Syndromic Scoliosis

Scoliosis is defined as a lateral curvature of the spine of greater than 10 degrees. Syndromic Scoliosis is a scoliosis associated with an underlying genetic syndrome such as Marfan Syndrome, Neurofibromatosis, Noonan Syndrome and many others.

Idiopathic Scoliosis is the most common form of scoliosis, but doesn't have a clearly defined underlying cause. In syndromic scoliosis, patients have an abnormal curvature in their spine which is due to an underlying condition affecting the growth and/or development of tissues in their bodies (dysplasia). There are a vast array of conditions that can lead to the development of scoliosis. These syndromes were traditionally diagnosed by typical clinical findings and physical features of patients; such as the short-limbed stature of people with Achondroplasia compared with the long slender limbs of people with Marfan Syndrome. With the emergence of the field of medical genetics, we are now able to test for a wide variety of conditions through genetic testing. The list of conditions that can be tested for continues to increase as we gain a better understanding of the human genome, and how it affects human development.

Many patients with syndromic scoliosis present with early onset scoliosis. Early onset means that the patient has a progressive curve before the age of 10 years or before the adolescent growth spurt. These are some of the most challenging cases of scoliosis to treat. Ongoing growth is needed for normal development of the body, including maturation of the chest wall and lungs. Unfortunately, this growth also drives the scoliosis and left untreated can result in severe spinal deformity. There are many treatment options for patients with early onset scoliosis with each having their own pros and cons.

It would not be realistic to put together a comprehensive list of every syndrome that could possible result in the development of scoliosis. Some syndromes, such as Muscular Dystrophy or Cerebral Palsy, are also considered more of a neuromuscular scoliosis rather than a syndromic scoliosis because of their disruption to the normal way that nerves and muscles function to control the alignment of the body. Below are brief descriptions of some of the more common syndromes that are associated with progressive spinal deformities in children.

Neurofibromatosis (NF)

Neurofibromatosis is the most common autosomal dominant inherited disorder in the world, with a prevalence of over 1 million cases and affects the growth of neural tissue. Spinal deformities are common with up to 35% of NF patients affected. Patients often have typical skin markings called café-au-lait spots. The spine deformities can be extremely variable with some patients having very mild curves that do not require treatment, while others can develop very severe stiff curves requiring treatment at a young age. Dystrophic scoliosis occurs in NF with a short, sharp severe curve often associated with a plexiform neurofibroma. Rarely cases of spinal subluxation and dislocation are seen in NF.

Marfan Syndrome

Marfan syndrome is a disease that affects the connective tissues of the body and specifically a

protein called fibrillin. People with Marfan syndrome typically have long slender limbs and hypermobile joints. Scoliosis develops in about 60% of people with Marfan syndrome, and has a broad spectrum from very mild curves that require no treatment to much more severe scoliosis. When spine deformities develop early in life, they can be relentlessly progressive and result in very severe curves. Scoliosis in Marfan syndrome is often resistant to brace treatment and may require surgical intervention at a young age. Because fibrillin is an important component of tissue throughout the body, Marfan syndrome affects much more than just bones and joints, with patients often having other medical issues such as cardiac valvular disease, aortic root dilatation, and ocular lens dislocation.

Osteogenesis Imperfecta (OI)

Osteogenesis Imperfecta (OI) is due to a genetic defect in the body's ability to make collagen, which is the primary protein building block for bones and joints in the human body. This results in bone that is extremely brittle and prone to fracture, hypermobile joints, and weak muscles. There are several types of OI based on the genetic abnormality and thus the degree to which collagen is affected. The more severe types have a greater impact on growth and development. Collagen is found throughout the body so many body systems can be affected by this disease. The rate of scoliosis found in individuals with OI is reported between 40% to upwards of 90% and depends on the type of OI. There is some evidence to suggest that some patients with OI may benefit from medical therapy with bisphosphonates to help slow down the rate of spinal curve progression, but it does not ‘cure’ the scoliosis.

Achondroplasia

Achondroplasia is the most common form of short stature. Patients with this have short arms and legs, but a normal height to their trunk. They often have an abnormal kyphosis (forward bend to the spine) rather than a scoliosis (side bend to the spine). Spinal deformity is present in over 90% of children with achondroplasia under the age of 1. This kyphosis usually improves once the child is able to start standing and walking (even if delayed); and significant deformity decreases to about 30% at age 3 and 10% of children at age 10. Occasionally bracing is needed to help support the spine during the early phases of growth. Patients typically also have a narrow spinal canal causing pinching of the nerves or spinal cord, which is called spinal stenosis. This can occur either high at the foramen magnum (where the skull meets the first cervical vertebra) or low down at the bottom of the lumbar spine (lower back).

Ehlers-Danlos Syndrome

Ehlers-Danlos Syndrome represents a group of conditions that affect connective tissues throughout the body. These patients usually present with hypermobile joints and ligamentous laxity. There are several sub-types of Ehlers-Danlos depending on the underlying genetic mutation that affects the severity of the disease and the body parts most affected. The overall rate of scoliosis in this population is about 30% with some sub-types being over 50%. About 10% of patients will require surgery for their scoliosis. Back or neck pain in patients with hypermobility can be a common presentation, and often responds to strength training. Other joints can be affected, typically the knee cap (patellofemoral joint), fingers and toes, and patients will have varying degrees of flexible flat feet (pes planus).

References:

Anissipour AK, Hammerberg KW, Caudill A, et al. Behavior of Scoliosis During Growth of Children with Osteogenesis Imperfecta. J Bone Joint Surg Am, 2014; 96-A(3): 237-43.

Crawford AH, Herrera-Soto J. Scoliosis Associated with Neurofibromatosis. Ortho Clinics North America, 2007; 38(4): 553-62.

Crawford AH, Parikh S, Schorry EK, et al. The Immature Spine in Type-1 Neurofibromatosis. Jl Bone Joint Surg – Am, 2007; 89-A (Suppl 1): 123-42.

Misra SN, Morgan HW. Thoracolumbar spinal deformity in achondroplasia. Neurosurg Focus, 2003; 14(1): 1-8.

Sponseller PD, Bhimani M, Solacoff D, et al. Results of Brace Treatment of Scoliosis in Marfan Syndrome. Spine, 2000; 25(18): 2350-4.

Sponseller PD, Sethi N, Cameron D, et al. Infantile Scoliosis in Marfan Syndrome. Spine, 1997; 22(5): 509-16.

Sponseller PD, Thompson GH, Akbarnia BA, et al. Growing Rods for Infantile Scoliosis in Marfan Syndrome. Spine, 2009; 34(16): 1711-15.

Stanitski DF, Nadjarian R, Stanitski CL, et al. Orthopaedic Manifestations of Ehlers-Danlos Syndrome. Clin Orthop Related Research, 2000; 376: 213-21.