Chapter 4
Overview and Comparison of Idiopathic, Neuromuscular, and Congenital Forms of Scoliosis

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Scoliosis is really a physical finding, a lateral curvature of the spine. There are a number of potential causes, not all of which are related to a primary spinal deformity. For instance, if a patient has one leg longer than another, they will display a curved spine; otherwise their trunk would leave the pelvis at an angle, causing the individual to always look like they are leaning to one side. In a similar way, if an individual leans their back to one side, they will also show a scoliosis on a radiograph. Radiographs are often used to make the diagnosis, but because of these "postural" causes, scoliosis is usually defined as a curve on a standing radiograph of the spine measuring at least 10° (Binstadt et al. 1978, Cobb 1958).

The causes of scoliosis can be broadly classified as congenital, neuromuscular, syndrome related, idiopathic, and due to a secondary cause (Beals 1973). Another way to think about this is that scoliosis can be caused by a primary problem related to the spine itself (idiopathic and congenital scoliosis) or can be secondary to an underlying more systemic disorder (neuromuscular or syndromic scoliosis) (Table 4.1).

Congenital scoliosis is due to a vertebral malformation during fetal development, which results in a deviation of the normal spinal alignment. Neurologic conditions can cause curves due to muscle weakness or muscle imbalance, with muscles on one side of the spine pulling laterally more than muscles on the other side of the spine. This includes conditions such as cerebral palsy, paralysis, and Duchenne muscular dystrophy. These curves often have a typical radiographic appearance, with a long curve over most of the spine, often in a "c" shape, so that the patient is made off balance by the curve. Syndromes that are not associated with a neuromuscular problem, but instead are associated with a structural defect in the connective tissues can also cause scoliosis. This is presumably because gene mutation causes the bone and soft tissues to weaken, resulting in skeletal deformity. Several syndromes fall into this category including Marfan syndrome, osteogenesis imperfecta, and Ehlers–Danlos.

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Table 4.1 Selected syndromes and neuromuscular conditions associated with scoliosis

Achondroplasia
Arthrogryposis
Cerebral palsy
Charcot–Marie–Tooth disease
Congenital hypotonia
Duchenne muscular dystrophy
Ehlers–Danlos syndrome
Marfan syndrome
Myelomeningocele
Neurofibromatosis
Osteogenesis imperfecta
Paralysis
Poliomyelitis
Spinal muscular atrophy

In Ehlers–Danlos and Marfan, laxity of the soft tissue ligaments connecting the vertebra is associated with scoliosis. Osteogenesis imperfecta is associated with both soft tissue laxity and also weak bones that easily deform, and this condition can also cause scoliosis (Raff and Byers 1996).

The cause of idiopathic scoliosis, as the name suggests, is not known, although there is a wealth of data showing a familial occurrence (Kouwenhoven and Castelein 2008). Indeed, linkage studies suggest a genetic etiology, and several scoliosis loci have been identified. Idiopathic scoliosis behaves clinically in different ways depending on the age at which the curve presents. As such, it has been subdivided into adolescent, juvenile, and infantile forms. Many cases of infantile scoliosis and a few cases of juvenile scoliosis will resolve on their own. For the other idiopathic scoliosis patients, as a general rule, the younger a child presents, the more severe the ultimate degree of scoliosis that will develop.

Routine screening for scoliosis is no longer recommended in most jurisdictions (Weinstein et al. 2008). As such, patients usually present with a noticeable spinal deformity or, more likely, chest wall and back asymmetry. Whether identified by the patient, their parents, or through identification by a primary caregiver, posterior chest wall prominence is the most outward manifestation of spinal curvature. Other body characteristics may include shoulder asymmetry and overall posture imbalance in the coronal plane.

Sometimes pain can cause an individual to stand with their back bent and cause a scoliosis. This includes painful conditions such as infection, secondary to trauma, or malignancy. Brain tumors, intraspinal tumors, boney tumors, and extra-spinal malignancies, such as a retroperitoneal tumors, can all present with scoliosis. As such, the first priority for a physician caring for a patient with scoliosis is to evaluate for such serious, potential life threatening conditions (Janicki and Alman 2007).

The two reasons for a physician to become involved in the care of an individual with scoliosis are to identify the cause and to treat potentially deleterious effects of the curve. The medical evaluation of a patient with scoliosis includes a medical history, physical examination, and appropriate diagnostic radiographic tests. Much

of the history and physical examination is focused on identifying more ominous causes of scoliosis. Young age at onset (younger than 10 years of age), rapid curve progression, and the presence of neurological symptoms are the most useful findings in identifying nonidiopathic causes of scoliosis.

Once scoliosis is suspected, spinal radiographs are usually obtained. These films are taken with a patient standing if possible. Curve magnitude is measured using a technique referred to as the Cobb method, in which the greatest angle between the end plates of the most tilted vertebra on each side of the apex of the curve is measured (Fig. 4.1). There is a high intra- and interobserver variability to this measurement. Studies show a 5° error of measurement in idiopathic cases, and studies report up to a 15° error of measurement in congenital cases. There are a number of clues that can be elucidated from the radiographs as to curve etiology. There should be two pedicles at every level, and absence of a pedicle suggests either a congenital, neoplastic, or infectious etiology. In idiopathic scoliosis, the apex of the curve usually points away from the heart, to the right, and there should be rotation of the spine, with the apex of the curve having the most rotation. Idiopathic scoliosis is not only a deformity in the coronal plane but also a rotational deformity. A scoliotic curve without rotation should be investigated for other causes. If back pain

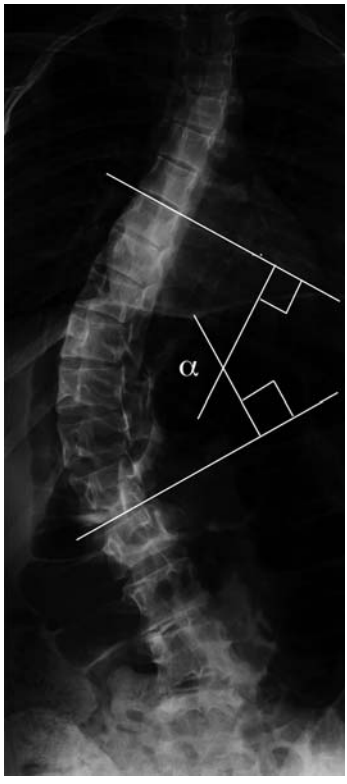

Fig. 4.1 Cobb angle (α) measurement on a standard anterior–posterior view spinal radiograph

is present, additional studies should be considered, such as a lateral radiograph of the spine including the lumbosacral region, to look for vertebral abnormalities associated with diagnoses such as spondylolysis (an idiopathic fracture of the posterior elements of the vertebral body), spondylolisthesis (a slipping forward of one vertebral body on the adjacent one), infection, or bony destruction. Further imaging for patients with scoliosis and back pain may include a bone scan or a magnetic resonance imaging (MRI) test. An MRI is not needed for the treatment of most patients with idiopathic scoliosis although some treating centers will request an MRI for any patient with scoliosis (Kotwicki 2008, Cassar-Pullicino and Eisenstein 2002, Carman et al. 1990, Morrissy et al. 1990).

In addition to the obvious differences in the etiology of the various types of scoliosis, each has differences in their clinical behavior and in the implications of spinal deformity (Howard et al. 2007, Mercado et al. 2007, Donaldson et al. 2007, Smith et al. 2006). Because radiographs are easy to observe, much of the literature on scoliosis outcome is based on the degree of curve progression. Despite this, there is a poor relationship between curve magnitude and the effect of scoliosis on an individual. Furthermore, depending on the location of a curve and an association with an underlying condition, curves of equal magnitude can have very different effects.

Several population studies have examined the natural history of curve progression in idiopathic, congenital, and neuromuscular scoliosis. These studies give an overall risk of progression in a population, but unfortunately, there is strong variability in how curves behave in individual patients, so that observation over time is still required to make treatment decision based on progression in individual patients. Further confounding issues of curve progression is the relatively poor reliability of measures of degree of scoliosis on radiographs. This means that one needs to measure at least a 5° change in the magnitude of the degree of curve to be considered a change (Carman et al. 1990, Morrissy et al. 1990).

Long-term studies of adolescent idiopathic scoliosis show little deleterious effect in most untreated patients. In patients with idiopathic scoliosis in whom surgery was recommended, but surgery was declined, there were little long-term differences in pain, physical function, general health, or occupation when compared to the general population. This makes it important to clearly define treatment goals in scoliosis. Indeed, these data provide the rationale for an ongoing prospective randomized trial of brace treatment versus no brace treatment for idiopathic scoliosis.

Severe curves centered in the thoracic spine can be associated with pulmonary function insufficiency, and changes in pulmonary function test results can be seen once a curve in the thoracic region exceeds 70°, and symptomatic changes are rare in curves measuring less than 90° (Barrios et al. 2005, Smyth et al. 1984, Weber et al. 1975). Some forms of congenital scoliosis are associated with more severe pulmonary compromise, and this is due to the overall short trunk height or to a severe curvature occurring in early infancy. Such deformities early in childhood can impede normal lung development, as there is insufficient space available for normal pulmonary development. In these cases there is often multiple rib abnormalities present which also contribute to the pulmonary insufficiency (Ramirez et al. 2007, Campbell et al. 2003). Patients with a neuromuscular etiology to their scoliosis often

have poor pulmonary function. It is, however, not clear if the poor pulmonary function is related to the spinal deformity or to the underlying weakness of the muscles controlling respiration (Finder et al. 2004). As such, pulmonary effects from scoliosis are primarily limited to large curves, which occur most commonly in either idiopathic scoliosis that present at a very early age or severe cases of congenital scoliosis.

Pain can occur in all forms of scoliosis, but it is not clear that the incidence of pain is greater than in the general population. While there are no population studies of pain in congenital scoliosis, this type of spinal deformity does not seem to be associated with a higher frequency of pain than idiopathic scoliosis. In some cases of neuromuscular scoliosis or syndromic scoliosis pain may be associated with spinal deformity, particularly because sitting and positioning may be difficult and associated with abnormal pressure on the skin in patients with poor muscle control. Indeed, this symptom is a major reason that children with neuromuscular scoliosis undergo surgery (Mercado et al. 2007).

One consequence of spinal deformity is cosmesis. In moderate or severe scoliosis, one can see that the effected individual has a spinal deformity. Interestingly, the degree of spinal deformity on radiographs does not correlate with cosmetic ratings (Donaldson et al. 2007, Smith et al. 2006). For many individuals with idiopathic or congenital scoliosis, cosmetic concerns are a major reason they seek intervention. For children with congenital scoliosis, involvement of fewer vertebra often has worse cosmetic effects than involvement of multiple vertebrae. The site of the congenital deformity is also important, as hemivertebrae near the top and bottom of the spine have more dramatics cosmetic effects than congenital deformities in the middle of the spine (Goldberg et al. 2002a, b).

Congenital scoliosis is due to skeletal abnormalities of the spine that are present at birth. These anomalies, which can include multiple levels, are the result of and broadly classified as a failure of formation or a failure of segmentation (or both) during vertebral development. Because these spinal deformities are present in utero, they may be first identified on fetal ultrasound. In addition, the underlying cause can be associated with abnormalities of other organ systems in up to 40% of cases (Arlet et al. 2003). Thus, it is important to identify associated anomalies with a thorough evaluation of the neurological, cardiovascular, and genitourinary systems (Arlet et al. 2003). In contrast, idiopathic scoliosis is not associated with a higher incidence of other organ malformations than in the normal population. Syndromic scoliosis can be associated with other anomalies depending on the cause. For instance, Marfan syndrome is associated with cardiovascular anomalies.

Treatment is based on the type, age of the patient, progression of the curve, the long-term implications of the spinal deformity, the location of the curve, and type of anomaly. Treatment can be divided into the broad categories of observation, bracing, and surgery. Observation is used for small curves that are unlikely to progress to the point of causing symptomatic problems. Bracing has been shown to be effective in slowing scoliosis severity progression in mild to moderate-sized idiopathic curves in skeletally immature children. It has not been demonstrated to alter curve progression in other forms of scoliosis, or in curves in skeletally mature individuals. Despite

this, bracing is sometimes used to hold the back in an overall straighter alignment. Surgery is used for more severe curves, and in most cases is spinal fusion using instrumentation to hold the back into a straighter alignment. While texts often list a degree of curvature as an indication for surgery, there are only relative, as opposed to absolute, indications for surgery. Natural history studies of patients with idiopathic scoliosis after skeletal maturity, found that curves less than 30° do not progress, while most curves of greater than 50° continue to progress. The progression is on average approximately 1° per year. These data are often used to suggest that curves greater than 50° require surgery as they will progress later in life. Not all curves actually progress (indeed almost a third of curves over 50° will not substantially increase in severity), and severe physiologic abnormalities have not been found in patients who had refused surgery with idiopathic scoliosis. In contrast severe curves, which are usually secondary to juvenile idiopathic scoliosis or congenital scoliosis can progress to a severity having deleterious physiologic effects and as such benefit from surgical intervention. Treatment for very young children with severe curves is rather problematic. In young children who still have substantial growth left, and for whom bracing is not an option, spinal fusion will shorten the length of the spine, and in the thoracic region this may decrease the space available for the lungs, resulting in impaired pulmonary function. In these individuals, non-fusion surgery is unusually advocated (Janicki and Alman 2007, Kim et al. 2009, Heary et al. 2008). This surgery involves either instrumentation for the spine without fusion or the use of instrumentation to distract the ribs. Non-fusion surgery techniques have less than ideal outcomes, and as such this is an area of intense current research into spine deformity management.

Scoliosis is a physical finding that can have a variety of etiologies. While there are some similarities between scoliosis of different etiologies, there are also substantial differences. As such, one needs to consider the etiology and natural history of the various forms of scoliosis to develop effective therapeutic approaches for patients with scoliosis.

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