Clinical use of the Kid-Short Marfan Score (Kid-SMS) as an additional tool for proper diagnosis of Marfan syndrome in children

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Background: Due to age dependent manifestations diagnosis of Marfan syndrome (MFS) in children is sophisticated. Although revised Ghent Criteria (GC) is a major step forward, its utility in children is still restricted due to expensive and technically advanced diagnosis. As early diagnosis submits long-term benefits concerning prognosis, the need of appropriate diagnostic tool for risk stratification is justified. In 2012 we developed the Kid-Short Marfan Score (Kid-SMS) to improve and simplify diagnosis in young patients. This study verifies the benefit of this new risk score.

Methods: Overall 187 paediatric patients were subject to a standardized diagnostic programme. Frequency of diagnosis with the revised Ghent nosology, genetics and the Kid-SMS were analysed and compared in all patients.

Results: During the verification process the Kid-SMS identified more suspected patients with MFS compared to revised Ghent nosology and genetics alone. The developed Kid-SMS covered for more than 96 % of our patients with MFS.

Conclusion: Whereas diagnosis of MFS is sophisticated, Kid-SMS is a useful tool for risk stratification of suspected children with MFS by easy executable diagnosis, especially for paediatricians and paediatric cardiologists. The clinical use may be recommended.