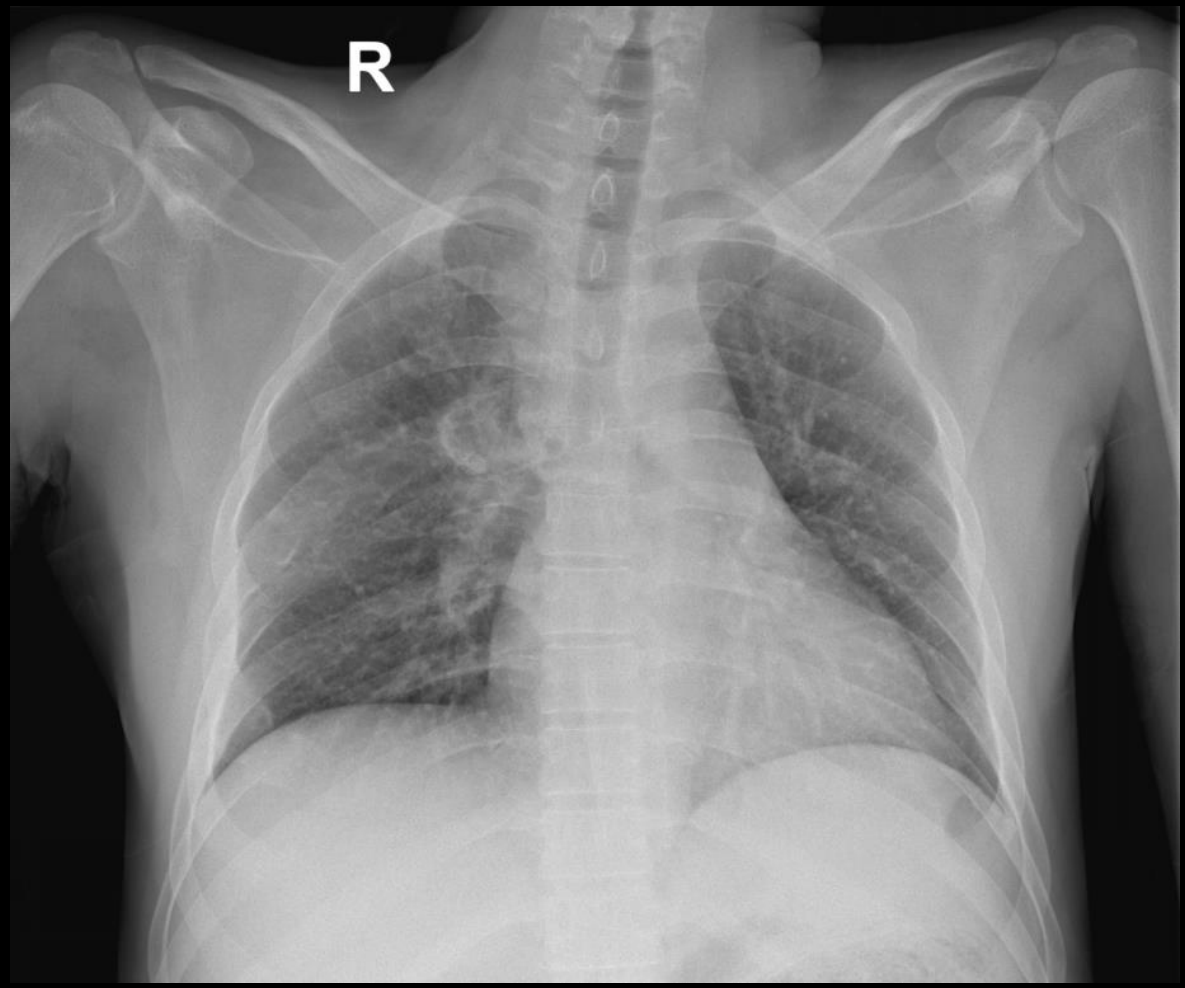
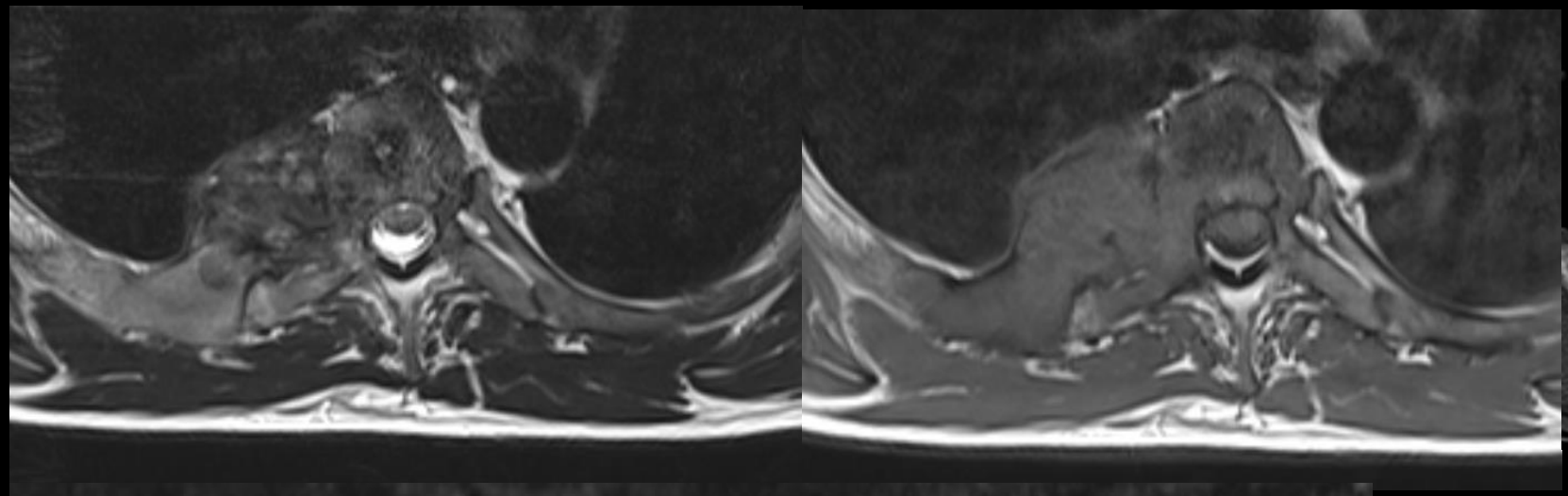
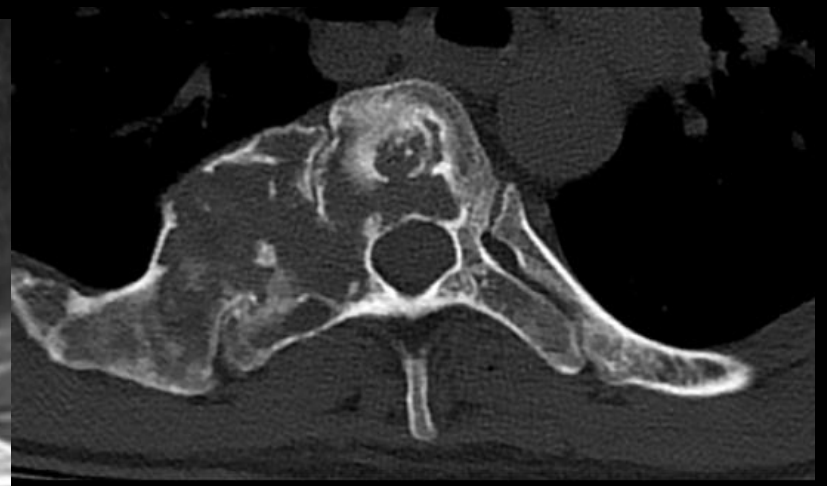
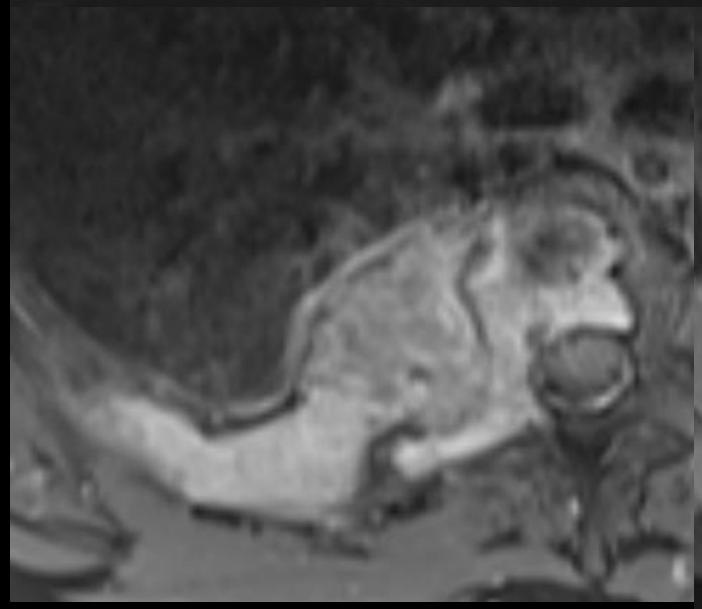
MICOD – 16/04/2024
Case contributor – Dr. Sonal Saran

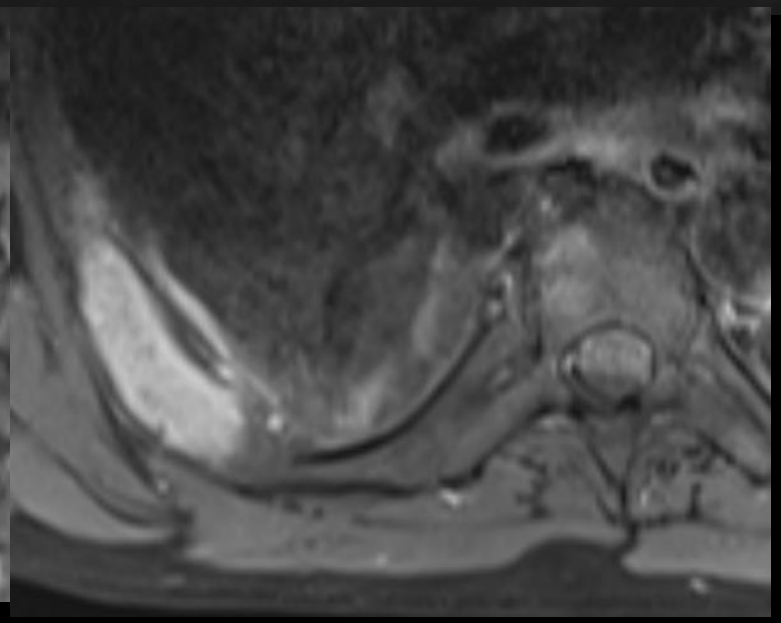


32 year old male with pain in back









Polyostotic cystic fibrous dysplasia (FD)

A lytic expansile lesion involving T5 vertebral body, right lamina and extending into adjacent right costovertebral joint and rib. It shows ground glass matrix. Axial T2W & post-contrast T1 weighted MR images show T2 heterogeneous, predominantly hypointense, lesion involving T5 vertebral body on right side extending into costovertebral joint and right rib with heterogeneous enhancement and causing no obvious compromise of spinal canal.

Polyostotic cystic fibrous dysplasia (FD)

- *Congenital* disorder from sporadic mutation of the *α-subunit of the Gs stimulatory protein*.
- FD arises *sporadically*, and there are no confirmed cases of vertical transmission.
- Osseous changes are characterized by the replacement and distortion of normal bone with *poorly organized, structurally unsound, fibrous tissue*.
- May be localized to a *single or multiple bones*. In *McCune-Albright syndrome (MAS),* FD is associated with hyperfunction of endocrine organs and overproduction of melanin in the skin, while *Mazabraud syndrome* FD is associated with intramuscular myxomas.
- In radiology, FD is very often automatically associated with the term *"ground glass matrix".*

Kushchayeva YS, Kushchayev SV, Glushko TY, Tella SH, Teytelboym OM, Collins MT, Boyce AM. Fibrous dysplasia for radiologists: beyond ground glass bone matrix. Insights Imaging. 2018 Dec

Polyostotic cystic fibrous dysplasia (FD)

Table 1

Nomenclature of the diseases associated with fibrous dysplasia (FD) lesions

Forms of fibrous dysplasia	Bone involvement		Café-au-lait spots	Endocrine disorders	Intramuscular myxomas
	Single	Multiple			
Monostotic	X				
Polyostotic		X			
McCune-Albright syndrome		X	X	X	
Mazabraud syndrome		X			X

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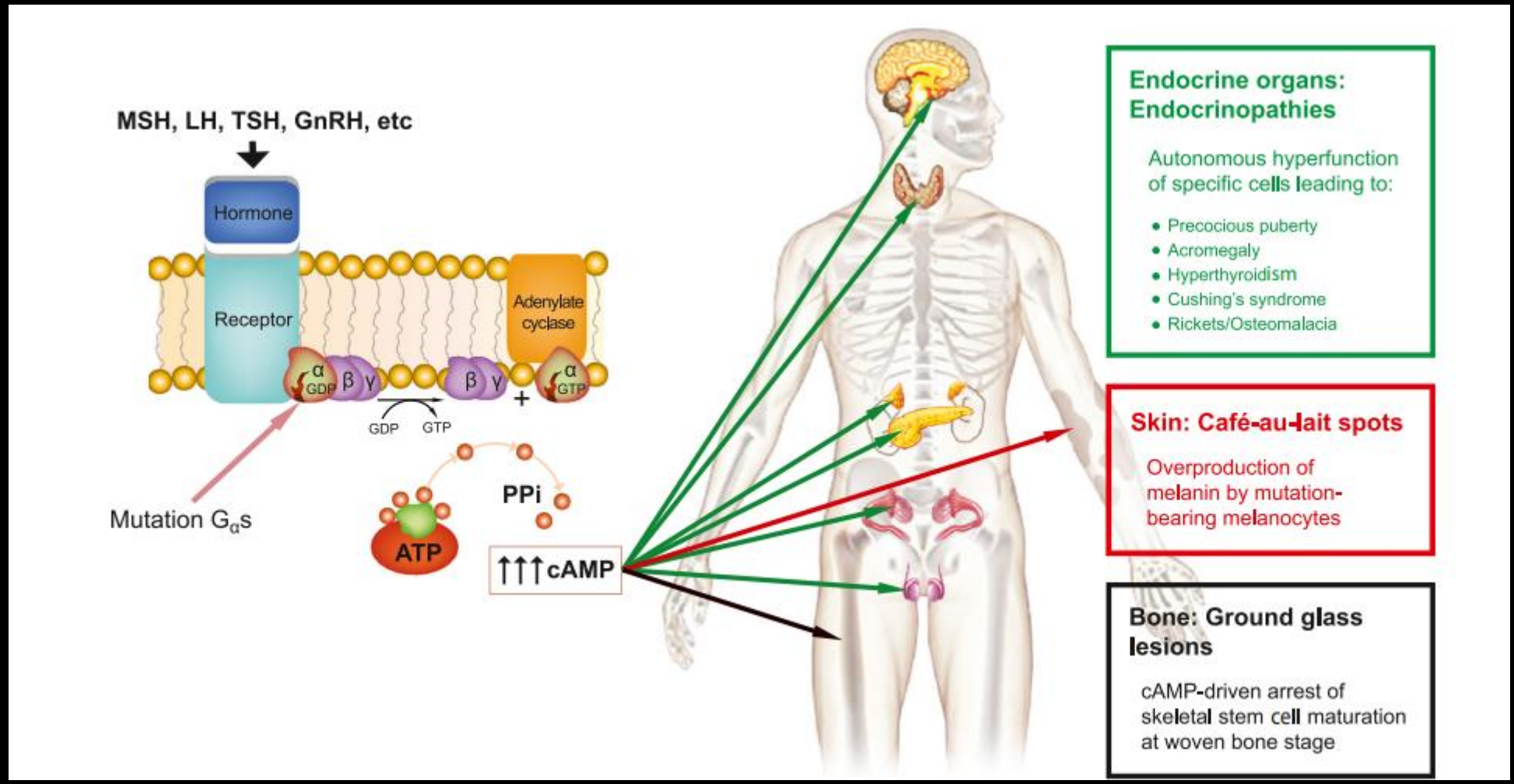
Polyostotic cystic fibrous dysplasia (FD)



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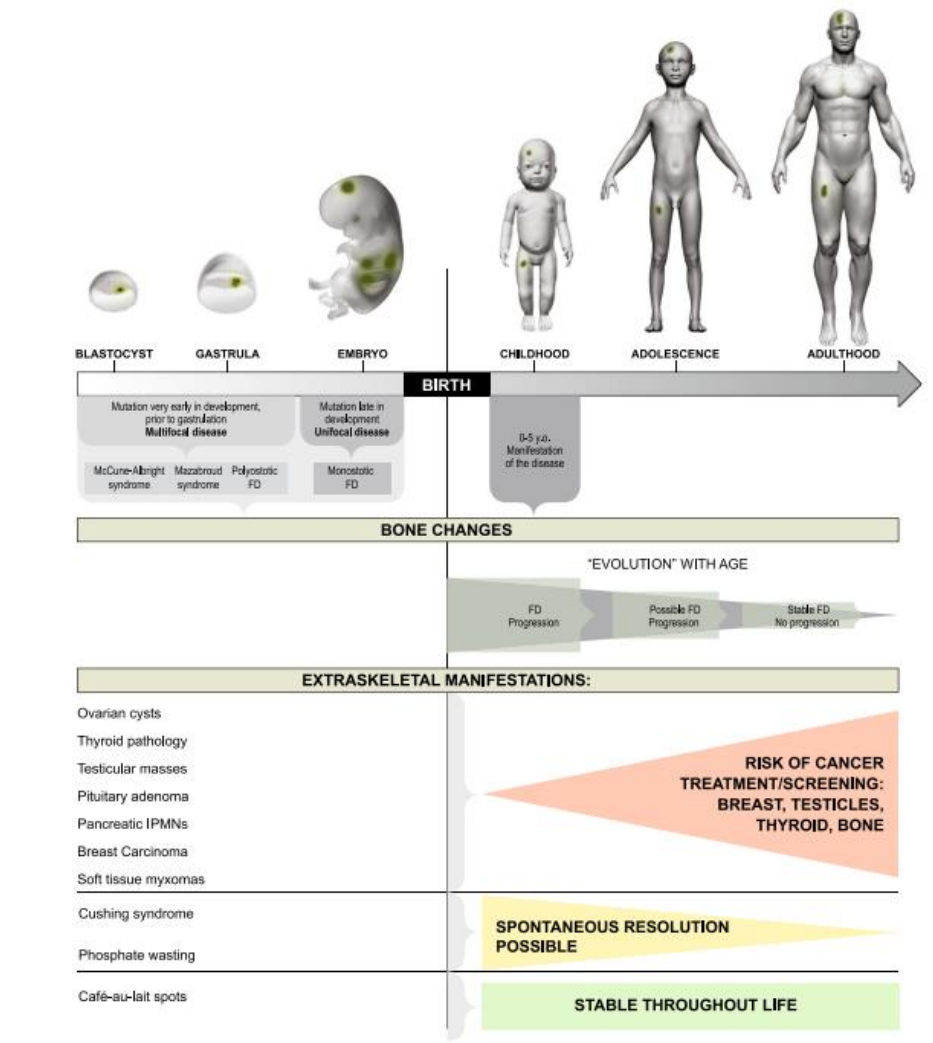

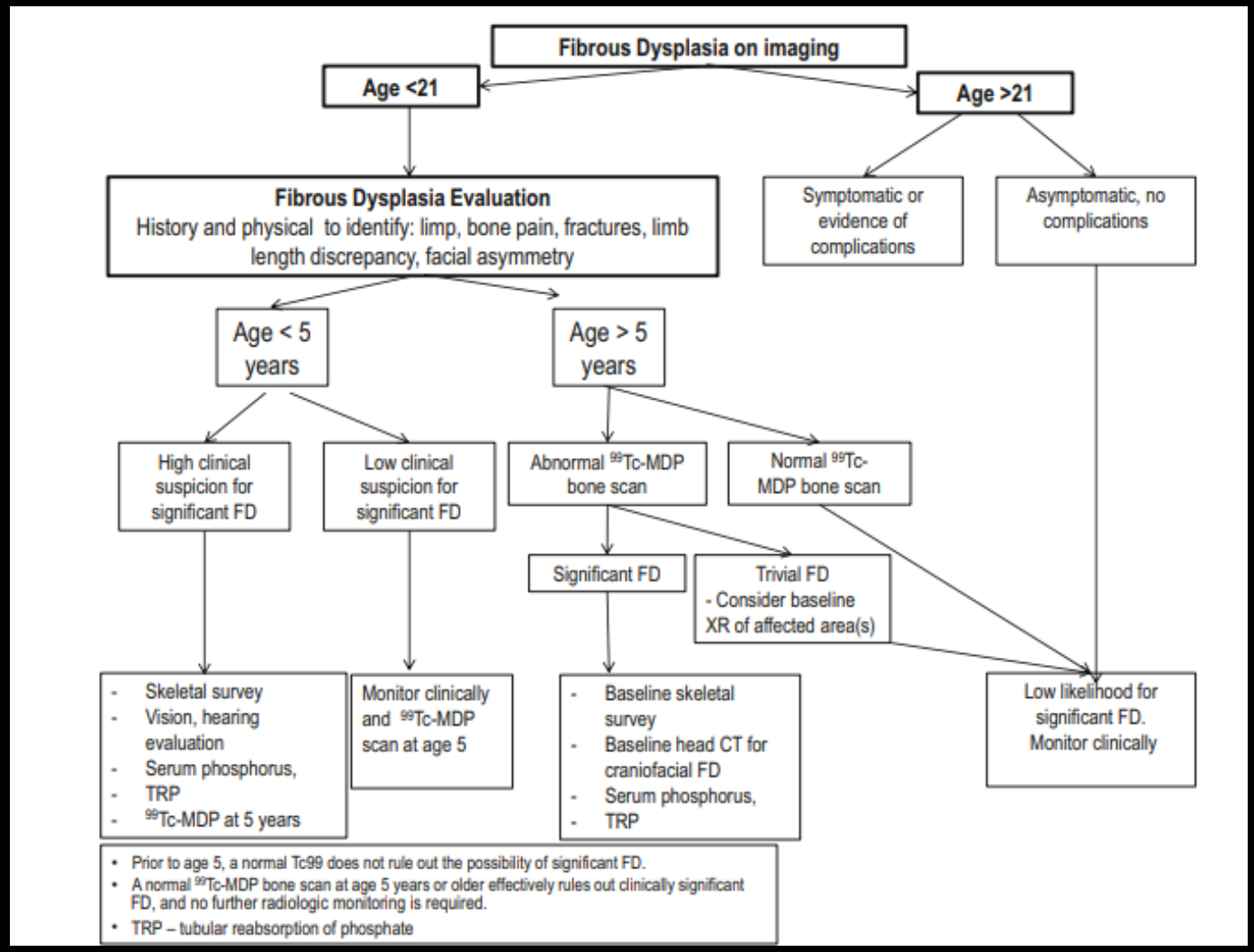

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Polyostotic cystic fibrous dysplasia (FD)

Table 2 Suggested follow-up imaging for patients with fibrous dysplasia, McCune-Albright syndrome and Mazabraud syndrome (an expert opinion, based on the NIH cohort)

Involvement	Organs involved	Frequency of involvement[a]	Clinical problem	Suggested radiological follow-up
Bone lesions	All lesions	100%	Fractures, benign and malignant matrix transformation	Initial bone scan to assess the extension of disease. CT of the affected area/bones to evaluate changes in pain, rapid enlargement, local changes.
	Craniofacial bones	80%	Vision/Hearing	Head CT at baseline. Repeat periodically in childhood to monitor progression. Repeat as needed for symptomatic lesions in adulthood.
	Femur	91%	Deformities	Measure neck-shaft angle to identify progressive femoral neck deformation on X-ray.
	Axial skeleton	63%	Scoliosis	Closely monitor for scoliosis on X-ray; surgical fixation if Cobb angle > 50 degrees.
Extra-skeletal	Thyroid	66%	Hyperthyroidism (38%), autoimmune thyroiditis, thyroid cancer (1.3%)	Thyroid ultrasound at baseline and periodically to follow abnormalities.
	Pituitary	10–15%	Adenoma, hyperplasia without adenoma	MRI brain at baseline for patients with abnormal pituitary function.
	Testicles	85%	Macroorchidism, Leydig or Sertoli cell hyperplasia, testicular germ cell tumour	Testicular ultrasound at baseline and periodically to follow abnormalities.
	Ovaries	85%	Autonomous ovarian cysts	Pelvic US if breast development, vaginal bleeding or signs of estrogenisation below age 6–7 years.
	GI tract	32%	Pancreatic IPMN	MRI of abdomen with MRCP follow-up in 6–12 months if IPMNs 10–20 mm; 6 months follow-up if > 20 mm or demonstrates suspicious features
	Intramuscular myxomas in Mazabraud syndrome	100%	Asymptomatic	No follow-up

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Polyostotic cystic fibrous dysplasia (FD)

- FD is a complex disease, characterized by age-related histological, radiographic and clinical transformations. *Radiologists play a crucial role in the identification of osseous complications*
- The craniofacial form of the disease is the most common type of FD and the most difficult form to manage. It requires clinical and radiological evaluation and follow-up.
- Patients with *MAS* may have different *extra-skeletal abnormalities (ovarian cysts, testicular changes, pituitary adenoma or IPMN),* which often require follow-up.
- Many patients with FD undergo repeated imaging with radiation; therefore, *high radiation exposure is a concern*. Efforts should be made to reduce cumulative radiation risks.

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Thank you