

The Dutch guidelines for the diagnosis and management of Marfan syndrome: between evidence and common sense

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Introduction

Because Marfan syndrome is relatively rare, five expert centres (Marfan clinics) have been established in the Netherlands. These centers provide Marfan syndrome diagnostics when a diagnosis is suspected on clinical grounds and ensure adequate clinical assessment and treatment. While the Marfan clinics are in close contact with one another, there are nevertheless differences in the approach to and organisation of care.

Moreover, there was no uniform policy for referral from primary and secondary centres to a Marfan clinic. In order to develop a uniform policy regarding the referral, diagnosis and treatment of Marfan patients, the scientific societies involved in the care for Marfan patients have decided, following an initiative of the Dutch Society of Clinical Genetics (VKGN), to develop a guideline with uniform, and as far as possible, evidence-based recommendations.

The development of the guideline was funded by the Stichting Kwaliteitsgelden Medisch Specialisten (SKMS). The Department of Professional Quality Support of the Dutch Order of Medical Specialists provided methodological support.

For the development of the guideline, a multidisciplinary working group was established in 2010, consisting of representatives from all relevant specialties involved in the assessment and care of Marfan syndrome. The working group consisted of clinical geneticists, cardiologists, a cardiothoracic surgeon, ophthalmologists, a gynaecologist, a paediatrician/-cardiologist, orthopaedic surgeons, a molecular geneticist and an anaesthetist. In 2012 the guidelines were implemented.

The guideline is primarily aimed at all healthcare professionals involved in the detection, diagnosis, monitoring and treatment of patients with Marfan syndrome: GPs, (paediatric) cardiologists, paediatricians, thoracic surgeons, clinical geneticists, ophthalmologists, gynaecologists, orthopaedic surgeons, midwives, paediatricians and doctors at health clinics. The directive is therefore not only intended for specialists linked to a Marfan clinic. The secondary target group is that of patients with Marfan syndrome.

Patients with Marfan syndrome, organised in the Contactgroup Marfan Netherlands, were involved in the creation of this guideline and made recommendations for the organisation of care.

Aim of the guideline

The guidelines create a tool for the provision of uniform care in the field of Marfan patient care.

It provides recommendations for referrers regarding referral policy, which amongst others provides guidelines regarding the characteristics required to indicate a referral to a Marfan clinic. In addition, recommendations are made for care providers at Marfan clinics regarding the diagnostic procedure, the logistics thereof, the clinical assessment and treatment of Marfan patients and family studies. Specific recommendations regarding prenatal diagnosis, pregnancy and childbirth are also made. No recommendations are provided for the treatment of disorders or problems that occur more

frequently in Marfan syndrome than in the normal population, but which do not require treatment other than that appropriate in non-Marfan patients.

Delineation of the guideline

The working group has formulated a number of clinical questions that form the basis for the various chapters of this guideline. The guideline does not aim to describe the entire process of clinical care, but focuses on specific bottlenecks.

For every clinical question the guidelines provide an introduction, a summary of relevant literature, conclusions, considerations and recommendations

Clinical questions were formulated by the working group based on critical issues. When possible, the most important and patient-relevant outcome measures were identified for each clinical question.

If possible, for the clinical questions a systematic literature search was performed and relevant articles were selected. For the remaining clinical questions, the available scientific evidence proved to be insufficient. To answer these questions, the expertise of the working group members was elicited, supported by scientific literature when available. Because Marfan syndrome is a rare disorder, the available literature is often limited to case series and small patient groups.

A summary of the relevant literature and the conclusions are in the guidelines together with the level of evidence supporting a conclusion.

Before making a specific recommendation, in addition to the scientific evidence, other factors must be considered including the expertise of the working group members, patient preferences, costs, availability of facilities or organisational aspects. These factors are, insofar as they have not been scientifically investigated, listed under the heading 'considerations'.

The recommendations provide an answer to the central clinical question and are based on both the available scientific evidence and on the most important considerations.

Evidence or common sense?

For most if not all clinical issues that are described in the guidelines no sufficient scientific evidence is available. Therefore the care for Marfan patients is largely based on expert opinion. As in the name, guidelines are a 'guide' and the care should be tailored for every individual patient. That leaves room for the clinicians to individualize the advice in close collaboration with the patient, based upon the correct scientific information or evidence together with common sense.

Future

As for all guidelines, these guidelines have a limited shelf life. That means that in the near future the guidelines will be revised.

Hopefully, future research will provide evidence for several aspects in diagnosis, therapy and surveillance of Marfan patients.