

A 14-year-old male, suffering from  
right hearing loss for 3-4 years

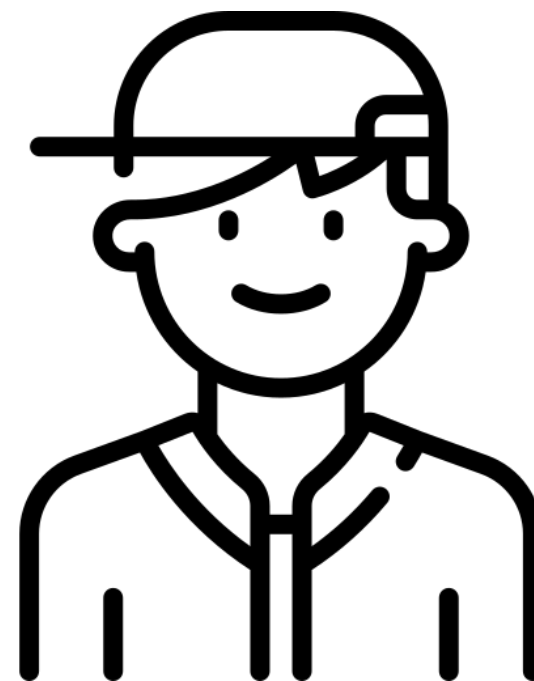
Date:2025/05/21

Presenter: PGY 2 徐維成

Supervisor: VS 洪偉誠

# Basic data

Chart Num:	H63416
Name:	黃OO
Age/Sex:	14 y/o / Male
Source of information	The patient's family
Social & Personal history:	No specific
Allergic / Family history:	NKA / no specific
Admission from:	OPD
Chief complaint	Right hearing loss for 3-4 years



1st Treatment course (2019-2020)

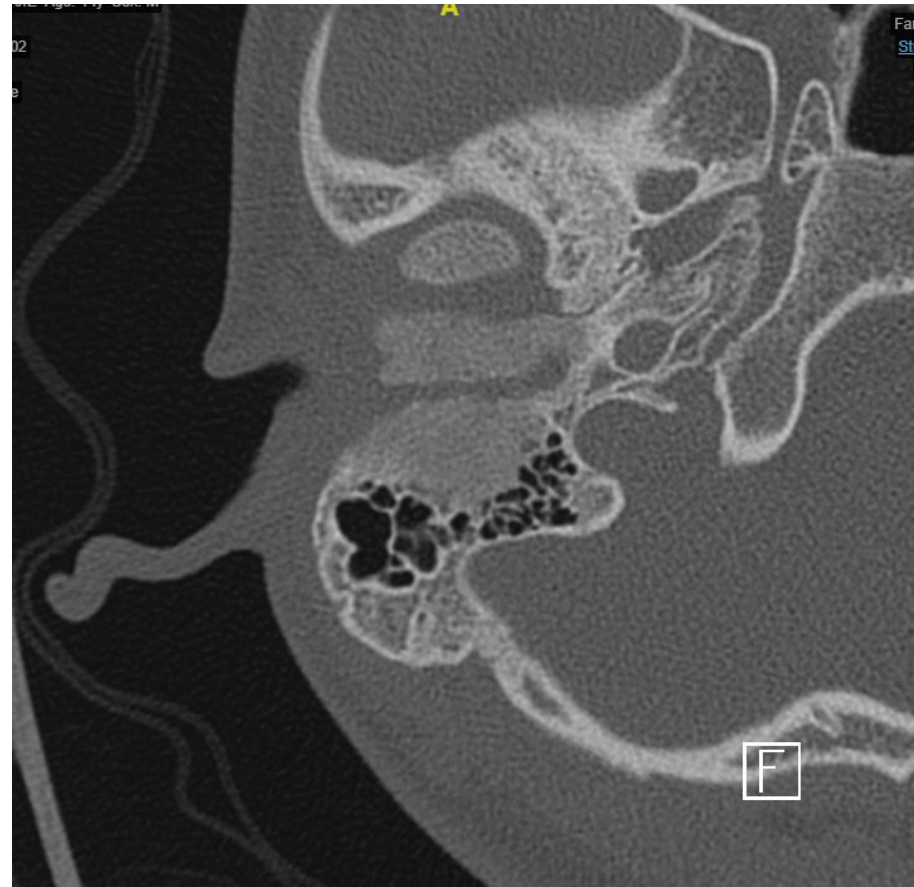
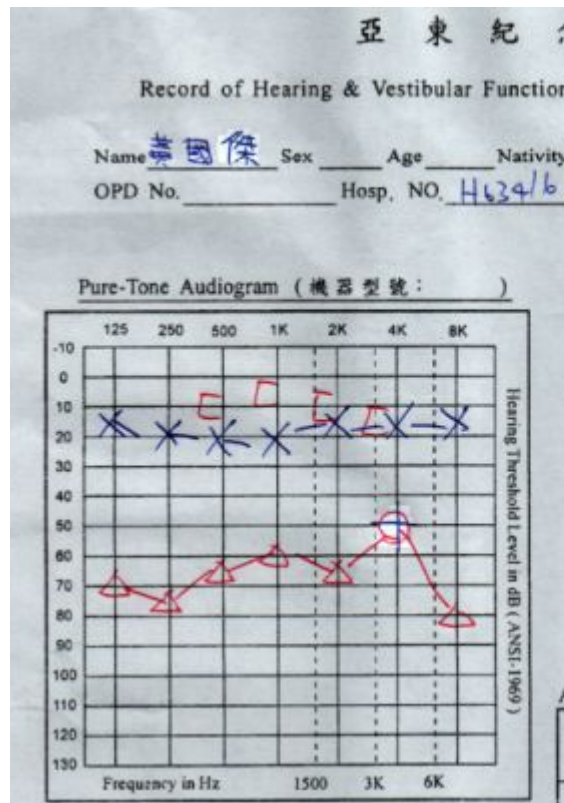
# Present illness

- 2019/09/19 OPD **initial visit:**
  - **Sx:** **Right hearing loss for 3-4 years**, right otalgia for 3-4 days
  - **PE:**
    - left eardrum: intact
    - right eardrum: cannot be seen, right EEC very stenosis
  - **Plan:**
    - Arrange PTA for hearing loss assessment
    - Arrange High Resolution CT for Temporal Bone
    - Arrange admission for surgical intervention

# Present illness (Initial visit)

2019/10/02 OPD:

- **Pure Tone analysis:** significant A-B gap: about 50 dB



- **HR-CT**
  - Ground-glass lesions in the mastoid process of right temporal bone, partially obliterating the mastoid air cells
- DDx: fibrous dysplasia

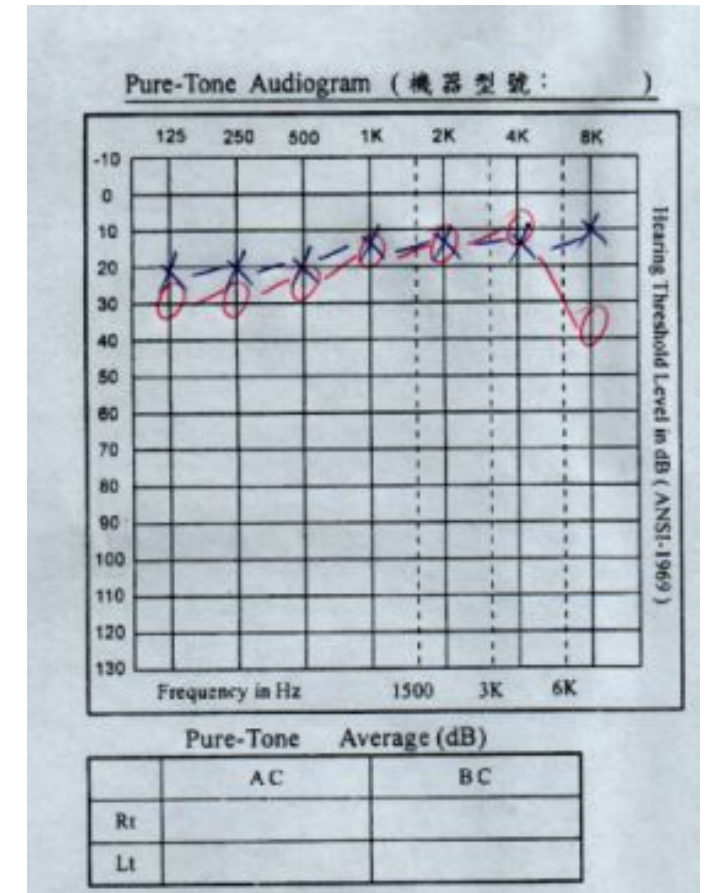
# Present illness (**1st OP**)

- Admission course: 2019/10/17-20
    - **Surgery performed: Right tympanoplasty type I with right canaloplasty**
- Post-op care and arrange OPD follow up on 2019/10/20.
- **Patho:**
    - fibro-osseous lesion composed of thin trabeculae of woven bone, suggestive of fibrous dysplasia.

# Present illness (**After 1st OP**)

OPD follow up **after 1st OP** till 2020/06/03

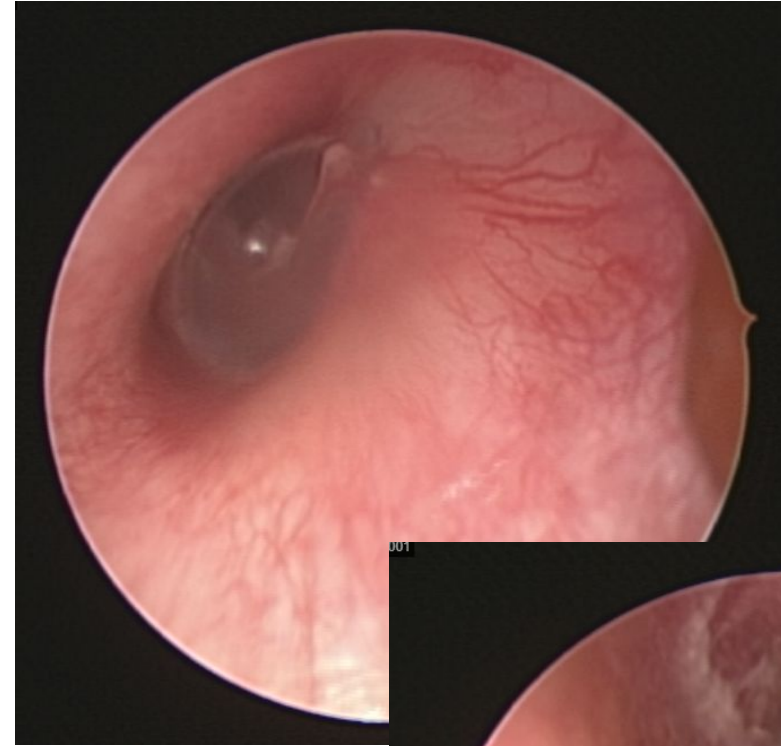
- **Sx:** Right ear itching and fullness after surgery, improved gradually
- **PE:** right eardrum: some cerumen and debris noted
- **Pure tone analysis on 2020/02/12**
  - hearing test generally normal
  - no A-B gap, no conductive hearing loss



# Present illness

OPD follow up **after 1st OP**

- **Sx: Right ear itching and fullness after surgery, improved gradually**
- **Otoendoscope:**
  - Left: intact eardrum
  - Right: patent ear canal





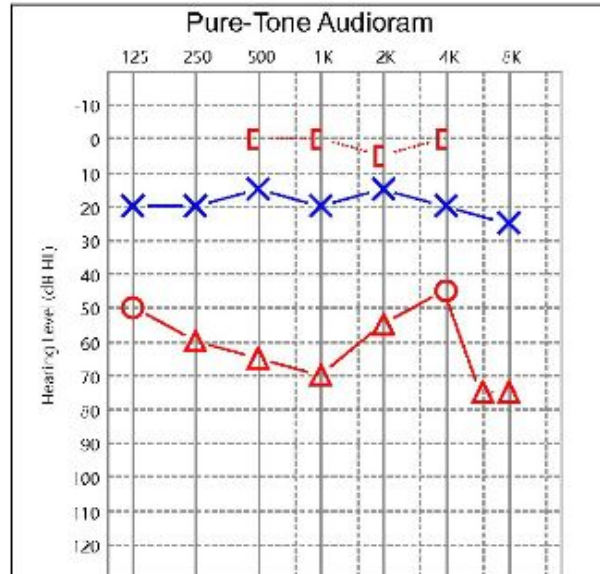
2nd Treatment course (2022-2024)

# Present illness (**Before 2nd OP**)

- OPD: 2022/06/15
- **Sx: Right hearing loss for more than 1 week**
- **PE:**
  - left eardrum: intact
  - right eardrum: stenotic EEC (06/15), more stenotic (07/25)
- **Plan:**
  - Arrange PTA for hearing loss assessment
  - Re-arrange High Resolution CT for Temporal Bone
  - Arrange admission for surgical intervention



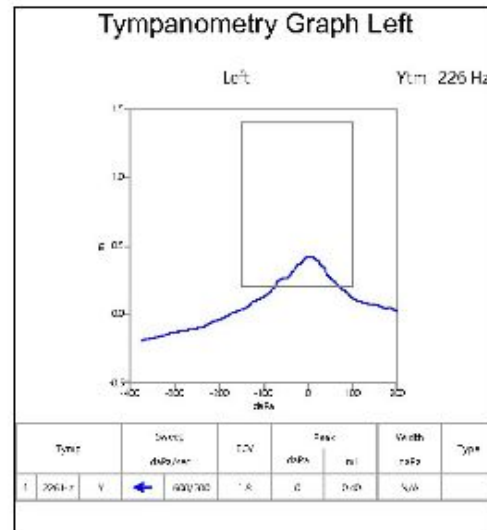
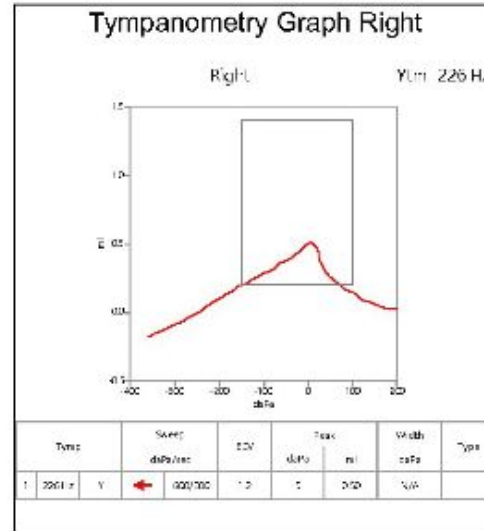
# Present illness (Before 2nd OP)



AC PTA **59 dB** BC PTA **1 dB** SII **4 %**

AC PTA **18 dB** BC PTA SII **95 %**

Speech Test				
Ear	Test Type	Aided	%	dB HL
L	SRT			20
R	SRT			65



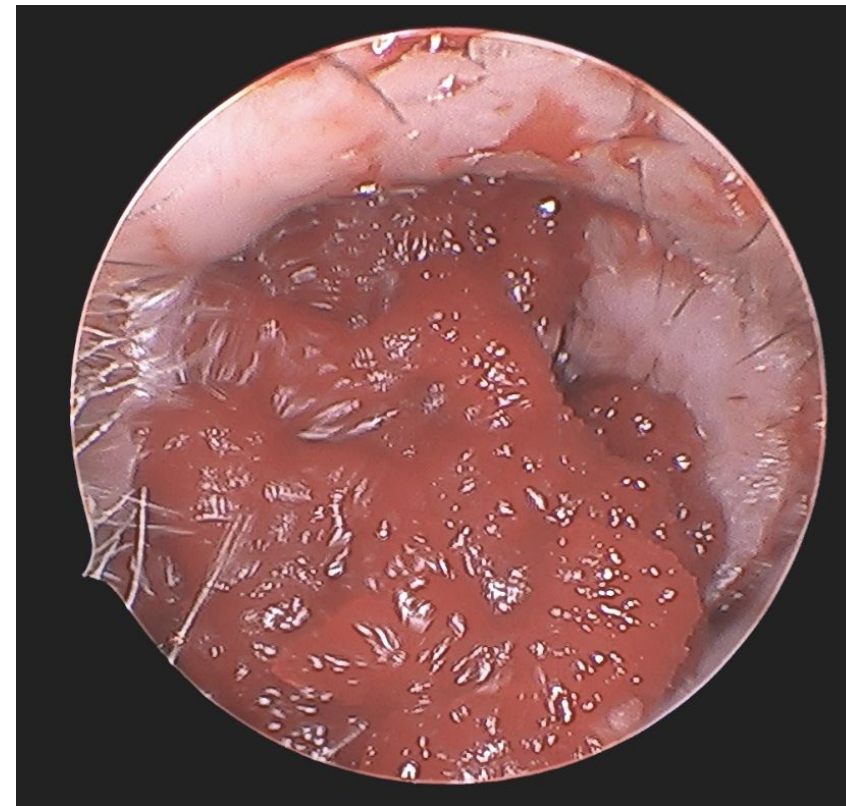
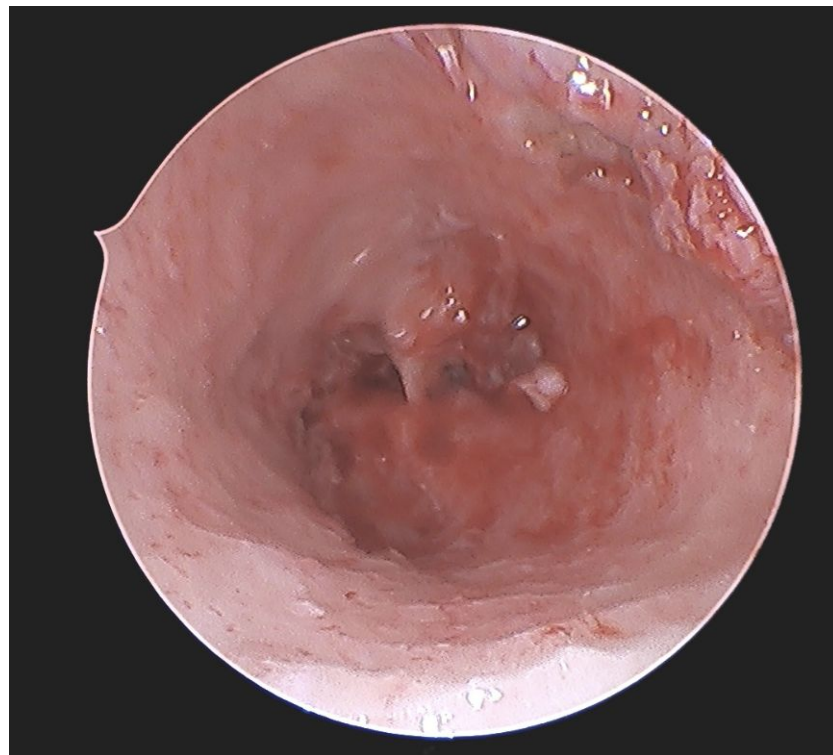
# Present illness (2nd OP)

- Admission course: 2022/10/27-30
  - **Surgery performed: Right tympanoplasty type I with right canaloplasty + mastoidectomy**
  - Operation time: 3 hours

Post-op care and arrange OPD follow up on 2nd./Nov./2022

- **Patho:**
  - Osseous tissue

# Operation image (2nd OP)

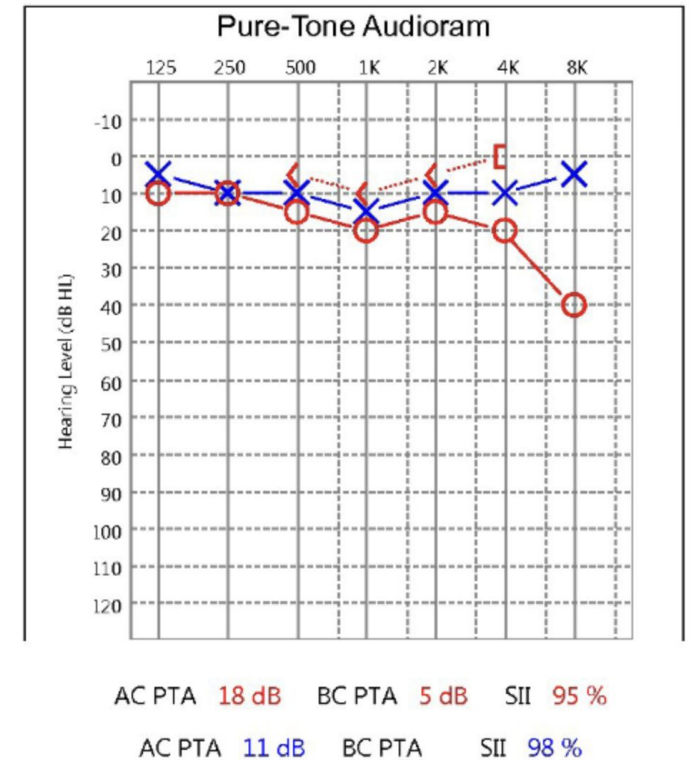




# Present illness (After 2nd OP)

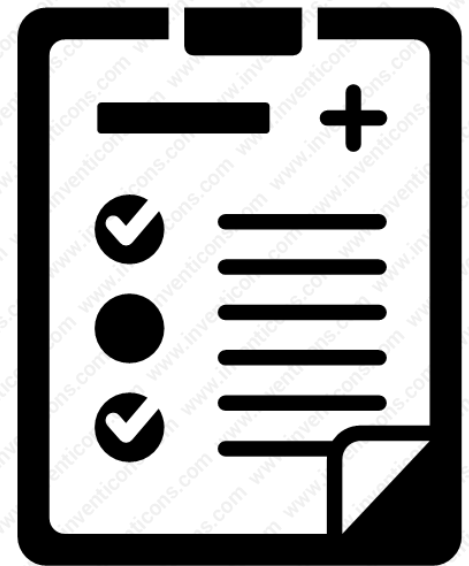
OPD follow up till **AFTER 2nd. OP**

- **Sx: right aural itchiness and mild otorrhea resolved during further follow up**
- **Pure tone analysis on 2nd./Feb./2022**
  - hearing test generally normal
  - no A-B gap, no conductive hearing loss



# Final Diagnosis

- Right external acoustic canal stenosis, due to fibrous dysplasia status post
  - right tympanoplasty (type I) + canaloplasty on 2019/10/18;
  - right tympanoplasty (type I) + mastoidectomy + canaloplasty on 2022/10/28;





► Int J Mol Sci. 2023 Oct 26;24(21):15591. doi: [10.3390/ijms242115591](https://doi.org/10.3390/ijms242115591)

## A Rare Skeletal Disorder, Fibrous Dysplasia: A Review of Its Pathogenesis and Therapeutic Prospects

[Ha-Young Kim](#)<sup>1,2</sup>, [Jung-Hee Shim](#)<sup>3,4</sup>, [Chan-Yeong Heo](#)<sup>1,2,3,\*</sup>

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Impact factor: 4.9

Review Article

## Craniofacial fibrous dysplasia: A review of current literature

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Impact factor: 3.5

# Review: Fibrous dysplasia



# Definition and classification

- **Definition:**
  - *"Fibrous dysplasia is a congenital, non-inherited, benign intramedullary bone lesion in which the normal bone marrow is replaced by abnormal fibro-osseous tissue."*
- **Types:**
  - **Monostotic** (involving a single bone)
  - **Polyostotic** (involving multiple bones)
  - **McCune-Albright Syndrome** (FD with endocrine abnormalities: precocious puberty, hyperthyroidism, Cushing syndrome and café-au-lait skin pigmentation).

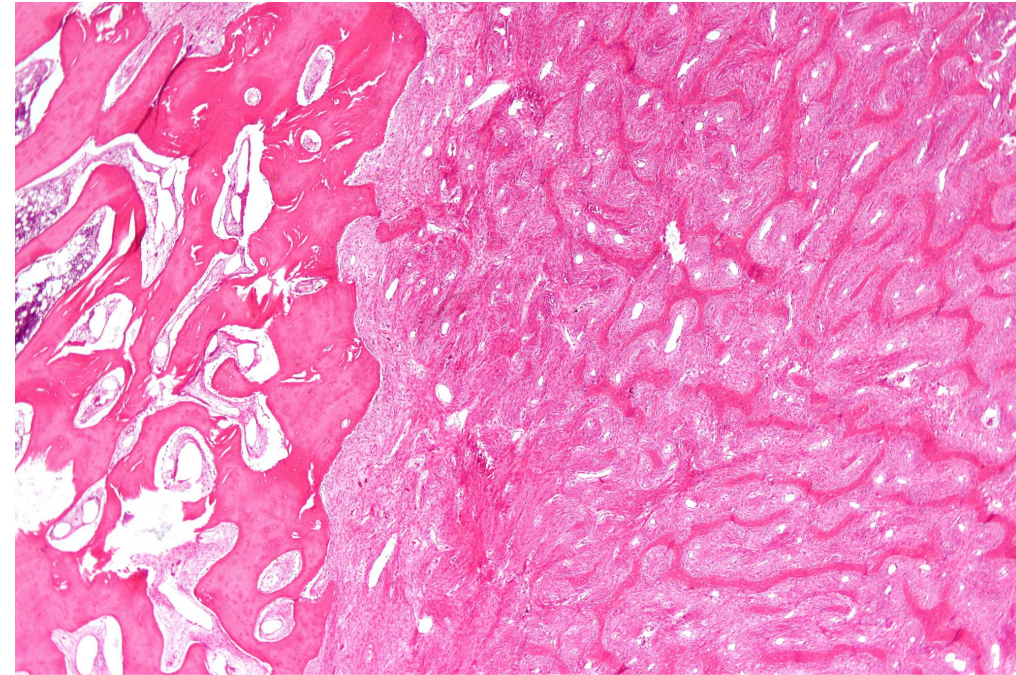


# Epidemiology and Pathogenesis

- **Prevalence:**
  - Approximately 1 in 5,000 to 10,000 individuals
- **Age of Onset:**
  - Monostotic FD: Typically presents in adolescence or early adulthood
  - Polyostotic FD: Often diagnosed before age 10
  - Gender Distribution: No significant gender predilection
- **Genetic Mutation:**
  - Activating mutations in the **GNAS** gene
  - Pathway: "*Somatic gain-of-function mutations in the cyclic adenosine monophosphate (cAMP)-regulating protein Gas alter differentiation of multipotent skeletal stem cells.*" (Boyce et al., 2023)
- **Result: Replacement of normal bone with fibro-osseous tissue**

# Histopathology

- **Microscopic Features:**
  - Irregularly shaped trabeculae of woven bone
  - Fibrous stroma lacking osteoblastic rimming
- **Characteristic Appearance:**
  - "Micrograph showing fibrous dysplasia with the characteristic thin, irregular bony trabeculae and fibrotic marrow space."



*fibrous dysplasia juxtaposed with unaffected bone (wikipedia)*

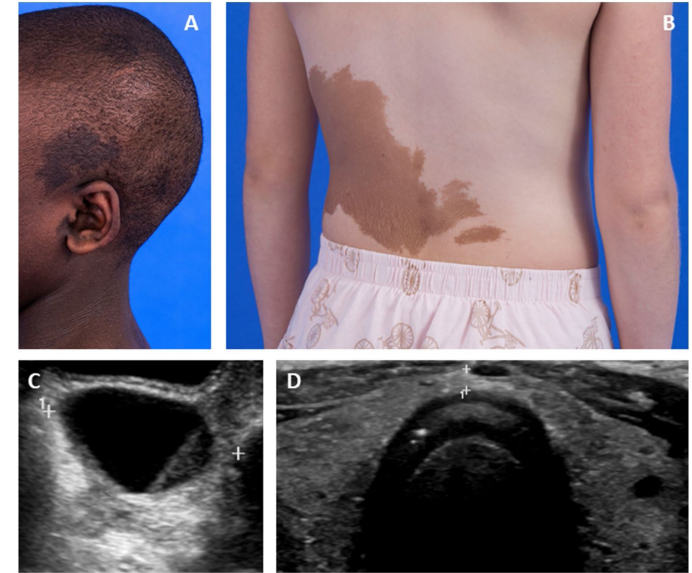
# Clinical presentation and Radiographic traits

- **Clinical presentation**

- Symptoms range from asymptomatic to severe deformity and bone pain, swelling, fractures
- Craniofacial deformity in 50% of polyostotic and 30 % in monostotic

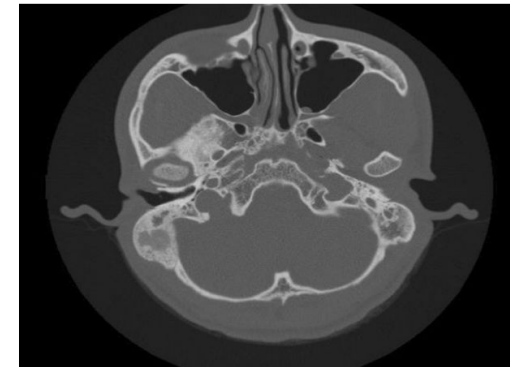
- **Radiographic traits**

- Ground-glass matrix on X-ray.
- Expansion of affected bone with cortical thinning.
- CT and MRI for precise lesion delineation.
- High-resolution CT is particularly valuable for temporal bone evaluation.



*Ovarian cyst*

*Heterogeneous thyroid gland*



**Fig. 3** HRCT showing heterogeneous area with ground glass appearance involving right mastoid portion of temporal bone

# Hearing loss and temporal bone involvement

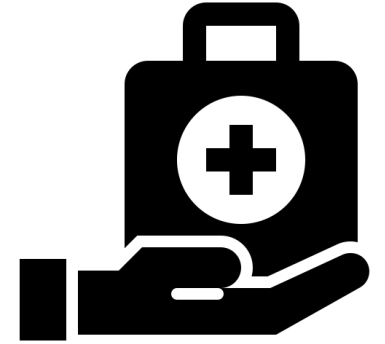
- **Temporal Bone Involvement in FD and Hearing loss**
  - Temporal bone is commonly affected in craniofacial FD(5%-10%) often within polyostotic presentations.
  - Symptoms include aural fullness, otalgia, and conductive hearing loss.
  - Lesions may involve the external auditory canal, ossicles, and middle ear cavity (sensorineural HL is less common unless inner ear was compressed)
  - "Fibrous dysplasia of the temporal bone may cause external auditory canal stenosis and ossicular chain fixation, leading to conductive hearing loss." (Triantafillou et al., 2023)



# Diagnosis and natural course

- **Diagnosis**
  - Based on clinical exam, radiologic features, and histopathology.
  - Differential diagnosis includes other osteogenesis disease: ossifying fibroma, low-grade osteosarcoma, and Paget's disease.
- **Natural History and Prognosis**
  - Lesions develop in early childhood and stabilize post-puberty.
  - Disease may remain stationary or slowly progress.
  - Risk of pathological fractures and deformity increases with age.

# Management and follow up

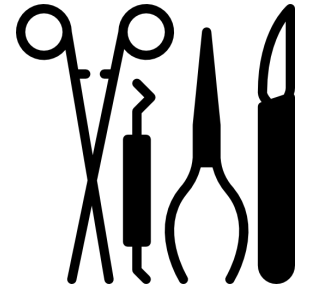


- **Conservative Management**
  - Observation for asymptomatic cases.
  - Pain managed with NSAIDs.
  - Regular monitoring with imaging and lab markers.
- **Bisphosphonates**
  - Used to reduce bone pain and lesion expansion, fractures
  - "The therapeutic efficacy of these agents in the control of disease activity remains uncertain." (Triantafillou et al., 2023)
- **Denosumab:**
  - RANKL inhibitor reduces osteoclast activity.
  - Shown to reduce lesion activity and pain.
  - Requires monitoring for rebound hypercalcemia.

# Management

- **Endocrine Abnormalities in MAS**
  - Precocious puberty, hyperthyroidism, Cushing syndrome.
  - Managed in collaboration with endocrinologists.
  - Requires hormone suppression or other medical therapy.





# Surgical management

- **Surgical Management**
  - Indicated for deformity, fracture, and severe pain.
  - Options include curettage, bone grafting, osteotomy, and internal fixation.
  - "Surgical treatment remains appropriate when pain is unresponsive to other medical treatments..." (Zoccali et al., 2024)
- **Temporal Bone Involvement**
  - Conductive HL is present in up to 50% of temporal bone FD cases. Sensorineural HL is less common
  - Conductive hearing loss is the most common type due to ossicular chain dysfunction or EAC narrowing.
  - Surgery may include canaloplasty or ossiculoplasty.

# What's next?

- **Quality of Life and Long-Term Follow-up**
  - Chronic pain, mobility limitations affect QOL.
  - Periodic imaging and functional assessments necessary.
  - Stigmatization and the psychosocial matter in craniofacial FD
- **Emerging Therapies**
  - Targeting molecular pathways (e.g., MEK inhibitors, RANKL antagonists).
  - "A small molecule RANKL inhibitor, AS2676239, shows promise in suppressing osteoclastogenesis." (Triantafillou et al., 2023)

# Other cases series

S4352

Indian J Otolaryngol Head Neck Surg (December 2022) 74(Suppl 3):S4350–S4355

**Table 1** Detailed Characteristics of patients

Case	Age/ Sex	Presenting Symptom	Otological examination	Radiological Extent of Lesion	Management	Outcome	Follow up for FD
1	24/ M	Hearing loss	EAC Stenosis PTA: 50 dB CHL	Entire temporal bone sparing Petrous portion and extending to zygomatic arch	Canaloplasty Revision canal wall down tympanomastoidectomy	Wide EAC Better hearing	Stable on routine follow ups
2	18/ M	Left temporal mass	Normal PTA: Normal hearing	Left squamous and mastoid part of temporal bone	MCFA approach Titanium Plate and screw for Reconstruction	Hearing normal Cosmesis better	No lesion on routine follow ups yearly
3	25/ M	Incidental finding Asymptomatic	Normal PTA: Normal hearing	Right Mastoid portion of temporal bone	Wait and watch	Patient on regular follow up	Stable upto 2 years follow up

*CHL* Conductive Hearing loss, *FD* Fibrous dysplasia, *MCFA* Middle Cranial Fossa Approach

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- Tuompo S, Mäkitie RE, Nieminen MT. Craniofacial fibrous dysplasia: A review of current literature. *Bone*. 2025;192:117377. doi:10.1016/j.bone.2024.117377

# Back to our patient

- **This was a 14 year-old boy presented with right hearing loss for 3-4 years.**
- **The patient underwent canaloplasty twice on 2019 and 2022.**
- **Conductive hearing loss improved significantly after surgery.**

**The disease, fibrous dysplasia, is a slow progressing, benign disease that causing bone pain and bone deformities. Symptoms varied according to size and location affected.**

THANK YOU FOR YOUR ATTENTION