



National Marfan Foundation

Marfan Syndrome

Overview of Orthopedic Management

Skeletal manifestations in the Marfan patient may develop or become more pronounced with age. Although not life-threatening, they can cause decreased mobility and chronic pain, thereby creating significant impact on the quality of life. This is becoming more and more evident as advances in cardiovascular surgery are extending life expectancy to nearly that of the general population.

MAJOR SKELETAL MANIFESTATIONS

- Scoliosis over 20 degrees by upright radiographs
- Spondylolisthesis
- Protrusio acetabulae by anteroposterior hip x-ray
- Severe hindfoot valgus, also noted as medial deviation of medial malleolus (by clinical finding, not imaging)
- Increased extremity to trunk ratio (armspan to height >1.05)
- Arachnodactyly as diagnosed by positive thumb and wrist signs. The thumb sign is when the entire distal phalanx of the thumb protrudes beyond the ulnar border of the clenched fist. The wrist sign is when the thumb can cover the entire fifth fingernail when wrapped around the contralateral wrist
- Dural ectasia (MRI preferred for evaluation)
- Sternal deformities (pectus carinatum or severe excavatum)
- Elbow flexion contracture (by clinical diagnosis; at least 10 degrees from full extension of the elbow)

MINOR MANIFESTATIONS MAY INCLUDE

- Distinctive craniofacial features, such as a long narrow cranium, downslanting palpebral fissures, high arched palate
- Joint laxity

OTHER FEATURES NOT IN THE 1996 GHENT CRITERIA

- Limb length inequality greater than 2 cm difference by clinical or radiographic measure
- Excessive kyphosis; thoracolumbar kyphosis by standing lateral spine x-rays
- Distinctive spinal features including thinned pedicles, vertebral scalloping and elongated transverse processes
- Hip dislocation in infants
- Osteopenia by DEXA scan
- Musculoskeletal pain and fatigue

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MANAGEMENT GUIDELINES

ON-GOING SCREENING

All growing children with Marfan syndrome require annual screening for scoliosis, kyphosis, and spondylolisthesis by physical examination performed by the primary care physician. The examination should include:

- Standing examination for kyphosis
- Forward bend test to detect asymmetry of any portion of trunk
- Palpation for bony step-off

If abnormality is found follow-up with:

- Standing posteroanterior and lateral radiographs
- Lateral radiograph of the lumbosacral spine for spondylolisthesis
- Or referral to orthopedic surgeon for further evaluation

All growing children with Marfan syndrome require screening at age 9-12 for leg length inequality.

- Palpate pelvis in standing position
- Note: “False-positive” may occur in cases of scoliosis
- Refer to orthopedic surgeon if significant difference is seen (1/2 inch, or 1.2 cm)

SCOLIOSIS

Medical Management

- Patients with scoliosis should be referred to a spine deformity expert for management
- Use of scoliosis bracing
 - ◆ No clear documentation of efficacy. Bracing may be appropriate for a subset of growing children, i.e. consider for curves of 15-30 degrees
 - ◆ No role for bracing after skeletal maturity
 - ◆ Low-yield for curves over 35-40 degrees
 - ◆ Curves over 40 degrees are likely to progress over time. Bracing is of little or no value in these cases. Consider use on an individual basis.

Surgical Management

- Surgery is an appropriate option for curves over 45 degrees
- Growing rods are a successful option for curves over 50-60 degrees in patients who are less than 9 years old
- For older children, between 9 years and skeletal maturity, may defer surgery until ~60 degrees to allow further growth
- Prior to surgery, obtain MRI or CT to assess dural ectasia and pedicle thinning
- Prior to surgery, obtain cardiology clearance within past 6 months

- Surgery should include all major curves; selective fusion is less successful than in idiopathic scoliosis
- Be aware of increased risk for blood loss, cerebrospinal fluid leak and failure of fixation
- Posterior approach is currently the preferred approach for most scoliosis surgery.

KYPHOSIS

Medical Management

- Patients with excessive kyphosis should be referred to a spine deformity expert for management
- Physical therapy/exercises are appropriate for mild cases. (Note: There is no clinical research in Marfan syndrome patients.)
- No evidence of efficacy of kyphosis bracing, but is an option to try if under 70 degrees

Surgical Management

- Spinal fusion is an option for curves over ~70 degrees in the thoracic spine and “significant” deformity of the thoracolumbar region

BACK PAIN

- Back pain is more common in Marfan syndrome than in the general population
- Causes include scoliosis, disc abnormalities, dural ectasia, spondylolisthesis
 - ◆ Consider MRI to evaluate for dural ectasia, a frequently seen feature of Marfan syndrome.
- Refer to orthopedic surgeon or rehabilitation specialist
- Treatment options do not differ significantly from general population

MARFAN FEET

- Problems include severe flat feet, foot pain, weakness of foot/ankle mechanics, significant calluses, bunions, and toe deformities
- Orthotic/brace treatment does not change growth of foot or create higher arches.
- Orthotics may be useful if pain is present. Custom-made soft orthotics may be needed to assure the proper fit essential for effective treatment
- Ankle weakness may respond to use of ankle-foot orthoses (AFO's). Evaluate for proper type to use
- Surgery is indicated only if foot deformities and pain do not respond to conservative treatment
- There is no proven success using artificial ankles or toe joints in the management of Marfan feet

PROTRUSIO ACETABULAE/HIP PAIN

- Although protrusion is common in Marfan patients, most people with MFS are not at risk for hip arthritis
- Protrusio accelerates degeneration only if extreme (Center-edge angle >50 degrees before age 11)
- Consider triradiate epiphyseodesis only if this degree of protrusion is seen before age 11

PECTUS EXCAVATUM

- May displace heart to left and limit exercise tolerance
- Considered significant if depression exceeds 2.5cm, or if the ratio of chest width to sterno-vertebral distance (Haller index) exceeds 3.2
- Surgery may be done via open or minimally-invasive approaches
 - ◆ Open approach (Ravitch procedure) consists of detaching rib cartilages and osteotomizing sternum, elevating it over a bar
 - ◆ Minimally invasive approach (Nuss procedure) consists of elevating sternum with a curved bar attached to ribs on each side. Bar is left in place for 2-3 years to promote remodeling of chest wall
- Optimal time for pectus excavatum surgery is in the early teen years
- Nuss procedure may be combined with cardiac surgical procedure in carefully selected situations
- Pectus excavatum may be corrected after prior median sternotomy

PECTUS CARINATUM

- May cause symptoms from pressure or impact
- Some patients may benefit from treatment for self-image concerns/personal preference
- Brace treatment in juvenile age is investigational
- Surgical treatment for severe cases may be offered at or near skeletal maturity. It involves rib cartilage detachment and osteotomy.

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