Overview of the medical and research literature of Marfan Syndrome in 2016-2017

This is a short overview of what has been going on in the research field during the past year. This list is not exhaustive and does not include case reports (report concerning one single patient). If you want a more detailed summary and access to the article please visit Pubmed (https://www.ncbi.nlm.nih.gov/pubmed). If you have questions you can contact me at l.janssen@marfan.be and I'll do my best to help you.

Cardiology


➢ “Abnormal heart rate recovery and deficient chronotropic response after submaximal exercise in young Marfan syndrome patients.” Peres P, Carvalho AC, Perez AB, Medeiros WM. Cardiol Young. 2016 Oct;26(7):1274-81


“Mitral valve prolapse and Marfan syndrome.” Thacoor A. Congenit Heart Dis. 2017 Jun 5. Review

Ophtalmology


“Comparative data on SD-OCT for the retinal nerve fiber layer and retinal macular thickness in a large cohort with Marfan syndrome.” Xu W, Kurup SP, Fawzi AA, Durbin MK, Maumenee IH, Mets MB. Ophthalmic Genet. 2017 Jan-Feb;38(1):34-38. → Study showing a reduced thickness of the fibrous layer of the optic nerve in Marfan patients.


Orthopedics

Dermatology


Endocrinology and Metabolism


Dural Ectasia


Obstetrics


➢ “Birth characteristics of women with Marfan syndrome, obstetric and neonatal outcomes of their pregnancies-A nationwide cohort and case-control study.” Kernell K, Sydsjö G, Bladh M, Joseffson A. Eur J Obstet Gynecol Reprod Biol. 2017 Aug;215:106-111. → Swedish study showing that Marfan woman are more likely to have prematured or small compared to age babies and delivery by c-section.

➢ “Sex, pregnancy and aortic disease in Marfan syndrome.” Renard M, Muiño-Mosquera L, Manalo EC, Tufa S, Carlson EJ, Keene DR, De Backer J, Sakai LY. PLoS One. 2017 Jul 14;12(7):e0181166. → Preliminary study comparing the aortic root diameters of a man group, a pregnant group, and non-pregnant woman group, showing an increased dilatation in the man and pregnant group, possibly showing a protective effect of estrogen in women (as 17β-estradiol is known to activate the production of fibrillin-1 in vascular smooth muscle cells in the aorta)

Genetic-phenotype


Éditorial concernant le même sujet que l’article précédent. “Genotype-phenotype correlations in Marfan syndrome.” Landis BJ, Veldtman GR, Ware SM. Heart. 2017 Jun 8.


“Associations of Age and Sex With Marfan Phenotype: The National Heart, Lung, and Blood Institute GenTAC (Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions) Registry.” Roman MJ, Devereux RB, Preiss LR, Asch FM, Eagle KA, Holmes KW, LeMaire SA, Maslen CL, Milewicz DM, Morris SA, Prakash SK, Pyeritz RE, Ravekes WJ, Shohet RV, Song HK, Weinsaft JW; GenTAC Investigators. Circ Cardiovasc Genet. 2017 Jun;10(3). Study showing that pulmonary, squeletal and aortic signs are more present in adult Marfan patients then in children, but not the other signs.

Cell and molecular research


Pain and quality of life


“Characterization of pain, disability, and psychological burden in Marfan syndrome.” Speed TJ, Mathur VA, Hand M, Christensen B, Sponseller PD, Williams KA, Campbell CM. Am J Med Genet A. 2016 Nov 14. Study on 245 Marfan patients, showing that 89% of the patients complain of pain, from which 28% are Marfan related pains, resulting in a psychological burden.

“Marfan Syndrome and Quality of Life in the GenTAC Registry.” Goldfinger JZ, Preiss LR, Devereux RB, Roman MJ, Hendershot TP, Kroner BL, Eagle KA; GenTAC Registry Consortium. J Am Coll Cardiol. 2017 Jun 13;69(23):2821-2830. American study showing that Marfan patients have a lower quality of life than a control group, but the perception of the quality of life is linked to socioeconomical factors and not to general health or severity of MFS.


General research