

the **international**
consortium for

FDMAS Ltd.



Inaugural meeting

1st Meeting in Oxford in 2015, 2nd Meeting in Lyon in 2016,
3rd Meeting in Leiden in 2017 (international patientenday), 4th Meeting in Florence in 2019
5th meeting held online on 14th of July, 2022



the international
consortium for

FDMAS Ltd.



Come together Work together

During the meetings

- updates of progress ongoing trials/projects
- “The Clinical Pathway”
- Kick off multiple collaborations

2017: ‘Core Writing group’: FD/MAS Alliance (USA), National Institute of Health (NIH – USA)
LUMC(Leiden the Netherlands), Patiëntenvereniging Fibreuze Dysplasie(NL), EAMS (Italy),
FDSSUK (UK)



Mission?



The mission of the International Consortium for FD/MAS is

- to improve the care and find solutions for the unmet needs of patients and their care providers through the promotion of multi-stakeholder, collaborative, and patient-centric clinical, translational, and basic research.
- The International Consortium for FD/MAS will encourage the testing and development of potential cures and treatments and promote the development of evidence-based standards of care.
- The consortium aims to improve access to information worldwide through dissemination and education initiatives.
- The consortium will support the development of cooperative scientific studies and initiatives with promise to serve the global FD/MAS patient community.

Contact platform; website and live meetings



ICFDMAS

Board of Directors (combination specialist / patients)
Dr. Natasha Appelman- Dijkstra (LUMC) and
Adrienne McBride (FD/MAS Alliance)

Support by

- Dr. Michael Collins (NIH) and Martine Dekker (PV Fibreuze Dysplasie) and Tovah Burnstein (FD/MAS Alliance)

Additive value for patients and doctors/researchers




WHAT HAS BEEN ACHIEVED SO FAR?

Clinical

Position statement | [Open Access](#) | [Published: 13 June 2019](#)

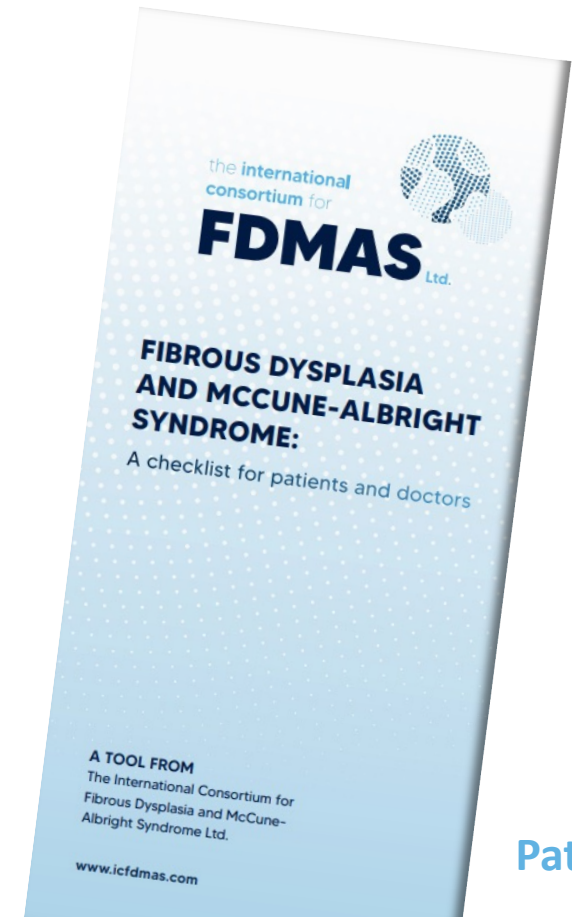
Best practice management guidelines for fibrous dysplasia/McCune-Albright syndrome: a consensus statement from the FD/MAS international consortium

[Muhammad Kassim Javaid](#) , [Alison Boyce](#), [Natasha Appelman-Dijkstra](#), [Juling Ong](#), [Patrizia Defabianis](#), [Amaka Offiah](#), [Paul Arundel](#), [Nick Shaw](#), [Valter Dal Pos](#), [Ann Underhil](#), [Deanna Portero](#), [Lisa Heral](#), [Anne-Marie Heegaard](#), [Laura Masi](#), [Fergal Monsell](#), [Robert Stanton](#), [Pieter Durk Sander Dijkstra](#), [Maria Luisa Brandi](#), [Roland Chapurlat](#), [Neveen Agnes Therese Hamdy](#) & [Michael Terrence Collins](#)

[Orphanet Journal of Rare Diseases](#) **14**, Article number: 139 (2019) | [Cite this article](#)

40k Accesses | **54** Citations | **22** Altmetric | [Metrics](#)

 A [Correction](#) to this article was published on 21 November 2019



Patient guide

the international
consortium for

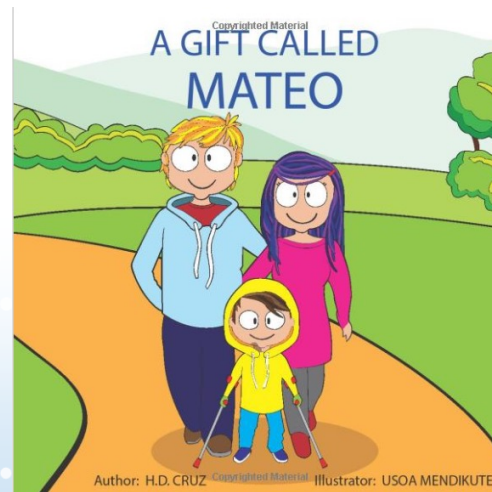
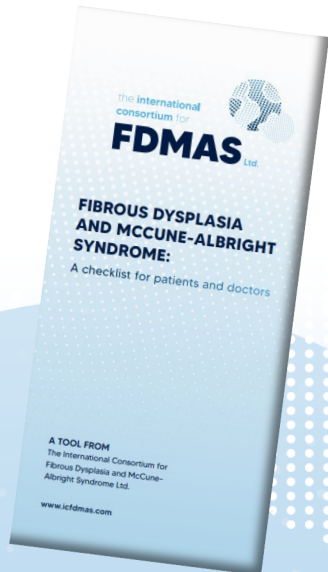
FDMAS Ltd.



WHAT HAS BEEN ACHIEVED SO FAR?

patient advocacy

- Patient guide
- Presentation of priorities in patient care (14.07.2022)
- Global FD/MAS Awareness Week – coordinated awareness campaign
- Children’s book: “A gift called Matteo” available in various languages on Amazon
- Children’s information flyer about FD/MAS
- Animation movie FD/MAS



WHAT HAS BEEN ACHIEVED SO FAR?

Research

More collaborations

- Clinical papers on breastcancer, mazabrauds syndrome, fractures, deformaties
- Patients associations defining the path of interest

Open discussions

- Perhaps leading to the development of multiple models for the disease
- Making it possible to circulate potential study protocols

the international
consortium for

FDMAS Ltd.



Consortium publications

1 **Coxa Vara Deformity in Fibrous Dysplasia/McCune-Albright Syndrome: Prevalence, Natural History and Risk Factors: A Two-Center Study.**
Cite Meier ME, Appelman-Dijkstra NM, Collins MT, Geels RES, Stanton RP, de Witte PB, Boyce AM, van de Sande MAJ.
Share J Bone Miner Res. 2023 Jul;38(7):968-975. doi: 10.1002/jbmr.4818. Epub 2023 May 21. PMID: 37102469

2 **Extent of Extraskkeletal Manifestations of Fibrous Dysplasia/McCune-Albright Syndrome in Patients with Mazabraud's Syndrome.**
Cite Hagelstein-Rotman M, Appelman-Dijkstra NM, Boyce AM, Chapurlat R, Dur NBJ, Gensburger D, Majoor BCJ, van de Sande MAJ, Dijkstra PDS.
Share Calcif Tissue Int. 2022 Mar;110(3):334-340. doi: 10.1007/s00223-021-00918-0. Epub 2021 Dec 2. PMID: 34854944

3 **Long Bone Fractures in Fibrous Dysplasia/McCune-Albright Syndrome: Prevalence, Natural History, and Risk Factors.**
Cite Geels RES, Meier ME, Saikali A, Tsonaka R, Appelman-Dijkstra NM, Boyce AM.
Share J Bone Miner Res. 2022 Feb;37(2):236-243. doi: 10.1002/jbmr.4463. Epub 2021 Nov 17. PMID: 34668234 [Free PMC article.](#)

4 **Correction to: Best practice management guidelines for fibrous dysplasia/McCune-Albright syndrome: a consensus statement from the FD/MAS international consortium.**
Cite Javaid MK, Boyce A, Appelman-Dijkstra N, Ong J, Defabianis P, Offiah A, Arundel P, Shaw N, Pos VD, Underhil A, Portero D, Heral L, Heegaard AM, Masi L, Monsell F, Stanton R, Dijkstra PDS, Brandi ML, Chapurlat R, Hamdy NAT, Collins MT.
Share Orphanet J Rare Dis. 2019 Nov 21;14(1):267. doi: 10.1186/s13023-019-1255-6. PMID: 31752972 [Free PMC article.](#)

5 **Best practice management guidelines for fibrous dysplasia/McCune-Albright syndrome: a consensus statement from the FD/MAS international consortium.**
Cite Javaid MK, Boyce A, Appelman-Dijkstra N, Ong J, Defabianis P, Offiah A, Arundel P, Shaw N, Pos VD, Underhil A, Portero D, Heral L, Heegaard AM, Masi L, Monsell F, Stanton R, Dijkstra PDS, Brandi ML, Chapurlat R, Hamdy NAT, Collins MT.
Share Orphanet J Rare Dis. 2019 Jun 13;14(1):139. doi: 10.1186/s13023-019-1102-9. PMID: 31196103 [Free PMC article.](#) [Review.](#)

6 **Increased Risk of Breast Cancer at a Young Age in Women with Fibrous Dysplasia.**
Cite Majoor BC, Boyce AM, Bovée JV, Smit VT, Collins MT, Cleton-Jansen AM, Dekkers OM, Hamdy NA, Dijkstra PS, Appelman-Dijkstra NM.
Share J Bone Miner Res. 2018 Jan;33(1):84-90. doi: 10.1002/jbmr.3286. Epub 2017 Sep 20.

Fibrous dysplasia animal models: A systematic review.

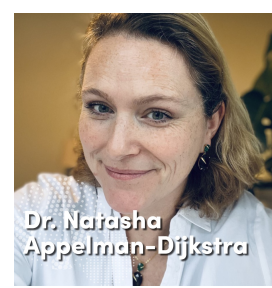
7 Hopkins C, de Castro LF, Corsi A, Boyce A, Collins MT, Riminucci M, Heegaard AM.
Cite Bone. 2022 Feb;155:116270. doi: 10.1016/j.bone.2021.116270. Epub 2021 Dec 4. PMID: 34875396 [Free article.](#)

Share BACKGROUND: **Fibrous dysplasia** (FD) is a rare genetic **bone** disorder resulting in an overproduction of cAMP leading to a structurally unsound tissue, caused by a genetic mutation in the guanine nucleotide-binding protein gene (GNAS). ...

> J Clin Endocrinol Metab. 2022 May 17;107(6):e2258-e2266. doi: 10.1210/clinem/dgac120.

Neuropathic-like Pain in Fibrous Dysplasia/McCune-Albright Syndrome

Tiahna L Spencer¹, Laura Watts², Anushka Soni³, Rafael Pinedo-Villanueva³, Anne-Marie Heegaard⁴, Alison M Boyce⁵, M Kassim Javaid³



What did we do?

- Website
- Linked in page
- Logo/formats
- Organizing the meeting
- Setting the legal base for the consortium

What will we do?

- Define the membership
- Engage with the community worldwide
- Encourage future collaborations
- Support the local organizing committee for 2025
- Plan Annual meeting February 2024