

Children with Marfan syndrome

Diagnostics and treatment

Why is this important?

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Children of parents with Marfan – diagnostic options

- * Early testing of all children – if the parents agree
- * Testing of children who have symptoms/ look marfanoid
- * Wait until the children are older (and can speak their minds)
 - * Annual echocardiograms
 - * Annual eye examinations
- * Several families we know have been told that they don't need to worry about cardiovascular exams or genetic testing until 10-12 years of age
- * This can be – and is - forgotten

Diagnostics in children with healthy parents

- * Clinical suspicion
 - * Skeletal features
 - * Ocular problems
 - * Other findings (hernia, pneumothorax...)
- * Sometimes family history
- * Knowledge among doctors is key
- * Impression: More children seem to get an early diagnosis

Risks related to delayed diagnosis

- * Irreversible visual problems, amblyopia
- * Aortic dissection
- * Late medication, earlier aortic surgery
- * Medicalisation of healthy children
- * Worried parents – and grandparents
 - * Some parents are not informed well enough and hence not worried

Early follow up and treatment

- * Regular echocardiograms, MRIs later
- * Regular eye exams
- * Beta blockers or losartan
 - * Immediately?
 - * At a certain Z score?
- * Prophylactic aortic surgery if needed
- * Glasses or eye surgery
- * Orthopedic footwear, physiotherapy, adjustments at school...

Case 1

- * 7-year-old girl
- * Marfan diagnosis 5 years old in 2014
- * Parents satisfied with the follow up at the local hospital
- * The family had not come to see us

Case 1

- * Summer 2015: Sinus valsalva 3 cm, Z-score 3,7
- * Plan: next controll 2 years later, no medication
- * Winter 2016: Surgery for pectus excavatum
- * Child cardiologist Oslo University Hospital: Beta-blocker, new echocardiogram in the fall

- * The parents don't always know what good follow up is

Case 2

- * 13-year-old girl
- * Mother and several other family members have Marfan syndrome
- * The mother had aortic surgery during her last pregnancy
- * The girl looks like her mother - who does not look marfanoid
- * The family had not received any information regarding genetic testing of the children
- * The girl had never had an echocardiogram