:: Marfan Syndrome

- This document is a translation of the French recommendations drafted by Prof. Guillaume Jondeau, reviewed and published by Orphanet in 2007.
- Some of the procedures mentioned, particularly drug treatments, may not be validated in the country where you practice.

Synonyms:
Marfan disease

Definition:
Marfan disease is an autosomal dominant genetic condition that is usually secondary to an anomaly of fibrillin type 1 and is clinically linked variably with skeletal (large size, arachnodactyly, scoliosis....), ophthalmologic (ectopic lens), cardiac (aortic dilation or dissection, mitral valve prolapse), cutaneous (striae), pulmonary (pneumothorax) signs.....

Further information:
See the Orphanet abstract

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Pre-hospital emergency care recommendations
Call for a patient suffering from Marfan Syndrome

Synonyms

- Marfan disease

Aetiology

- structural anomaly of fibrillin 1 (tissue protein)

Special risks in an emergency

- aortic dissection
- spontaneous pneumothorax
- retinal detachment

Frequently used long term treatments

- beta-blockers
- anticoagulants

Complications

- be suspicious of any thoracic pain
- be suspicious of any acute dyspnoea

Specific medical care prior to hospitalisation

- maintain systolic arterial blood pressure < 130mmHg (beta-blockers, calcium-channel blocking agents)
- avoid large volume changes

Recommendations for hospital emergency departments

Emergency situations

A. Emergencies related to Marfan syndrome

1. Aortic dissection

An aortic dissection should be suspected every time a patient presents with intense thoracic pain or one of its complications: tamponade, haemorrhagic shock.

- **Immediate diagnostic measures:**
  - **Imaging technique**:
    - trans-thoracic ultra sound and, if necessary, trans-oesophageal
      or: scanner
      or: MRI
      The technique to use is that which is available and that which is most familiar to the team.

> - The MRI is contra-indicated if a pacemaker is present or if a Harrington rod has been used for surgical treatment of scoliosis.

- **Immediate therapeutic measures:**
  - **Maintain arterial blood pressure < 130 mmHg** using beta-blockers and vasodilators such as nitroprusside or calcium-channel blocking agents.
  - The persistence of thoracic pain and its progression indicates that the dissection is extending.
  - **Once a diagnosis of aortic dissection has been confirmed, the patient should be transferred to the cardiac surgery unit** as an emergency **for a dissection of the ascending aorta** (emergency transfer by a medical team if necessary), **or closely monitor under medical treatment if the dissection does not involve the ascending aorta.**

2. Pneumothorax

Suspect it if there is thoracic pain preventing deep respiration and which is associated with dyspnoea.

- **Immediate diagnostic measures:**
  - Pulmonary radiography
  - Thoracic scan if there are any doubts.
- **Immediate therapeutic measures:**
  - Depending on how well tolerated:
    - simple observation or
    - drainage of the pneumothorax
  - Transfer to intensive care unit or pneumology department depending on what is available locally.

3. Retinal detachment

Retinal detachment is an ophthalmic emergency. It occurs more frequently in myopic patients. Retinal detachment is usually unilateral.

- **Immediate diagnostic measures:**
  - Look for functional signs of retinal tearing:

- myodesopsia and phosphene (light flashes).
- localised loss of field of vision, loss of visual acuity (if the macula is involved), central scotoma or total blindness.
- Note: there is never any ocular pain or redness in these cases (unless there is a concomitant problem).

- If functional signs are present: urgent ophthalmic examination, the retinal detachment can extend very quickly and jeopardise the sight in a relatively short time.

- **Immediate therapeutic measures:**
  - Surgical (ophthalmologic) treatment

B. Emergencies unrelated to Marfan syndrome:

4. Aortic dilation

This is an important problem that justifies:

- Asking the patient questions about his/her aortic diameter.
- Measuring this diameter, by trans-thoracic ultrasound usually, if the data is old and the situation quite urgent.
- Avoiding sudden blood pressure changes when taking care of the patient in so far as this is possible (avoid large volume changes, hypertensive pressures).
- Ask the patient if he/she has any mechanical prosthesis, which indicates continuous anticoagulant therapy and could contra-indicate a MRI.
- Check that there is no valvular leaking which could require preventive measures for Osler's endocarditis.

5. Parturition

- **Avoid sudden blood pressure changes during parturition whatever the method used.**
- **A caesarean section is preferable when aortic dilation is greater than 40 mm**, vaginal delivery is possible below this value.

Drug interactions

- Take into account the patient's treatments which are usually:
  - beta-blockers
  - anticoagulants

Anesthesia

- Problems related to epidural anaesthesia:
  - technical problems due to spinal deformity
  - blood pressure problems to be avoided as indicated above
  - incomplete (unilateral) anaesthesia

Additional therapeutic measures and hospitalisation

- adjust the bed for large sized patients.
- take into account of their sensitivity to cold (Raynaud's phenomenon).
- provide the patient with post-operative information if a mechanical valve is inserted.
- take into account of the spinal pain that often occurs after sternotomy.

Organ donation

- **Organ donation is contraindicated** with this disease

Documentary resources

- Erbel R, Alfonso F, Boileau C, Dirsch O, Eber B, Harverich A, Rakowski H, Struyven J, Radegran K, Sechtem U, Taylor J, Zollikofer C, Klein WW, Mulder B, Providencia LA . Diagnosis and management of aortic dissection. Eur Heart J. 2001;22:1642-81.
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- Judge DP, Dietz HC. Marfan's syndrome. Lancet. 2005;366:1965-76.

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