (present at birth) malformations of the vertebrae do not cause clinical signs
• A thorough diagnostic work-up should be performed when a congenital malformation results in nervous system abnormalities
• A genetic basis is suspected for malformations of the vertebrae; avoid breeding affected animals
• Early surgical intervention often is necessary to alleviate pressure on the spinal cord and to prevent further damage