Special Needs Corner

Marfan's Syndrome

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Marfan's Syndrome is an autosomal dominant genetic disorder impacting connective tissue. The mutation alters the manufacture of functional fibrillin-1. A decrease of FN1 leads to an associated decrease of microfibril formation, an increase in growth factors and a decrease in elasticity which results in instability of the tissues impacted. Marfan's affects approximately 1 in 5000 individuals with an estimated 50% transmission to children, however about 25% of all cases are new mutations (i.e. neither parent diagnosed with MFS).[1] Symptomology can include the following observed characteristics: long arms, fingers and legs; generalized tall and thin individuals, pectus excavatum or carinatum, flexible joints, flat feet, crowded teeth or scoliosis. Systems impacted by Marfan's include cardiovascular,

ocular, pulmonary and dural swelling, among others. The two predominant issues for these patients typically are cardiovascular and ocular, with potential for aortic dissection and myopia, glaucoma, early cataract formation and dislocation of the retina and/or the lens. The condition can be fatal early in life, however general life expectancy can be middle to late adulthood.[2]

Diagnosis of the disorder in adults is done using the Ghent criteria, however in children, it is not a strong diagnostic test and the Kids-Short Marfan survey[3] (FIGURE 1) is considered to be more appropriate. Key factors in the diagnosis include family history, dilatation of large arteries (specifically the aorta and pulmonary artery), dislocation of

FIGURE 1. Kid- Short Marfan Score (Kid SMS) (adapted from Mueller et al 2013)

Revised Ghent Criteria (skeletal signs)

Wrist AND/OR thumb sign   Pectus carinatum/excavatum deformity
Hindfoot deformity   Pneumothorax
Dural ectasia   Protrusio acetabuli
Reduced US/LS AND increased

Required manifestations	Risk category for likelihood of MFS
Dilatation of aortic root + Ectopia lentis	Very high risk
• dilatation of aortic root	High risk (Complete examination of all symptoms of the revised Ghent Criteria is strictly recommended as soon as possible. Patient should see Marfan Syndrome specialists)
+ mitral valve prolapse + tricuspid valve prolapse	
or	
+ dilatation of pulmonary artery or	
+ at least 3 skeletal features of the systemic score of the revised Ghent criteria	
• Ectopia lentis	
+ mitral valve prolapse + tricuspid valve prolapse	
or	
+ dilatation of pulmonary artery	
Family history dilatation of aortic root	Moderate risk (Patient needs to be verified or excluded with further diagnostic procedures other than or echocardiography and clinical examination)

arm/height AND no severe scoliosis
Scoliosis or thoracolumbar kyphosis   Reduced elbow extension
Facial features   (dolichocephaly, enophtalmos, downslanting palpebral fissures, malar hyoplasia, retrognathia)
Skin striae   Myopia > 3 diopters
Mitral valve prolapse (all types)

the lens and skeletal anomalies. Diagnosis in a child can be very challenging, as several of the key symptoms are more age dependent and may not be noted a younger patient. However, should a diagnosis of Marfan's be suspected, genetic testing and follow up with specialists is imperative.[4]

There currently exist no chiropractic care models for Marfan's syndrome; there is a single case report of care to a young adult following a motor vehicle accident with function being returned following whiplash within four visits.[5] While Marfan's presents a complicated case picture, there are avenues for chiropractic care to assist overall quality of life. Adjusting techniques may need to be altered to accommodate for increased flexibility and hyper mobility. Several of the key skeletal features would be expected to either respond or at least allow for care to have some palliative effect. These would include scoliotic curves, rib restriction and subluxation secondary to sternal deformity and pes planus. Kinesiotaping or similar techniques also could be applied in areas where the skin is not overly fragile to assist with breathing mechanisms, scoliotic correction and normalization of joint motion. Craniosacral therapy may also prove to be of benefit to patients especially around dural swelling, crowding in the mouth or an elevated palate.

Contraindication do exist in this disorder, both absolute and otherwise. A provider needs to be fully aware of the potential or existence of cardiovascular complications, including but not limited to aortic aneurysms, bronchioectasis and ocular symptoms of lens dislocation and retinal detachment. Technique can be modified to accommodate for a patient's mobility, hyper or restricted, as some joints may develop contracture over time. Care should be provided with full communication with the health team that your patient has developed; if they are beginning their journey, we can assist them in finding literate providers who have expertise with Marfan's Syndrome.

For further information and resources, the patient, parent or provider can visit www.marfan.org .[6]

References:

1. http://rarediseases.org/rare-diseases/marfan-syndrome/

2. Cattalini M, Khubchandani R, Cimaz R. When flexibility is not necessarily a virtue: a review of hypermobility syndromes and chronic or recurrent musculoskeletal pain in children. *Pediatric Rheumatology Online Journal*. 2015;13:40.

3. Mueller, G. C., Stark, V., Steiner, K., Weil, J., von Kodolitsch, Y. and Mir, T. S., The Kid-Short Marfan Score (Kid-SMS) — an easy executable risk score for suspected paediatric patients with Marfan syndrome. *Acta Paediatrica*. 2013;102: e84—e89.

4. http://ghr.nlm.nih.gov/condition/marfan-syndrome .

5. Tuling JR, Crowther ET, McCord P. Clinical considerations in the chiropractic management of the patient with Marfan syndrome. *J Manipulative Physio Therapy*. 2000 Sep;23(7):498-502.

6. www.marfan.org .

PANDAS : A Synopsis

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PANDAS is an abbreviation for Pediatric Autoimmune Neuropsychiatric Disorders Associated with Streptococcal Infections. This term is used to describe children and adolescents who have OCD (obsessive compulsive disorder) and /or tic disorders that follow or worsen following streptococcal infections.[2]

Most commonly, OCD and tic symptoms occur gradually over time, but in PANDAS, the symptom onset is unusually abrupt. Often, in PANDAS, the symptoms are noted to occur "out of the blue and overnight" and usually reach full intensity in 24 to 48 hours.[3] These OCD symptoms were usually accompanied by other neuropsychiatric symptoms such as anxiety attacks, extreme mood swings, hyperactivity, and difficulty with school subjects.[3] These symptoms usually occurred following a strong stimulant to the immune system such as a viral or bacterial infection.[2]

Because these symptoms have been noted to increase with intensity following an untreated or partially treated streptococcal infection, the term "pseudo-autoimmune" reaction is used to describe the immune response. This term is used to indicate that the immune system is not really abnormal but causes harmful effects between the anti-strep response and the body's own tissues.[2] This can be attributed to the ability of the strep bacteria to 'hide' from the immune system for long periods through mimicry of the host tissue. When the immune system finally recognizes the foreign strep, it makes antibodies that not only react to the strep molecules, but with the host tissue that was mimicked. These 'cross-re-

active' antibodies trigger an immune response that attacks the host's own tissues such as brain tissue, which can lead to the OCD and other neuropsychiatric symptoms.[2]

Diagnosis of PANDAS uses the criteria of: 1. Presence of clinically significant obsessions, compulsions and/or tics; 2. Unusually abrupt onset of symptoms or a relapse-remitting course of symptom severity. This means that during the initial and subsequent occurrences, the OCD symptoms can be plotted to show a worsening immediately following a strep infection.[2] The OCD symptoms usually decrease significantly and occasionally completely resolve between episodes. Proper testing for streptococcal bacteria is essential to ensure a proper diagnosis.

The best treatment for acute episodes of PANDAS is to eliminate the strep infection causing the symptoms.[1] If a throat culture for strep is positive, a single course of antibiotics will usually get rid of the strep infection and allow PANDAS symptoms to subside. Preventative measures that can be taken are to sterilize or replace things such as toothbrushes or to evaluate family members as to if they are 'strep carriers' so that all sources of the strep infections can be noted and potentially remediated. Prophylactic antibiotics have been shown to be effective to avoid future PAN-DAS episodes as well.[2]

To date, there have been no studies directly involving chiropractic care and PANDAS. Because of the relationship the immune system plays in this disease though, it can be postulated that since chiropractic has been shown to increase more natural function of the systems of the body, it may benefit those suffering from PANDAS through a better immune response. This is of course hypothetical and needs to be studied in order to make any direct relation with chiropractic as a treatment for PANDAS, but with the impressive anecdotal evidence from chiropractic and other neuropsychological maladies, it is definitely a worthwhile treatment in conjunction with treatment for active or latent bacterial infection.

References

1. The National Institute of Mental Health: www.nimh.nih.gov/labs-at-nimh/research-areas/clinics-and-labs/pdnb/web.shtml

2. The National Institute of Health: http://www.nimh.nih.gov/health/publications/pandas/index.shtml

3. Murphy TK, Storch EA, Lewin AB, Edge PJ, Goodman WK. Clinical factors associated with pediatric autoimmune neuropsychiatric disorder associated with streptococcal infections. *Journal of Pediatrics* 2012 Feb; 160 (2):314-9.