PCP Handout

Caring for Persons with Marfan Syndrome

Marfan syndrome is a disorder of connective tissue, most commonly affecting the ocular, musculoskeletal, and cardiovascular systems, including the risk of aortic aneurysms or dissections. Signs and symptoms vary widely in severity, timing of onset, and rate of progression. Classic body habitus features are tall and slender, elongated fingers and toes, and arm span that exceeds body height. Incidence is ~1 in 5,000 worldwide, autosomal dominant inheritance in 75%, new mutations in 25% of FBN l gene on chromosome 15 which makes the protein, fibrillin-1. Life expectancy similar to general population.

System	Information	Follow up
Dental	▪ Crowded teeth, high palate	▪ Routine dental care
Endocrine	▪ Taller than mean parent height, 40% have inc. leg to trunk or arm span to height, inc. osteopenia	▪ Use Dx specific growth curves
Eye	▪ Significant myopia, lens ectopis/ dislocation 72%, retinal detachment 36%, glaucoma 15%, early cataracts	▪ Annual eye exam, avoid Lasik
Face	▪ "Classic" - deep set eyes, long narrow face, down slanting eyes, malar hypoplasia, micrognathia	
Cardio-vascular	▪ Thoracic aorta dilation, esp. at Ao root 60-80%, abd aorta dilation 10% esp. as adult, aortic valve regurgitation, risk of dissection esp. as adult, coronary artery aneurysm reported in adults ▪ Mitral valve prolapse 52-68%, regurgitation, enlarged pulmonary art root 43-74%, atrial septal defect 4% ▪ Diastolic dysfunction nearly 100%, CHF ▪ Inc. arrhythmia – A fib 8%, ventricular dysrhythmias, abn repolarization QT lengthen, STdepress, axis shifts	▪ Semi to annual echo for aortic size, z-score greater >=2 , inc. screen in pregnancy ▪ Add screen of whole aorta in late teens ▪ Beta blockers and/or ACE/ARB ▪ Surgery if adult ao > 5 cm ▪ Avoid contact sports, isometrics, caffeine, decongestants, caution w/ ADHD stimulants ▪ Increased anesthesia risk
GI	▪ Esophageal dilation , achalasia, diaphragmatic hernia, diverticulosis, renal/liver cysts, abd pain, IBS, constipation, rarer volvulus	▪ Evaluate for sx
GU	▪ Fertility unaffected, 50% transmission , ▪ High risk pregnancy, increased risk aortic dissection	▪ Genetics and perinatology consultations
Musculo-skeletal	▪ Hyperextensible joints, muscle fatigue, overuse injury, increased spondylolithesis ▪ Pectus excavatum / carinatum 60%, scoliosis 18%, kyphosis, atlantoaxial translation 54%, basilar impression 36%, cervical kyphosis 14% - some neck pain, headaches ▪ Pes planus 60%, medial malleolar medial deviation ▪ Protrusio acetabulae of hip 14%, pain, early osteoarthritis	▪ Nuss procedure substernal plate for extreme p. excavatum heart/lung compression ▪ Scoliosis screen during growth, > 45 degrees consider correction ▪ Ankle orthoses as needed ▪ Consider severe hip protrusion surgery
Neurology	▪ Dural ectasia 90% (pouching of dura), lumbosacral, most asx, can cause bone erosion, nerve entrapment ▪ Pain in back, abdomen, legs, head	▪ CT/MRI for symptoms, orthopedic,neurosurgery as needed ▪ Risk CSF leak post epidural/spontaneous ▪ Avoid triptans vasoconstriction
Pulmonary	▪ Throat laxity, increased sleep apnea - obstructive, central and mixed ▪ Lung bullae 5-15%, spontaneous pneumothorax, restrictive disease from pectus/ scoliosis/ kyphosis with exercise intolerance	▪ Screen for apnea symptoms ▪ Avoid smoking, scuba diving, consider inc. risk of wind instruments ▪ PFTs for pulmonary complaints
Skin	▪ Skin striae, hernias with recurrences	▪ Hernia mesh often needed

References: Tinkle BT, et al. Health Supervision for Children and Adolescents with Marfan Syndrome. Pediatrics Apr 2023.

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Center for Youth and Adults with Conditions of Childhood (CYACC)