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Fibrous dysplasia and McCune–Albright syndrome: Imaging for positive and differential diagnoses, prognosis, and follow-up guidelines

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ABSTRACT

Purpose: The radiologist plays a critical role at all steps of the management of patients with fibrous dysplasia (FD) and McCune–Albright syndrome (MAS). The development of a standardized approach to the management of FD/MAS is crucial given the low incidence and multiple clinical presentations of these conditions. Our aim was to develop recommendations for bone imaging in FD/MAS management.
Materials and methods: The establishment of National Reference Centers in France as part of a Health Ministry program for orphan diseases has triggered the development of recommendations for the clinical management of FD/MAS. We used a well-established robust methodological approach involving an extensive literature review by a multidisciplinary working group (20 healthcare professionals) and scoring by a peer-review group (20 healthcare professionals different from the 20 previous ones). There were four phases: a systematic literature review, drafting of initial recommendations, peer-review of this initial draft, and drafting of the final recommendations.
Results: Fifty-seven specific recommendations are provided as key points for the diagnosis, prognosis, and follow-up of patients with FD/MAS. Issues of special interest are highlighted in the discussion, and areas in which future research is needed are identified.

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Conclusion: We believe the dissemination of these recommendations within the radiology community may facilitate communication between radiologists and other healthcare providers, thereby substantially improving the management of patients with these rare but potentially disabling conditions.

1. Introduction

Fibrous dysplasia of bone (FD) or Jaffe–Lichtenstein disease [1] is a rare benign bone disease that is congenital but not inherited. Fibrous tissue proliferates at one or more marrow sites in a variable number of bones [2]. Monostotic forms account for 70–90% of all cases [3–7]. No bone is exempt, but the most common sites of involvement are the femoral neck, craniofacial bones, and ribs, with variations in relative frequencies across studies [3–5,7]. The radiological features vary with the amount and degree of mineralized tissue within the lesion [8]. About 2–3% of patients with FD have McCune–Albright syndrome (MAS), which is characterized by café-au-lait spots and endocrine abnormalities, among which peripheral precocious puberty is the most common [1,2,9,10]. Hyperthyroidism, acromegaly, and hypercorticism are present in some patients. Renal phosphate wasting and soft tissue myxomas (Mazabraud syndrome) may also be encountered [2].

The establishment of National Reference Centers in France as part of a Health Ministry program for orphan diseases has triggered the development of recommendations for the clinical management of FD/MAS. FD/MAS is a rare disease whose broad spectrum of clinical manifestations results in the involvement of a wide variety of healthcare professionals (Table 1). The radiologist plays a crucial role at all steps of the management of FD/MAS, since bone imaging provides essential information for the diagnosis, prognostic evaluation, and patient follow-up. Optimal selection of single or combined imaging modalities and the best frequency of follow-up in each specific situation are questions that require clear answers, most notably for non-specialists of FD/MAS.

The aim of this work was to develop recommendations for bone imaging in FD/MAS, with the goal of improving the diagnosis, prognostic evaluation, and follow-up of patients with FD/MAS. We used a well-established methodology involving a systematic literature review and an analysis of the retrieved data by a group of experts.

2. Materials and methods

The French National Authority for Health (HAS) asked the FD/MAS project leaders (PO and RC) to produce high-quality recommendations for this disease [11,12]. These recommendations were developed by two groups of participants, a working group and a peer-review group, and involved four phases: a systematic literature review, drafting of initial recommendations, peer-review of this initial draft, and drafting of the final recommendations [11,12]. Although the entire process is described below, only the recommendations involving bone imaging are presented.

2.1. Participants

The working group was a multidisciplinary group of 20 healthcare professionals (including a radiologist, VB) and a representative of the French patient organization ASSYMCAL. These professionals were experts in FD/MAS and were strongly motivated to develop the recommendations. The project leaders (PO and RC) coordinated the efforts of the working group, and a project officer (CM) was in charge of the systematic literature search and selection of relevant publications, in collaboration with the HAS.

The peer-review group was a multidisciplinary group of 20 healthcare professionals (different from the 20 in the working group) including three radiologists (JDL, NMD, and AF).

Both groups were representative of the wide variety of healthcare settings and geographical sites of practice, as checked by the project leaders and validated by the HAS.

2.2. Method

2.2.1. Systematic literature review

The project officer conducted a systematic search of bibliographic databases for data published over the 11-year period from 1999 to 2009 and retrieved all published scientific papers, clinical practice guidelines, consensus conference reports, articles on medical decision-making, systematic reviews, metaanalyses, and other types of studies, in English or French. Table 2 lists the searched databases and Table 3 the indexing terms used for the search. The project leaders and project officer developed a list of issues for which recommendations were needed. Among these issues, six were in the field of radiology:

- What are the radiological features for the diagnosis of FD?
- When is a bone biopsy indicated and what are the technical requirements?
- How to radiologically assess the osseous/articular prognosis?
- What are the best modalities and frequency for imaging follow-up?
- What are the potential complications and best imaging modalities for their diagnosis?
- Which patients require referral to reference centers?

Of the 567 references retrieved, 189 articles were selected as relevant papers in a variety of specialties. The project leaders, project officer, and working group members critically analyzed these 189 articles, and assigned a level of evidence to each article (Table 4). Furthermore, of the 567 references, 83 that contained words relative to imaging in the title, were written by radiologists, or were published in radiology journals; the working group radiologist reviewed these 83 publications for answers to the issues of interest and assigned a level of evidence to each.

The project leaders and project officer wrote a report describing the evidence from the selected articles.

2.2.2. Drafting of the initial recommendations

During two full-day meetings, the working group discussed the evidence report and suggested recommendations based on the evidence, existing practice, and each member's personal experience. The working group, led by the project leaders and project officer, drafted initial recommendations, which were approved by all working group members before being submitted to the peer-review group.

2.2.3. Peer review

The project officer emailed the evidence report and initial recommendations to the peer-review group members, who rated each