

Best practice management guidelines for fibrous dysplasia/McCune-Albright Syndrome

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Best practice management guidelines for fibrous dysplasia/McCune-Albright syndrome: a consensus statement from the FD/MAS international consortium

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<https://ojrd.biomedcentral.com/articles/10.1186/s13023-019-1102-9>

Why did Researchers create these guidelines?

- FD/MAS is a very complex disease and challenging for clinicians
 - No 2 patients are alike
 - Many different specialties involved, who approach problems from different perspectives
- This has led to huge discrepancies in the types of care patients are receiving

How did Researchers create them?

- Consensus statement created by 51 FD/MAS clinical experts from 13 countries in the USA, Europe and Asia
- Core Guideline Development Group formed at the first FD/MAS International Consortium meeting in Oxford, UK, 2015
- Draft document circulated to the wider group. Comments were reviewed, & items with $\geq 70\%$ consensus were included in the guidelines
- Patient representatives independently developed a separate patient checklist to help patients & doctors use the guidelines

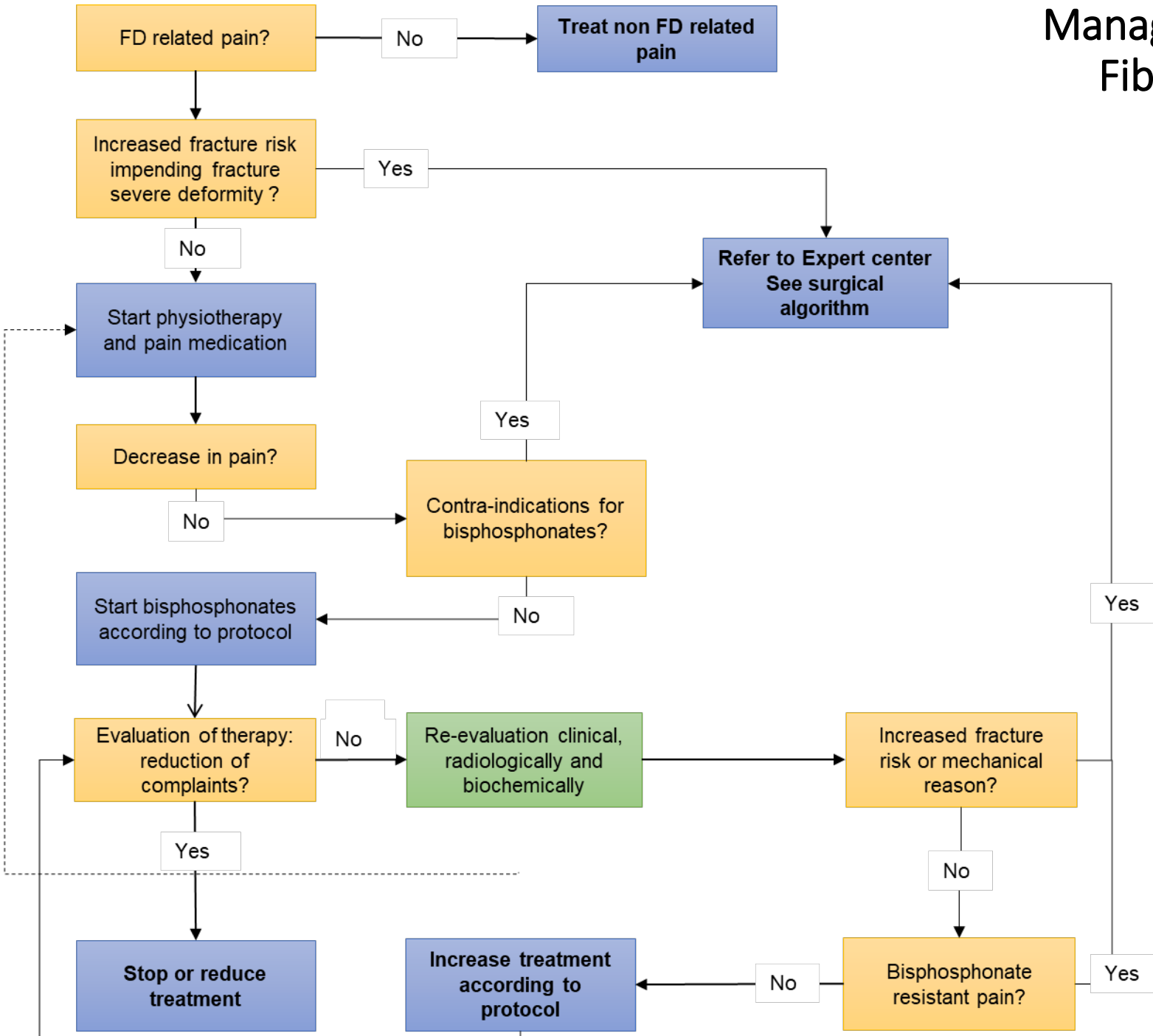
What changes do the guidelines represent in FD/MAS care?

- They define disease terms!
- McCune-Albright syndrome: the combination of FD and one or more extraskeletal features, OR 2 or more extraskeletal features
- Extraskeletal features:
 1. Café-au-lait skin macules with characteristic features
 2. Recurrent ovarian cysts in girls and women OR testicular lesions in boys and men
 3. Thyroid lesions with or without hyperthyroidism
 4. Growth hormone excess
 5. Neonatal hypercortisolism
 6. Intramuscular myxomas

What changes do the guidelines represent in FD/MAS care?

- They outline the full diagnostic workup that most patients need
- They inform doctors in making management decisions
- The guidelines do NOT:
 - Dictate care for every patient
 - Dictate surgical approaches
 - Replace the need for doctors to think about patients individually

Management of Pain in Fibrous Dysplasia



Precocious puberty management in girls

Bone age advanced ≥ 2 years,
frequent bleeding, and/or
psychological distress

Treatment

- Letrozole 2.5 mg daily
- If ineffective--add or replace with tamoxifen, fulvestrant
- Monitor for and treat central puberty

Bone age advancement < 2 years,
infrequent bleeding, and
no psychological distress

Monitoring

Bone age, growth
velocity, H&P every
6 months

Monitoring

Fibrous Dysplasia and McCune-Albright Syndrome: A Checklist for Patients and Doctors

A Tool from the FD/MAS International Consortium

About This Resource

Fibrous dysplasia/McCune-Albright syndrome (FD/MAS) is a rare and complicated disease that can affect the bones, skin and hormone glands.

Which parts of my body are affected by this disease?

<p><i>Write down the answers here.</i></p>	<p>Why is this important?</p> <p>FD is usually present from the time you were born, and the bone disease is usually visible on bone scans by the age of 5. It can occur in almost any bone in the body but usually occurs in the head, legs and ribs. The same mutation that causes FD in the bone also can occur in skin, endocrine tissue (the glands in your body that make hormones) and other organs, so it's important to check to see if those parts of your body are affected too.</p>
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Do I need more tests to find out if other parts of my body are affected?

<p><input type="checkbox"/> Blood and urine tests to look for problems with my:</p> <ul style="list-style-type: none"> <input type="checkbox"/> Ovaries <input type="checkbox"/> Testicles <input type="checkbox"/> Thyroid <input type="checkbox"/> Growth Hormone <input type="checkbox"/> Adrenal <input type="checkbox"/> Phosphate levels 	<p><input type="checkbox"/> Specialist eye sight screening</p> <p><input type="checkbox"/> Hearing screening</p> <p><input type="checkbox"/> Nuclear/isotope bone scan</p> <p><input type="checkbox"/> CT scan</p> <p><input type="checkbox"/> MRI scan</p> <p><input type="checkbox"/> Ultrasound scan of the thyroid</p> <p><input type="checkbox"/> Ultrasound scan of ..</p>	<p>Why is this important?</p> <p>All of these tests can help locate what areas of your body the disease affects, and how seriously they are affected.</p> <p>Some of these tests can detect hormonal imbalances that can make your bone disease worse, unless they are treated correctly.</p>
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How can patients and clinicians make use of the guidelines?

- Patients: Recommend it to your doctors
 - Consider bringing a physical copy and/or sending them an electronic link to the full guideline document
 - Bring a physical copy of the Patient Checklist to your visit and discuss it with your doctor
- Physicians in the FD/MAS community: share them with colleagues and consulting clinicians
- Share on social media

Thank you! Questions?

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