CENTRAL PRESENTATION
DHAKA MEDICAL COLLEGE & HOSPITAL

A 52-YEAR-OLD WOMAN PRESENTED WITH MULTIPLE JOINT PAIN AND FACIAL ASYMMETRY

DR. TANIA SULTANA
FCPS P-2 TRAINEE (CMT)
MEDICINE UNIT 5

PARTICULARS OF THE PATIENT

- Name : Mrs. X
- Age : 52 years
- Sex : Female
- Address : Gazipur
- Occupation : Housewife
- Marital status : Married
- Religion : Islam
- Date of admission : 4/12/24
- Date of examination: 4/12/24

THE PRESENTING COMPLAINTS

- Pain in multiple joint for 5 years
- Swelling of the both lower limbs for 1 year
- Shortness of breath for 1 year

HISTORY OF PRESENT ILLNESS

According to the statement of the patient, she was reasonably well 5 years back. Then she developed pain in multiple joint that:

- Involved both hands and feet
- Initially small joints later involved large joints too
- Pain was mild but gradually it has become severe
- Morning stiffness persists for 4 hours and was relieved by activity

HISTORY OF PRESENT ILLNESS

She complained of swelling of both lower limbs for 1 year that was:

- Gradual in onset
- Increased progressively over months
- No pain/ facial puffiness
- Not improved by elevation / rest
- No history of trauma /surgery / radiation exposure

HISTORY OF PRESENT ILLNESS

She also complained of shortness of breath that was:

- Mainly exertional
- Progressively worsen over time
- Associated with fatigue and palpitation
- Not aggravated by lying flat
- No chest pain/ seasonal variation

HISTORY OF PRESENT ILLNESS

There were no history of:

- Fever
- Weight loss
- Skin tightening and rash
- Oral ulcer
- Dry eye and mouth
- Contact with TB patient
- Color change in cold exposure
- Blood loss or repeated blood transfusion
- Muscle weakness
- Documented COVID-19 infection

HISTORY OF PRESENT ILLNESS

- She was normotensive and non-diabetic
- Her bladder and bowel habits were normal

HISTORY OF PAST ILLNESS

There were no significant past medical history

DRUG HISTORY

She used to take NSAIDs occasionally for last 5 years

PERSONAL HISTORY

- Non smoker, non alcoholic, non beetle nut chewer
- Uses sanitary latrine and drinks arsenic free water

FAMILY HISTORY

- Eleventh issue of non consanguineous parents
- No history of such kind of illness

SOCIOECONOMIC HISTORY

- She belongs to a lower middleclass family
- Lives in a tin shed house

OBSTETRIC AND GYNAECOLOGICAL HISTORY

- Menopausal for 3 years
- Mother of three child
- Her all three pregnancies were uneventful

IMMUNIZATION HISTORY

She was vaccinated against:

- BCG
- Tetanus
- MMR

No history of:

- COVID-19
- Influenza
- Pneumococcal vaccination

GENERAL EXAMINATION

- Appearance **: Ill looking, angle of mouth is deviated to right and pre and post auricular skin tags with incomplete development of left external ear (since birth)**
- Body build : Average
- Nutrition : Average
- Co operation : Co operative

GENERAL EXAMINATION

- Decubitus : On choice
- Anaemia : Severe
- Oedema : Bilateral pedal oedema
- Jaundice : Absent
- Cyanosis : Absent
- Koilonychia : Absent

GENERAL EXAMINATION

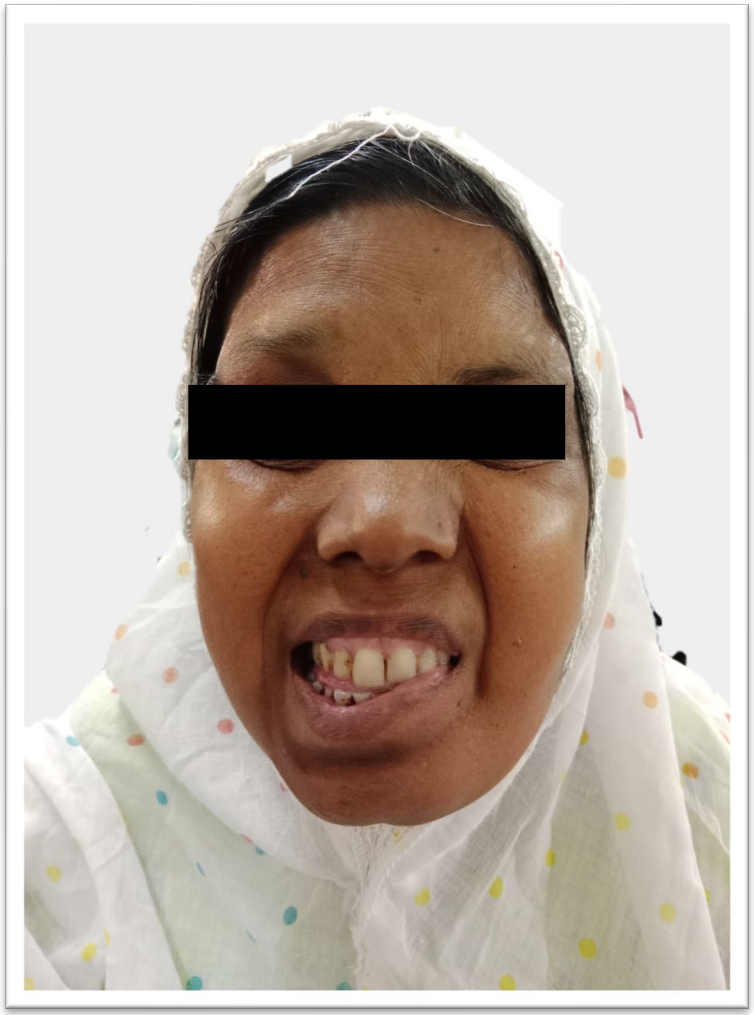
- Leukonychia : Absent
- Temp : 98º F
- Saturation : 95% in room air
- BP : 120/70 mm of Hg (no postural drop)
- Pulse : 112 beats/min and regular
- RR : 16 breaths/min

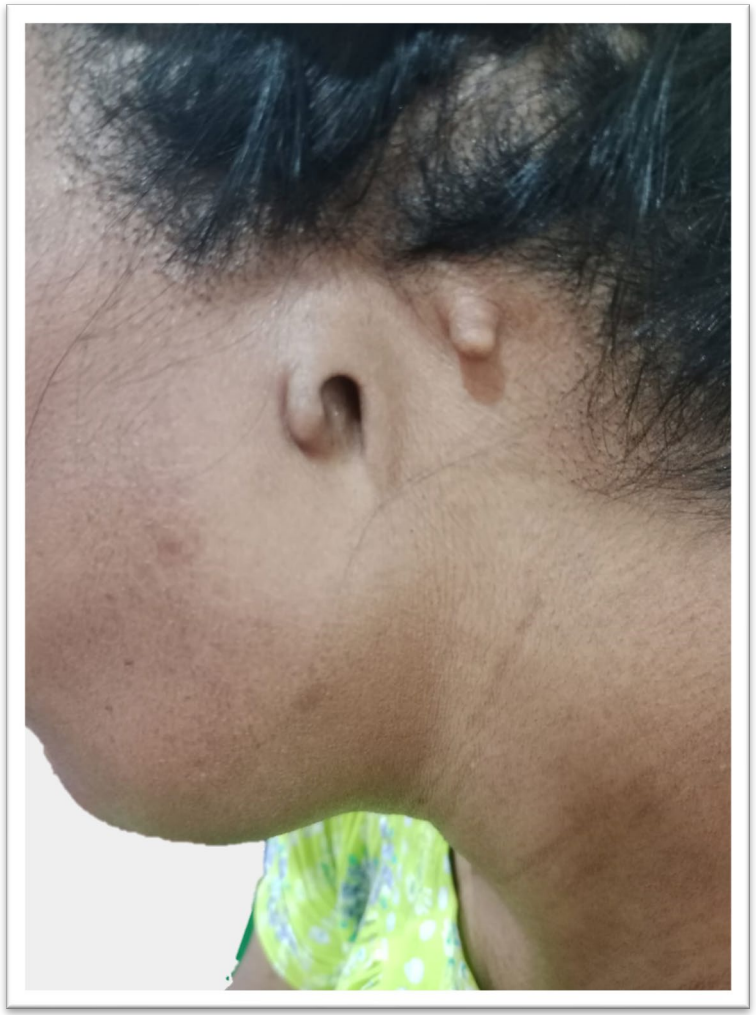
GENERAL EXAMINATION

- JVP : Not raised
- Lymph node : Not palpable
- Thyroid gland : Not enlarged
- Pigmentation : Absent
- Body hair distribution : Normal

GENERAL EXAMINATION

- Skin texture : Tightening, thickening and corrugation of both leg skin
- Cannula in situ in right hand
- Ophthalmoscopy : Normal
- Bed side urine : Negative for protein and sugar





Pictures taken with the consent from patient

SYSTEMIC EXAMINATION

MUSCULOSKELETAL SYSTEM

Examination of hand:

Inspection:

- Both wrist joints are swollen
- PIP joints of both hands are spindle shaped
- Swan neck deformity of right index & middle and left index finger
- Dorsal guttering in right hand

MUSCULOSKELETAL SYSTEM

- Z deformity of both thumbs and ulnar deviation of both hands
- Movements are restricted in both wrists and fingers of both hands with impaired functional activity
- Flexion deformity of fingers as evident by prayer sign
- Wasting of thenar and hypothenar muscle

MUSCULOSKELETAL SYSTEM

Palpation:

- All PIP and MCP joints of both hands are tender
- Synovial thickening is present in both wrist joints

MUSCULOSKELETAL SYSTEM

Examination of feet:

- All metatarsophalangeal and interphalangeal joints are swollen and deformed in both feet
- Lateral deviation of toes with some flexion deformity

MUSCULOSKELETAL SYSTEM

Examination of spine:

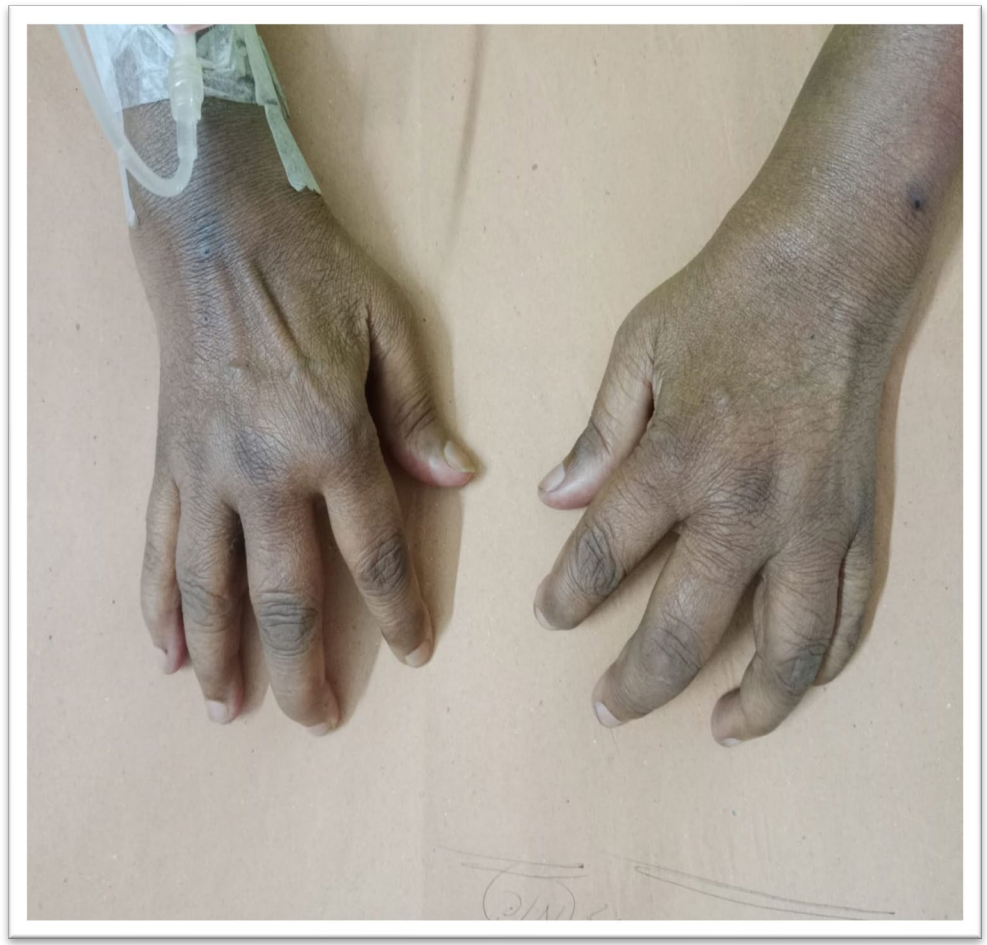
Lateral bending of dorsal spine is seen

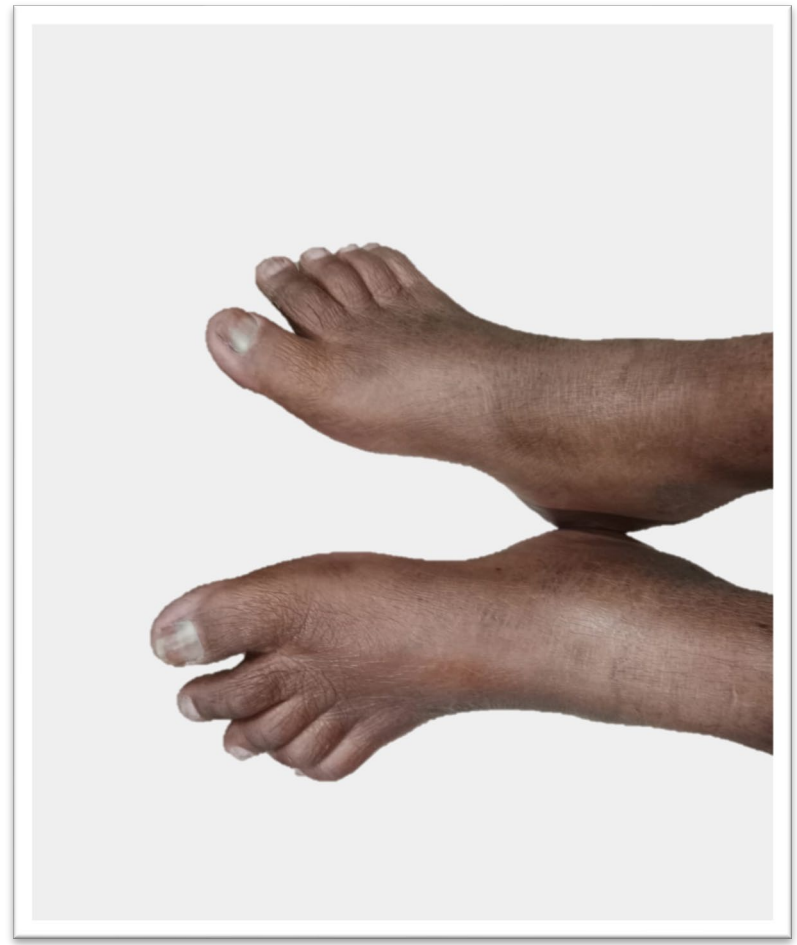
MUSCULOSKELETAL SYSTEM

Examination of other joints:

- Knee joints are swollen and tender
- There is no Baker or Popliteal cyst
- Elbow joints are swollen, tender with some flexion deformity

DAS 28 score- 7.09 (using ESR)





Pictures taken with the consent

RESPIRATORY SYSTEM

Inspection:

- Movement is restricted in left side
- No scar mark , deformity, visible impulse or engorged vein

RESPIRATORY SYSTEM

- **Palpation:**
    - Trachea is centrally placed
    - Chest expansibility is reduced in left side
    - Apex beat is palpable over left 5th intercostal space
- **Percussion**:
    - Resonant over both lung fields except area of liver dullness

RESPIRATORY SYSTEM

- **Auscultation**:
  - Breath sound is vesicular
  - Biphasic coarse crepitation over 6th intercostal space to downward in left side which was not altered with cough
  - Vocal resonance is normal over both lung fields

CARDIOVASCULAR SYSTEM

- **Inspection**:
    - No visible cardiac impulse and scar mark
- **Palpation**:
    - Apex beat is situated in left 5th intercostal space just medial to the midclavicular line
    - No thrill/heave/palpable P2

CARDIOVASCULAR SYSTEM

- **Auscultation**:
  - 1st and 2nd heart sound is audible in all four areas
  - There is a systolic murmur over tricuspid and pulmonary area with no radiation

GASTROINTESTINAL SYSTEM

Inspection :

- Abdomen is slightly distended
- Umbilicus centrally placed and inverted
- No visible peristalsis/ pulsation/ scar mark/ engorged veins/ pigmentation

GASTROINTESTINAL SYSTEM

Palpation :

- No tenderness/ organomegaly

Percussion :

- Dull in flanks
- Ascites was present as evidence by shifting dullness
- Upper border of liver dullness present in right 5^{th} intercostal space

GASTROINTESTINAL SYSTEM

Auscultation :

- Bowel sound present

OTHER SYSTEMIC EXAMINATION

Revealed no abnormalities

SALIENT FEATURES

SALIENT FEATURES

Mrs. X, 52 years old normotensive , non-diabetic, non-asthmatic muslim housewife, hailing from Gazipur, got admitted to Dhaka Medical College Hospital on 4th December 2024 with the complaints of pain and swelling in multiple joint of hands and feet for 5 years which involved both small and large joints.

SALIENT FEATURES

Severity of pain was increased over time that associated with morning stiffness which persists for about 4 hours and was relieved by activity.

She developed gradual swelling of both lower limbs for 1 year that had increased progressively over months with no history of anasarca.

SALIENT FEATURES

She also complained of shortness of breath that was mainly exertional ,progressively worsening , associated with fatigue and palpitation that hampered her daily activities with no history of orthopnoea.

.

SALIENT FEATURES

On examination she has facial asymmetry, incompletely developed external ear with pre & post auricular skin tags, average body built, severely anaemic, bipedal pitting oedema. Her BP was 120/70 mm of Hg with no postural drop, pulse was 112 b/min and regular.

SALIENT FEATURES

On musculoskeletal system examination, both wrist and elbow joints are swollen. PIP joints of both hands are spindle shaped. There is swan neck deformity of right index & middle and left index finger, Z deformity of both thumbs and ulnar deviation of both hands, flexion deformity of fingers as evident by prayer sign.

SALIENT FEATURES

Movements are restricted in both wrists, elbows and fingers of both hands.

All metatarsophalangeal and interphalangeal joints are swollen and deformed in both feet with lateral deviation of toes . Knee joints are swollen and tender.

SALIENT FEATURES

Examination of respiratory system revealed chest movement and expansibility is reduced in left side. Biphasic coarse crepitation over 6^{th} intercostal space to downward in left side which is not altered with cough.

On cardiovascular system , there is a systolic murmur over tricuspid and pulmonary area with no radiation.

SALIENT FEATURES

Examination of gastrointestinal system revealed, presence of ascites

PROBLEM LIST FROM AUDIENCE

Multiple joint pain

Leg swelling

Shortness of breath

Facial asymmetry

Absent external ear

Scoliosis

Problem list

Anaemia

Pitting oedema

Tenderness , swelling and deformity of joints

Coarse crepitation in left lung

Systolic murmur

WHAT COULD OUR DIAGNOSIS BE ?

PROVISIONAL DIAGNOSIS

Rheumatoid Arthritis with Interstitial lung disease with Goldenhar Syndrome

DIFFERENTIAL DIAGNOSIS

- Systemic lupus erythematosus
- Osteoarthritis

with Goldenhar Syndrome

INVESTIGATION PROFILE

COMPLETE BLOOD COUNT

Parameter	14/03/24	30/11/24	12/12/24
Haemoglobin	6.8 g/dl	6.0 g/dl	9.1 g/dl
MCV	47.9 fl	48.9 fl	56.5 fl
MCH	12.9 pg	13.4 pg	17.6 pg
MCHC	27.0 g/dl	27.4 g/dl	29.2 g/dl
ESR	65 mm in 1st hour	98 mm in 1st hour	55 mm in 1st hour
WBC	6920 /cmm	8220 /cmm	8900 /cmm
Neutrophils	72 %	82 %	78 %
Platelet	543000 /cmm	535000 /cmm	431000 /cmm

PERIPHERAL BLOOD FILM (30/11/24)

- RBC : Microcytic hypochromic with target, pencil , tear drop cell and few schistocytes
- WBC : Normal
- Platelets : Increased

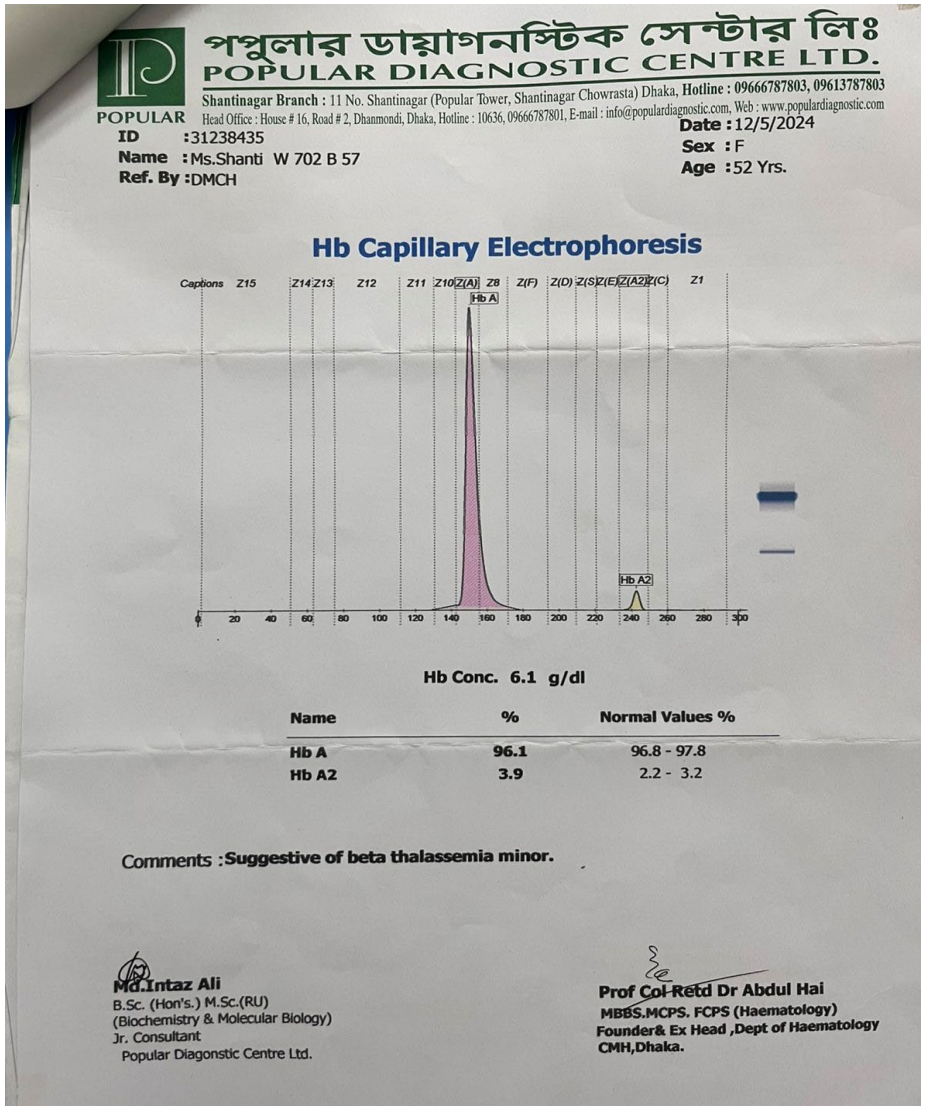
Comment : Suggestive of microcytic hypochromic anaemia with thrombocytosis

IRON PROFILE

Parameter	04/12/24	Normal value
S. Iron	78.80	37.0-170.0 µg/dL
TIBC	330.0	274-494.0 µg/dL
Tsat	23%	
Ferritin	9.45	15-120 ng/mL

HB. ELECTROPHORESIS (05/12/24)

Comment : Beta Thalassemia minor



পপুলার ডায়াগনস্টিক সেন্টার লিঃ
POPULAR DIAGNOSTIC CENTRE LTD.
Shantinagar Branch : 11 No. Shantinagar (Popular Tower, Shantinagar Chowrasta) Dhaka, Hotline : 09666787803, 09613787803
Head Office : House # 16, Road # 2, Dhanmondi, Dhaka, Hotline : 10636, 09666787801, E-mail : info@populardiagnostic.com, Web : www.populardiagnostic.com
POPULAR

ID :31238435
Name :Ms.Shanti W 702 B 57
Ref. By :DMCH

Date :12/5/2024
Sex :F
Age :52 Yrs.

Hb Capillary Electrophoresis

Hb Conc. 6.1 g/dl

Name	%	Normal Values %
Hb A	96.1	96.8 - 97.8
Hb A2	3.9	2.2 - 3.2

Comments :Suggestive of beta thalassemia minor.

Md.Intaz Ali
B.Sc. (Hon's.) M.Sc.(RU)
(Biochemistry & Molecular Biology)
Jr. Consultant
Popular Diagonstic Centre Ltd.

Prof Col Retd Dr Abdul Hai
MBBS.MCPS. FCPS (Haematology)
Founder& Ex Head ,Dept of Haematology
CMH,Dhaka.

BIOCHEMISTRY

Parameter	07/12/24	Normal value
Na+	143	135-145 mmol/L
K+	4.1	3.5-5.5 mmol/L
Cl-	103	97-107 mmol/L
S. Creatinine	1.29	0.6-1.40 mg/dl
S. Albumin	3.1	3.40-5.00 g/dl
NT-proBNP	1282.40	<125.0 pg/mL

BIOCHEMISTRY

Parameter	06/12/24	Normal value
RF	48.3	<15.9 IU/ml
ACPA	>200	<5 U/ml
CRP	47.5	Upto 3.0 mg/L
ANA	Not detected	
Anti dsDNA	2.83	<30 U/mL

BIOCHEMISTRY

Parameter	01/12/24	Normal value
S. TSH	2.05	0.35-5.50 µIU/ml
FT4	1.61	0.89-1.7 ng/dl
RBS	5.25	<7.8 mmol/L
SGPT	22	<40.0 U/L
HBsAg	Negative	Negative

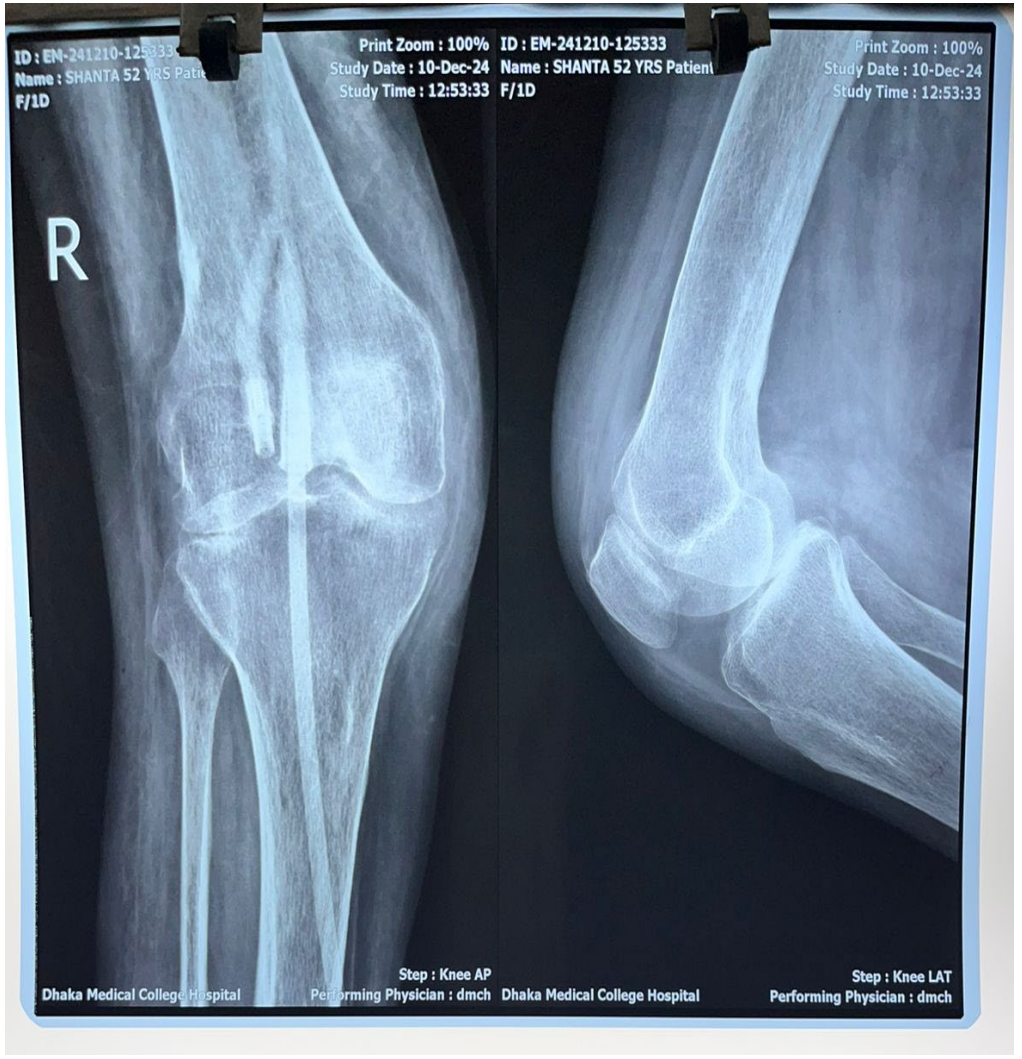
URINE R/E

Parameter	30/11/24	04/12/24
Protein	Trace	Nil
Glucose	Nil	Nil
Pus cell	0-2 /HPF	2-4 /HPF
RBC	Nil	Nil

- **24 hours UTP:** 0.15 g

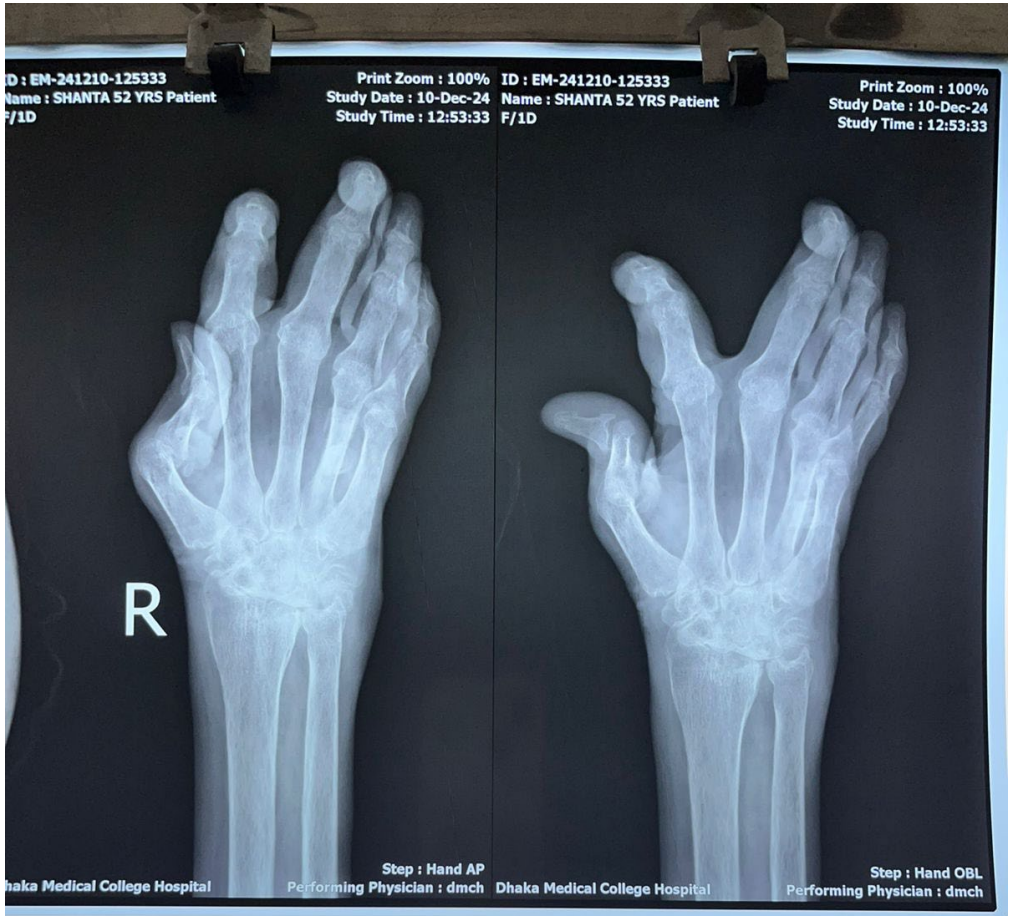
X-RAY OF RIGHT KNEE JOINT (11/12/24)

Comment: Features of Rheumatoid Arthritis of right knee



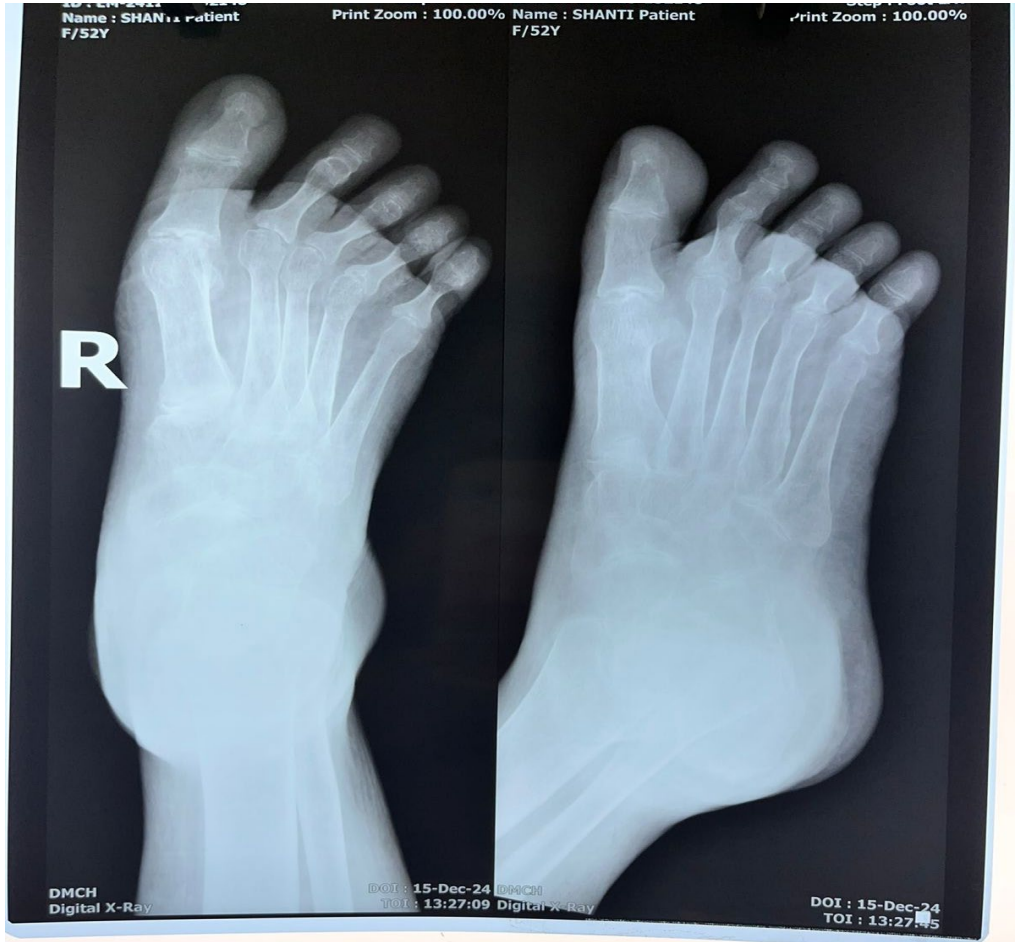
X-RAY OF HAND (13/12/24)

Comment: Features consistent with Rheumatoid arthritis of right hand



X-RAY OF RIGHT FOOT (13/12/24)

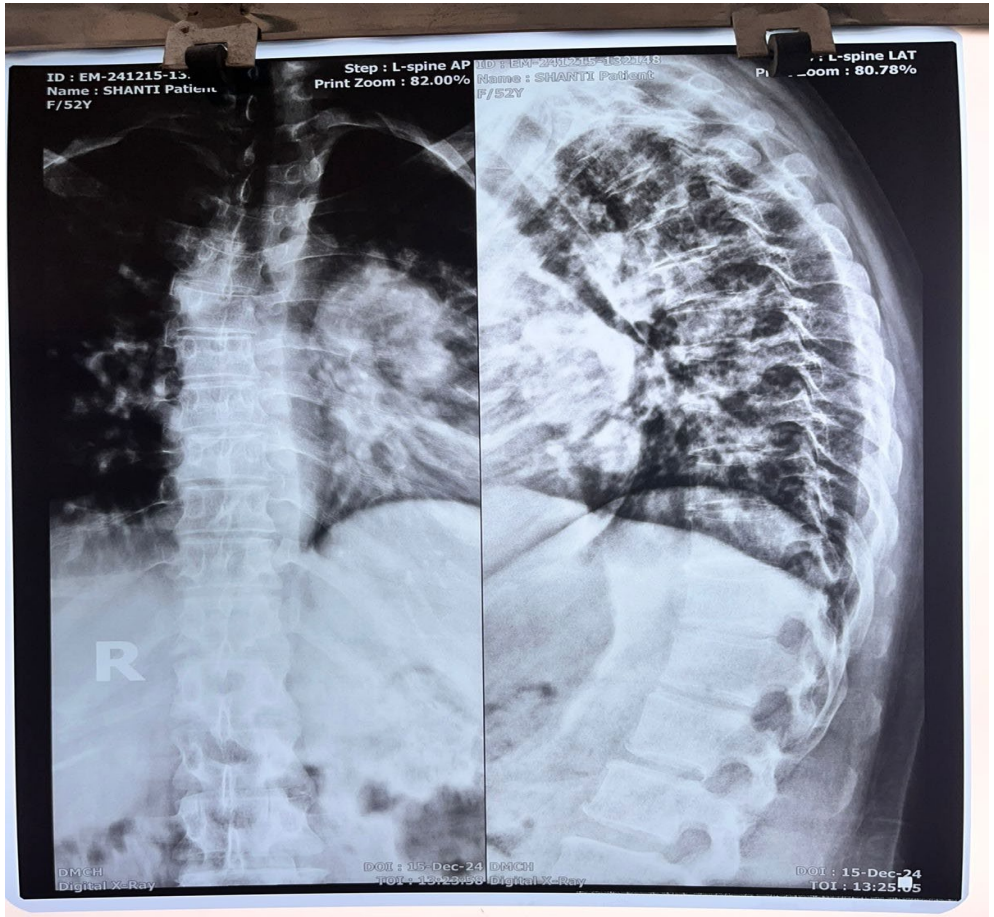
Comment : Features consistent with Rheumatoid arthritis of right foot



X-RAY OF DORSOLUMBAR SPINE (15/12/24)

Comment :

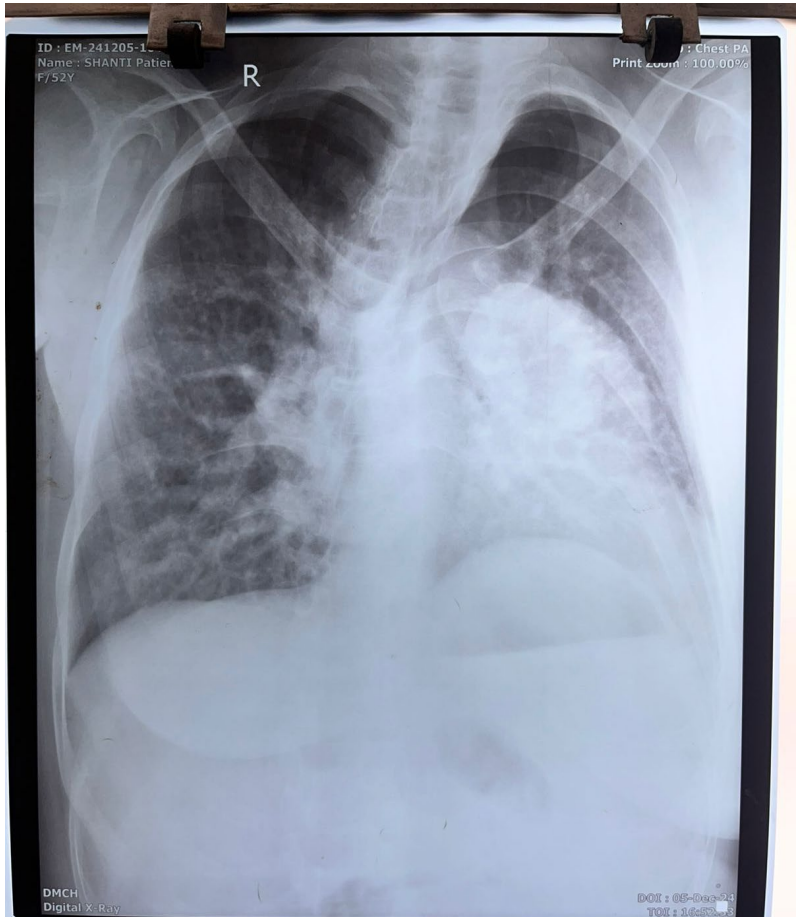
- Scoliosis
- Degenerative change in dorsal spine



CHEST X-RAY P/A VIEW (05/12/24)

Comment:

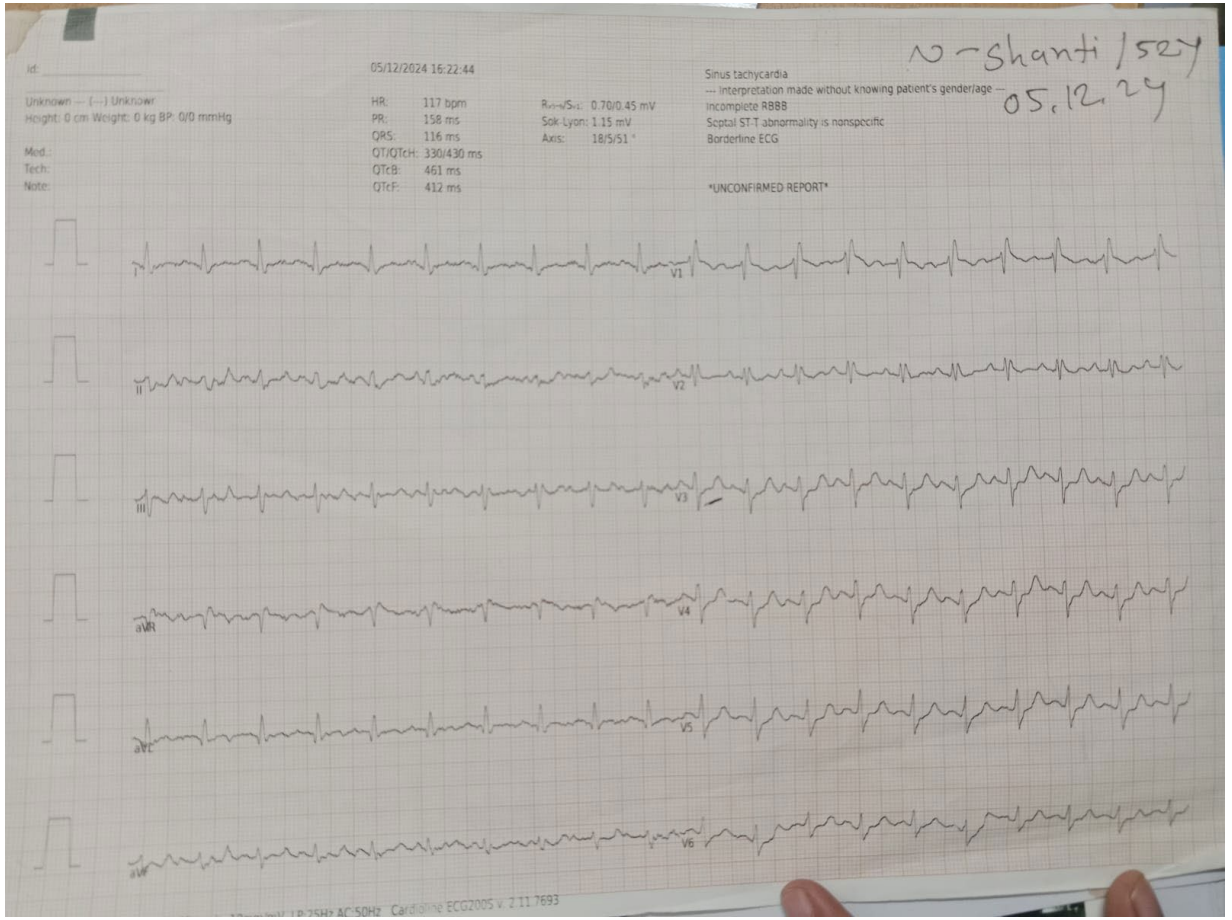
- Cardiomegaly with pulmonary arterial hypertension
- Scoliosis



ELECTROCARDIOGRAM (05/12/24)

Comment :

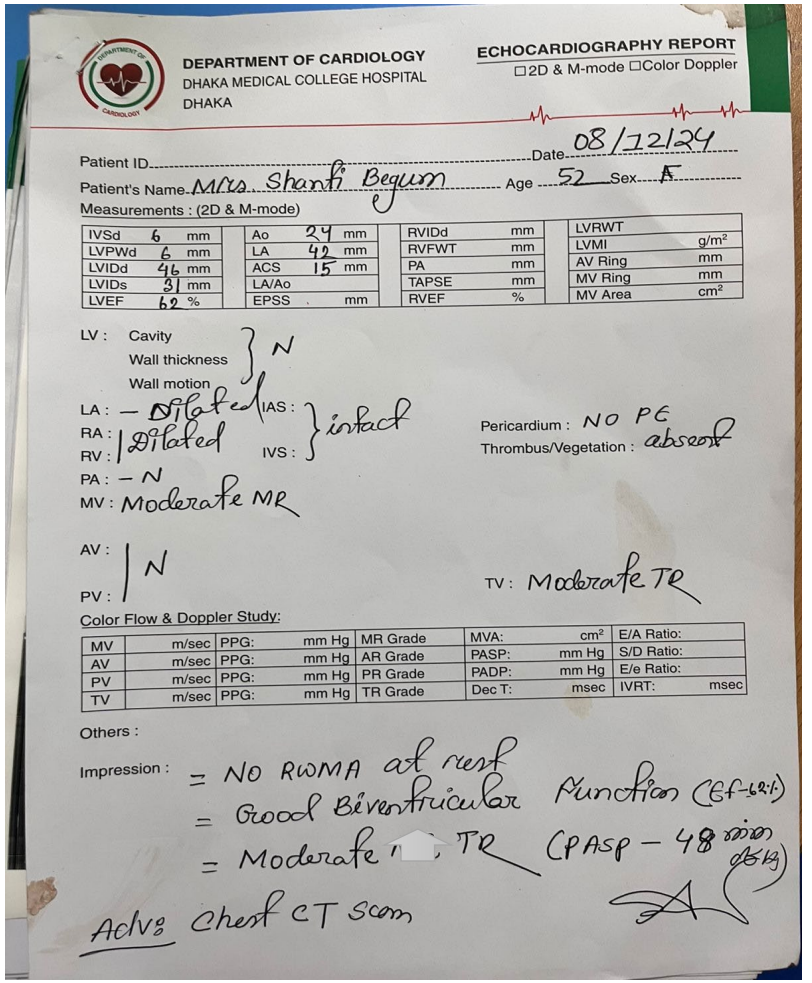
- Sinus tachycardia
- Incomplete RBBB



ECHOCARDIOGRAM (08/12/24)

Comment:

- Good biventricular function with EF-62%
- Moderate TR
- PASP – 48 mm of Hg



DEPARTMENT OF CARDIOLOGY
DHAKA MEDICAL COLLEGE HOSPITAL
DHAKA

ECHOCARDIOGRAPHY REPORT
☐2D & M-mode ☐Color Doppler

Patient ID.....
Date 08/12/24
Patient's Name Mrs. Shanti Begum Age 52 Sex F
Measurements : (2D & M-mode)

IVSd	6 mm	Ao	24 mm	RVIDd	mm	LVRWT	
LVPWd	6 mm	LA	42 mm	RVFWT	mm	LVMI	g/m²
LVIDd	46 mm	ACS	15 mm	PA	mm	AV Ring	mm
LVIDs	31 mm	LA/Ao		TAPSE	mm	MV Ring	mm
LVEF	62 %	EPSS	mm	RVEF	%	MV Area	cm²

LV : Cavity, Wall thickness, Wall motion } N
LA : – Dilated IAS : } intact
RA : } Dilated IVS :
RV :
PA : – N
MV : Moderate MR
AV :
PV : } N
TV : Moderate TR
Pericardium : NO PE
Thrombus/Vegetation : absent

Color Flow & Doppler Study:

MV	m/sec	PPG:	mm Hg	MR Grade	MVA:	cm²	E/A Ratio:
AV	m/sec	PPG:	mm Hg	AR Grade	PASP:	mm Hg	S/D Ratio:
PV	m/sec	PPG:	mm Hg	PR Grade	PADP:	mm Hg	E/e Ratio:
TV	m/sec	PPG:	mm Hg	TR Grade	Dec T:	msec	IVRT: msec

Others :

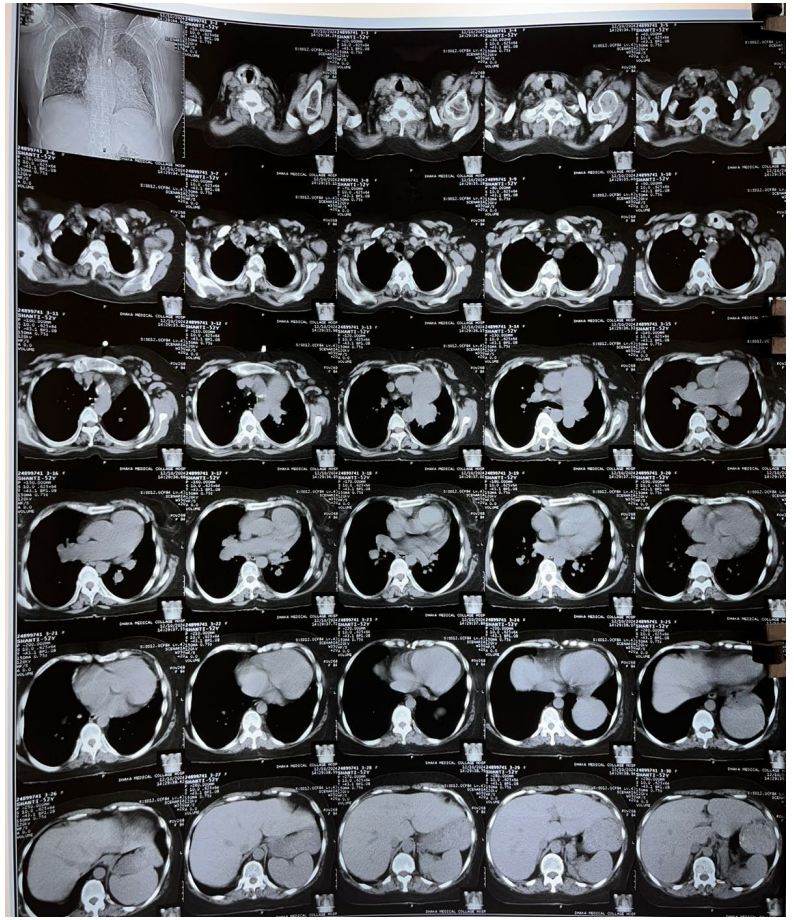
Impression : = NO RWMA at rest
= Good Biventricular Function (EF-62%)
= Moderate TR (PASP – 48 mm of Hg)

Advs: Chest CT scan

CT SCAN OF CHEST (10/12/24)

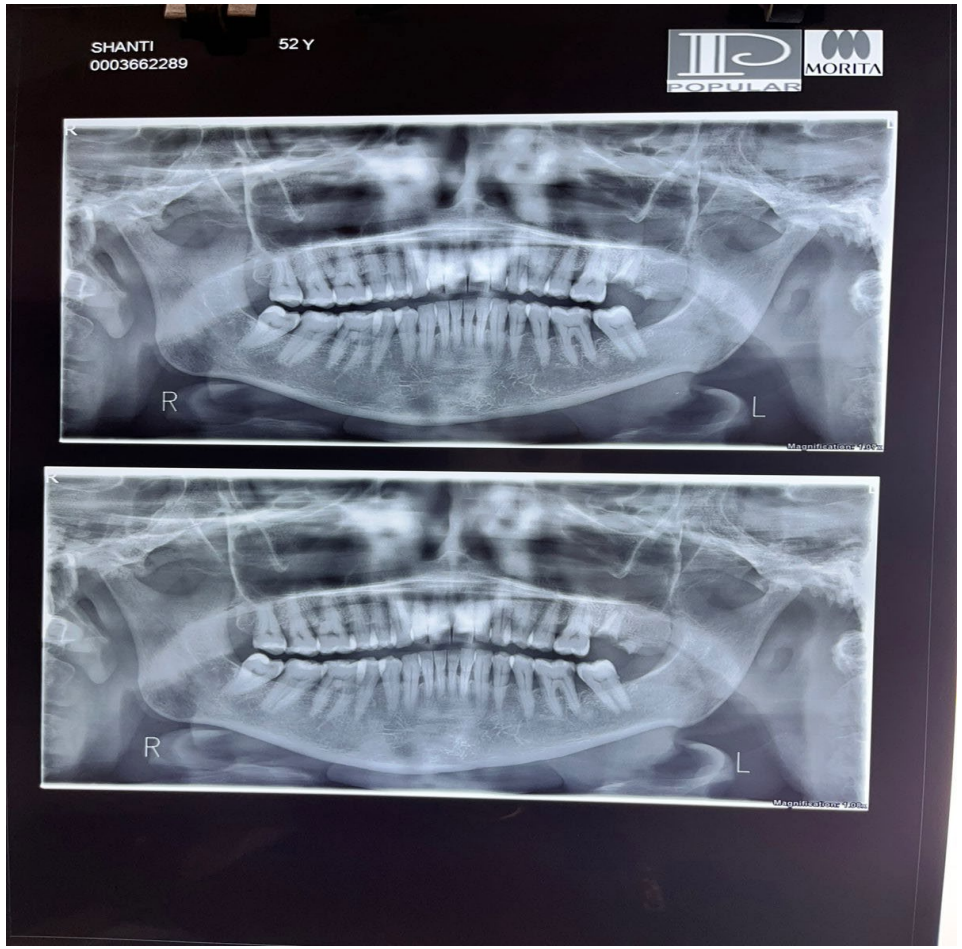
CT SCAN OF CHEST (10/12/24)

Comment : Cardiomegaly with dilated pulmonary vessels (more dilated pulmonary trunk)



ORTHOPANTOMOGRAM (15/12/24)

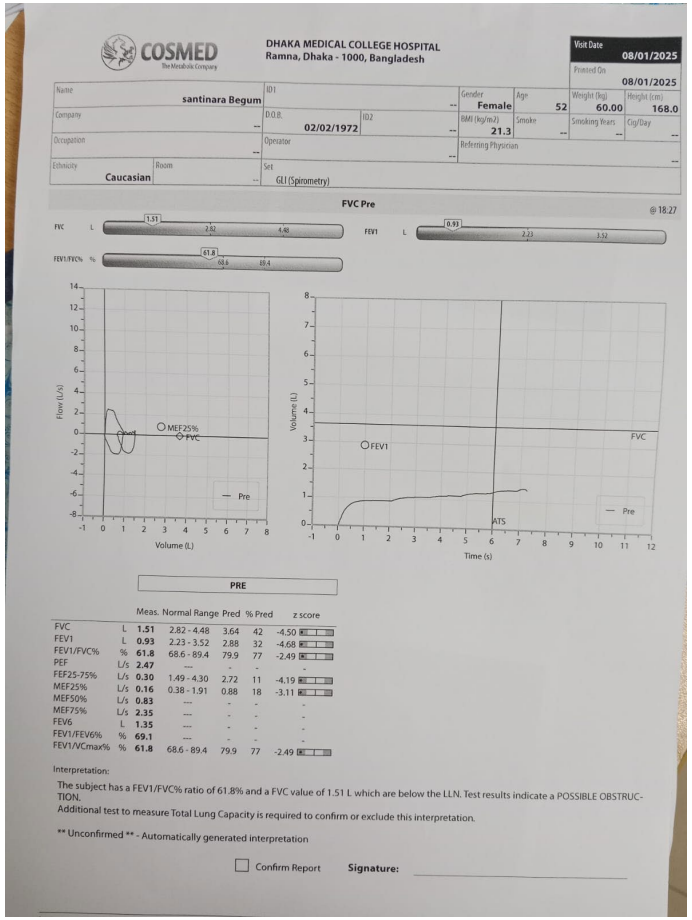
Comment : Short ramal height & width and reduced height of body of mandible in the left side



SPIROMETRY (08/01/25)

- FEV1/FVC ratio 61.8%
- FEV1 value 0.9 L
- FVC value 1.5 L

Comment: Possible obstruction



COSMED
The Metabolic Company

DHAKA MEDICAL COLLEGE HOSPITAL
Ramna, Dhaka - 1000, Bangladesh

Visit Date 08/01/2025
Printed On 08/01/2025

Name: santinara Begum	ID1: --	Gender: Female	Age: 52	Weight (kg): 60.00	Height (cm): 168.0
Company: --	D.O.B.: 02/02/1972	ID2: --	BMI (kg/m2): 21.3	Smoke: --	Smoking Years: --	Cig/Day: --
Occupation: --	Operator: --		Referring Physician: --			
Ethnicity: Caucasian	Room: --	Set: GLI (Spirometry)				

FVC Pre @ 18:27

FVC L 1.51 (2.82 - 4.48)
FEV1 L 0.93 (2.23 - 3.52)
FEV1/FVC% % 61.8 (68.6 - 89.4)

PRE

		Meas.	Normal Range	Pred	% Pred	z score
FVC	L	1.51	2.82 - 4.48	3.64	42	-4.50
FEV1	L	0.93	2.23 - 3.52	2.88	32	-4.68
FEV1/FVC%	%	61.8	68.6 - 89.4	79.9	77	-2.49
PEF	L/s	2.47	---	-	-	-
FEF25-75%	L/s	0.30	1.49 - 4.30	2.72	11	-4.19
MEF25%	L/s	0.16	0.38 - 1.91	0.88	18	-3.11
MEF50%	L/s	0.83	---	-	-	-
MEF75%	L/s	2.35	---	-	-	-
FEV6	L	1.35	---	-	-	-
FEV1/FEV6%	%	69.1	---	-	-	-
FEV1/VCmax%	%	61.8	68.6 - 89.4	79.9	77	-2.49

Interpretation:
The subject has a FEV1/FVC% ratio of 61.8% and a FVC value of 1.51 L which are below the LLN. Test results indicate a POSSIBLE OBSTRUCTION.
Additional test to measure Total Lung Capacity is required to confirm or exclude this interpretation.

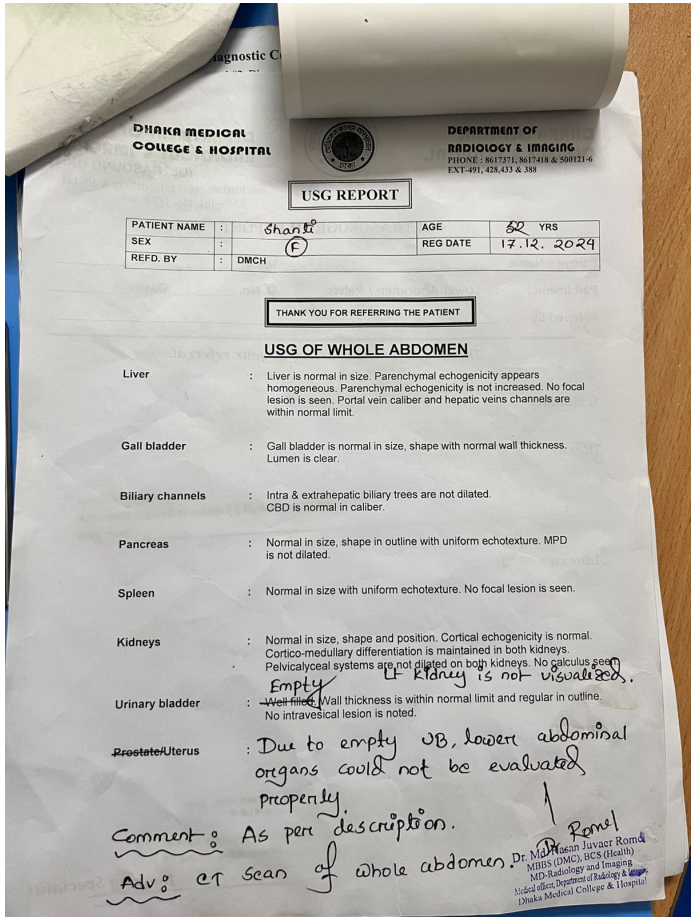
** Unconfirmed ** - Automatically generated interpretation

☐ Confirm Report Signature: _____

USG OF WHOLE ABDOMEN (17/12/24)

Comment:

Normal finding



DHAKA MEDICAL COLLEGE & HOSPITAL

DEPARTMENT OF RADIOLOGY & IMAGING
PHONE : 8617371, 8617418 & 500121-6
EXT-491, 428,433 & 388

USG REPORT

PATIENT NAME	:	Shanti		AGE	52 YRS
SEX	:	(F)		REG DATE	17.12.2024
REFD. BY	:	DMCH			

THANK YOU FOR REFERRING THE PATIENT

USG OF WHOLE ABDOMEN

Liver	:	Liver is normal in size. Parenchymal echogenicity appears homogeneous. Parenchymal echogenicity is not increased. No focal lesion is seen. Portal vein caliber and hepatic veins channels are within normal limit.
Gall bladder	:	Gall bladder is normal in size, shape with normal wall thickness. Lumen is clear.
Biliary channels	:	Intra & extrahepatic biliary trees are not dilated. CBD is normal in caliber.
Pancreas	:	Normal in size, shape in outline with uniform echotexture. MPD is not dilated.
Spleen	:	Normal in size with uniform echotexture. No focal lesion is seen.
Kidneys	:	Normal in size, shape and position. Cortical echogenicity is normal. Cortico-medullary differentiation is maintained in both kidneys. Pelvicalyceal systems are not dilated on both kidneys. No calculus seen. Lt kidney is not visualized.
Urinary bladder	:	~~Well filled~~ Empty. Wall thickness is within normal limit and regular in outline. No intravesical lesion is noted.
~~Prostate~~/Uterus	:	Due to empty UB, lower abdominal organs could not be evaluated properly.

Comment: As per description.

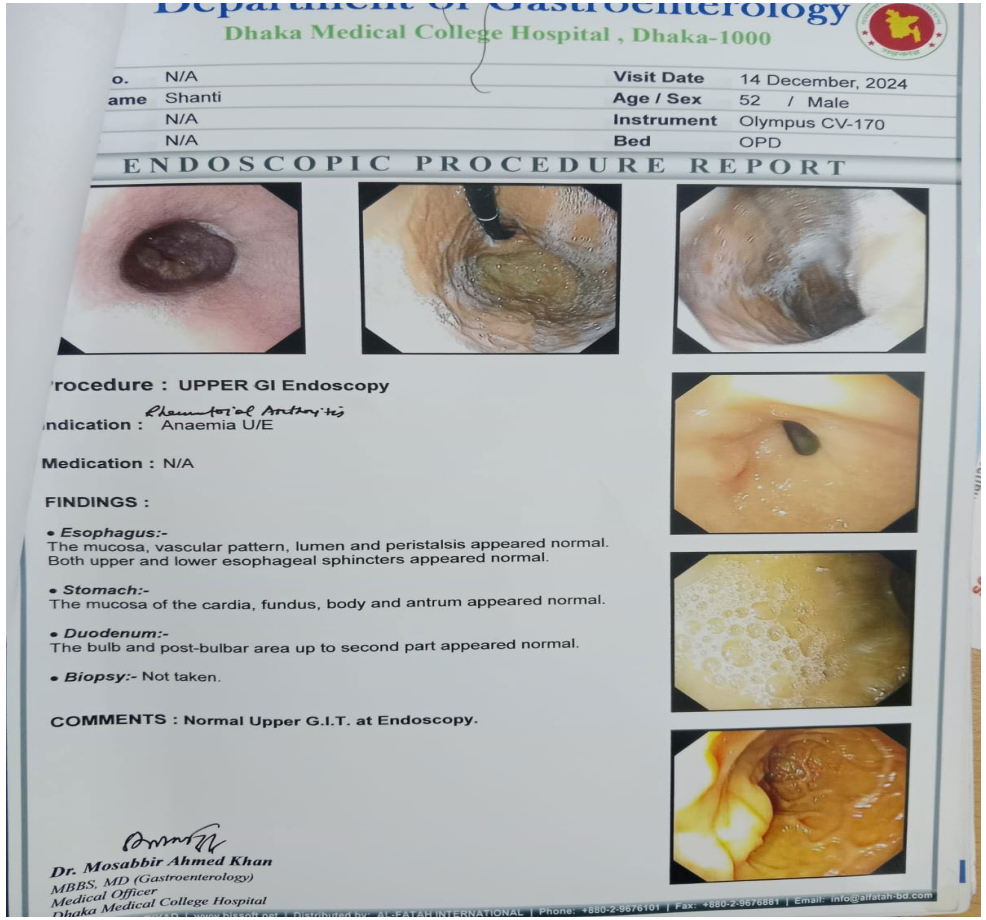
Adv: CT scan of whole abdomen.

Dr. Md. Hasan Juvaer Romel
MBBS (DMC), BCS (Health)
MD-Radiology and Imaging
Medical officer, Department of Radiology & Imaging
Dhaka Medical College & Hospital

UPPER GI ENDOSCOPY (14/12/24)

Comment:

Normal findings



Department of Gastroenterology
Dhaka Medical College Hospital , Dhaka-1000

o.	N/A	Visit Date	14 December, 2024
ame	Shanti	Age / Sex	52 / Male
	N/A	Instrument	Olympus CV-170
	N/A	Bed	OPD

ENDOSCOPIC PROCEDURE REPORT

rocedure : UPPER GI Endoscopy

ndication : Rheumatoid Arthritis
Anaemia U/E

Medication : N/A

FINDINGS :

- *Esophagus:-*
The mucosa, vascular pattern, lumen and peristalsis appeared normal.
Both upper and lower esophageal sphincters appeared normal.
- *Stomach:-*
The mucosa of the cardia, fundus, body and antrum appeared normal.
- *Duodenum:-*
The bulb and post-bulbar area up to second part appeared normal.
- *Biopsy:-* Not taken.

COMMENTS : Normal Upper G.I.T. at Endoscopy.

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Medical Officer
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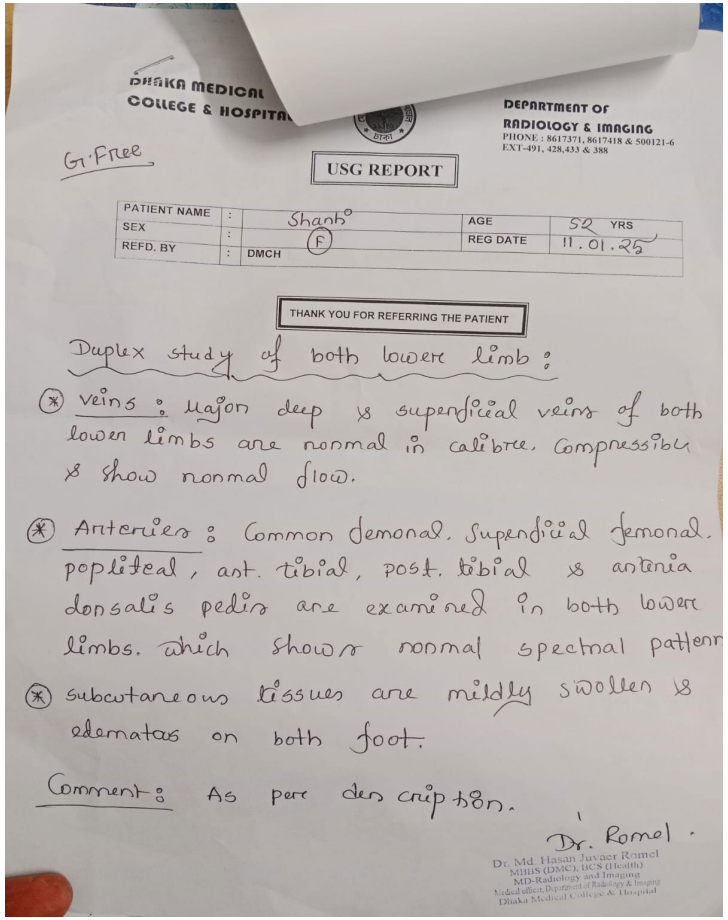
DUPLEX STUDY OF BOTH LOWER LIMBS (11/01/25)

- Veins- Normal in caliber, compressible and flow
- Arteries- Normal finding
- Subcutaneous tissues are mildly edematous and swollen on both foot



DHAKA MEDICAL COLLEGE & HOSPITAL

DEPARTMENT OF RADIOLOGY & IMAGING
PHONE : 8617371, 8617418 & 500121-6
EXT-491, 428,433 & 388

G-Free

USG REPORT

PATIENT NAME	:	Shanto		AGE	52 YRS
SEX	:	(F)		REG DATE	11.01.25
REFD. BY	:	DMCH			

THANK YOU FOR REFERRING THE PATIENT

Duplex study of both lower limb :

(*) Veins : Major deep & superficial veins of both lower limbs are normal in calibre, compressible & show normal flow.

(*) Arteries : Common femoral, superficial femoral, popliteal, ant. tibial, post. tibial & arteria dorsalis pedis are examined in both lower limbs. which shows normal spectral pattern

(*) subcutaneous tissues are mildly swollen & edematas on both foot.

Comment : As per description.

Dr. Romel

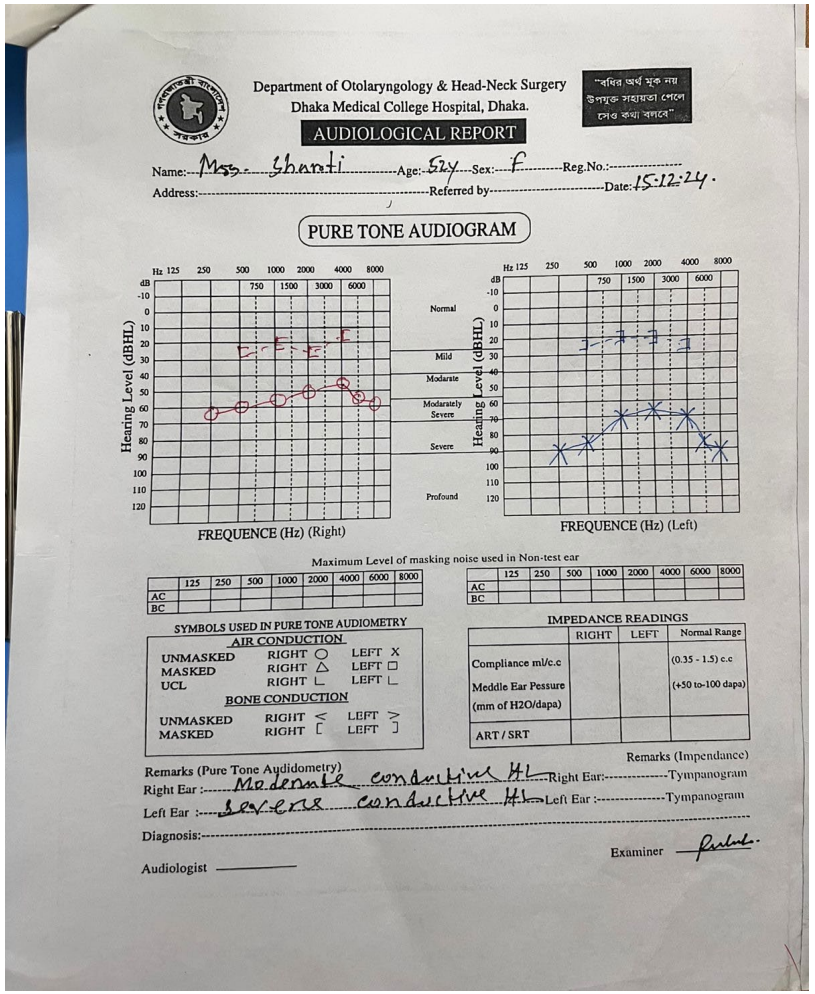
Dr. Md. Hasan Juyaer Romel
MBBS (DMC), BCS (Health)
MD-Radiology and Imaging
Medical Officer, Department of Radiology & Imaging
Dhaka Medical College & Hospital

PTA AND SRT (15/12/24)

Right ear- Moderate conductive hearing loss

Left ear- Severe conductive hearing loss

SRT: Absent



Department of Otolaryngology & Head-Neck Surgery
Dhaka Medical College Hospital, Dhaka.

AUDIOLOGICAL REPORT

"বধির অর্থ মূক নয়
উপযুক্ত সহায়তা পেলে
সেও কথা বলবে"

Name: Mrs. Shanti Age: 52y Sex: f Reg.No.:
Address: Referred by: Date: 15.12.24.

PURE TONE AUDIOGRAM

Hz 125 250 500 1000 2000 4000 8000
dB -10 0 10 20 30 40 50 60 70 80 90 100 110 120
750 1500 3000 6000

Hearing Level (dBHL)

Normal
Mild
Moderate
Moderately Severe
Severe
Profound

FREQUENCE (Hz) (Right)

FREQUENCE (Hz) (Left)

Maximum Level of masking noise used in Non-test ear

	125	250	500	1000	2000	4000	6000	8000
AC								
BC								

	125	250	500	1000	2000	4000	6000	8000
AC								
BC								

SYMBOLS USED IN PURE TONE AUDIOMETRY

AIR CONDUCTION

UNMASKED	RIGHT ○	LEFT X
MASKED	RIGHT △	LEFT □
UCL	RIGHT ∟	LEFT ∟

BONE CONDUCTION

UNMASKED	RIGHT <	LEFT >
MASKED	RIGHT [	LEFT]

IMPEDANCE READINGS

	RIGHT	LEFT	Normal Range
Compliance ml/c.c			(0.35 - 1.5) c.c
Meddle Ear Pessure (mm of H2O/dapa)			(+50 to-100 dapa)
ART / SRT			

Remarks (Pure Tone Audiometry)
Right Ear : Moderate conductive HL
Left Ear : Severe conductive HL

Remarks (Impendance)
Right Ear: Tympanogram
Left Ear : Tympanogram

Diagnosis:

Audiologist

Examiner

FINAL DIAGNOSIS

Rheumatoid Arthritis with Heart failure with preserved ejection fraction with Pulmonary hypertension with Goldenhar Syndrome with Beta Thalassemia Minor

TREATMENT GIVEN

- Methotrexate
- Folic acid
- Prednisolone
- NSAIDs
- Furosemide

Goldenhar Syndrome

Goldenhar syndrome is a syndrome of complex structures developing from first and second branchial arches during blastogenesis. The etiology of this rare disease is not fully understood, as it has shown itself variable genetically and of unclear causes. The disorder is characterized by a wide spectrum of symptoms and physical features that may vary greatly in range and severity from case to case.

Case Report

Goldenhar Syndrome: A rare case report

Ruchi Bhuyan, Abhishek Ranjan Pati[1], Sanat Kumar Bhuyan[1], Bikash Bishwadarshee Nayak[1]

Departments of Oral Pathology and Microbiology and [1]Oral Medicine and Radiology, Siksha O Anusandhan University, Khandagiri, Bhubaneswar, Odisha, India

Abstract Goldenhar Syndrome or oculoauriculovertebral spectrum is a complex syndrome characterized by an association of maxillomandibular hypoplasia, deformity of the ear, ocular dermoid and vertebral anomalies and the most severe form of hemifacial microsomia. Here, we describe a 26-year-old male patient with unilateral hemifacial microsomia, preauricular ear tags, macrosomia on the right side of the face.

Key Words: Hemifacial microsomia, ocular dermoid, preauricular ear tags

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Received: 11.07.2015, **Accepted:** 30.05.2016

INTRODUCTION

In 1881, the first observation of oculo-auriculo-vertebral (OAV) dysplasia was reported by Von Atlt[1] and in 1952, Dr. Maurice Goldenhar, a renowned Swiss ophthalmologist classified the clinical features and named the malformation complex as Goldenhar Syndrome and described it as a congenital defect characterized by constellation of malformations classically involving the face, eyes and ears.[2]

In 1960, Gorlin and Pindborg noted that the disorder varied from mild to severe and the facial involvement occurred unilaterally as well as bilaterally. In 1963, Gorlin *et al.* suggested the use of the term oculoauricularvertebral dysplasia to describe the syndrome characterized by epibulbar dermoids, auricular appendages, blind-ended auricular fistulas and vertebral anomalies.[3] They considered this syndrome to be a variant of this complex.

It is otherwise known as oculoauriculovertebral syndrome, hemifacial microsomia, first arch syndrome, first and second branchial arch syndrome, Goldenhar–Gorlin Syndrome, lateral facial dysplasia, unilateral craniofacial microsomia, otomandibular dysostosis, unilateral intrauterine facial necrosis and auriculo-branchiogenic dysplasia., Facio-auricular dysplasias represent a single disorder with great variability of expression and an isolated ear malformation may represent the mildest expression of the disorder.[4]

Abnormalities are unilateral in 85% of cases and bilateral in 10–33% of the cases and the right side is more frequently affected.[5] The incidence of Goldenhar Syndrome has been reported to be varying from 1:3500 to 1:5600 live births and it is present in 1:1000 children with congenital deafness with a male to female ratio of 3:2.[6] The disease is seen as sporadic and its etiology is not fully understood; however, positive family histories have been reported suggesting autosomal dominant or recessive inheritance. Some researchers have suggested that multifactorial inheritance are caused by the interaction of many genes, possibly in combination with environmental factors.[7]

Patients with Goldenhar Syndrome may exhibit a wide range of anomalies as described in Table 1.[8-22]

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Table 1: Anomalies associated with Goldenhar Syndrome

Dentofacial abnormalities	Unilateral facial hypoplasia
	Facial bones
	Prominent forehead[8]
	Hypoplasia of the zygomatic area
	Maxillar and mandibular hypoplasia[9]
	Mouth
	Lateral facial cleft (unilateral macrostomia)[9]
	Facial paresis[10]
	Intraoral findings
	Cleft lip, cleft palate, tongue cleft, unilateral tongue hypoplasia[10]
	Gingival Hypertrophy[9,10]
Salivary glands	Aplasia of parotid gland[8]
Ear	Preauricular skin tags, fistulas and nodes[8]
	Preauricular dimples to imperforated external acoustic meatus, deafness[11]
	Posteriorly angulated ears[9,10]
	Pretragal anotia[12]
	Bilateral preauricular pits[13]
	Other external ear malformations - dysplasias and asymmetries
	Aplasias and atresias of the external meatus
	Middle and internal ear anomalies[13]
Eye	Epibulbar dermoid (unilateral/bilateral)[13,14]
	Hypertelorism, ptosis[14,15]
	Small eye with notched upper lid,[15] short palpebral fissures, hypoplasia of supraorbital ridges[11,13]
	Prominent eyes with cloudy corneas[13]
	Juvenile glaucoma[16]
	Bilateral epicanthal folds[15]
	Lipodermoidoid (mostly bilateral)[17]
	Colobomas of the upper eyelid iris, choroidea and retina[18,19]
	Other eye anomalies
	Antimongoloid palpebral fissures
	Anophthalmia
	Cataract
	Blepharophimosis[20]
Vertebral column anomalies	Atlas occipitalization
	Synostosis. hemivertebrae
	Fused vertebrae,
	Kyphoscoliosis
	Bifid spine[20]
	Butterfly vertebrae[11]
	Equinovarus deformity[10]
	Missing ribs
	Clinodactyly of the 5th fingers
	Short stubby fingers with ulnar deviation, hypoplastic distal phalanges.
	Club feet with prominent heel[13]
Other systemic abnormalities	Rib anomalies and anomalies of the extremities
	Congenital heart disease (ventricular septal defect, atrial septal defect, pulmonary stenosis)[20]
	Growth retardation, marked hair on body,[11] torticollis,[21] upturned nose,[22] flat nasal bridge with broad nasal root,[13] deep creases on the feet[11]
	Severe respiratory distress due to upper respiratory tract infection, tracheoesophageal fistulas[10,13]
	Urogenital and gastrointestinal system: ectopic kidneys[20]
	Uretropelvic junction obstruction, imperforate anus
	Central nervous system occipital encephalocele[20]
	Anomalies of the larynx and lungs tracheoesophageal fistula esophageal atresia
	Retardation of mental development[20]

CASE REPORT

A 25-year-old male patient reported to the Department of Oral Medicine and Radiology with the chief complaint of malalignment of teeth since childhood. History revealed that the patient had difficulty in hearing from the left ear since childhood.

On extra-oral examination, a transverse cleft was present in right angle of mouth, with preauricular tissue tags and incompetent lips. Bilateral asymmetry of face and macrostomia was present and inter-incisal distance was 43 mm. On intraoral examination, 16 and 47 were missing with spacing between 11 12 13 14 42 43 44, proclined 11 12 13 43, cross bite in relation to 24 34, increased overjet and high arched palate [Figures 1-5].

Correlating the history and clinical findings a provisional diagnosis of hemifacial microsomia was given. Differential diagnosis of Goldenhar Syndrome was considered but could be difficult because of the variety of clinical signs. Collins and

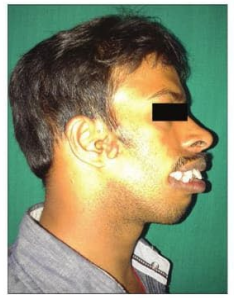

Figure 1: Lateral profile view of the patient showing preauricular skin tags, nodes and unilateral macrosomia

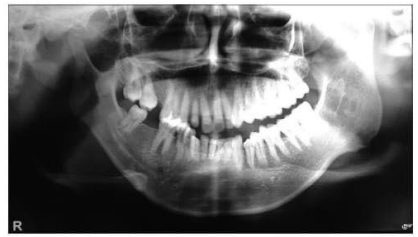

Figure 2: Orthopantomogram showing underdeveloped mandible on the right side

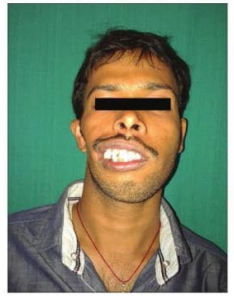

Figure 3: Front view demonstrating unilateral macrosomia, hypoplasia of the zygomatic area

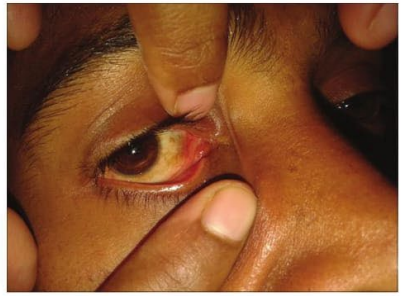

Figure 4: Medial aspect of eye demonstrating epibulbar dermoid

Wildervanck Syndrome (*Syndroma cervicoocuioacusticum*) must also be distinguished. Goldenhar Syndrome can be classified as a variant of hemifacial microsomia, OAV spectrum, or OAV Dysplasia by some authors.[20]

Other craniofacial anomalies such as Treachers Collin's Syndrome, mandibulofacial dysostosis,[23] Miller Syndrome, a very rare genetic condition, often referred to as "postaxial acrofacial dysostosis" must also be ruled out. These disorders are characterized by distinctive craniofacial malformations that occur in association with limb abnormalities.[24]

Investigations were done where OPG revealed reduction in size of body of mandible, condyles, coronoid process and ramus on the right side. The border of the mandible was well defined and intact. The trabecular pattern and the course of mandibular canal were normal with normal surrounding structures. PA view of skull was taken to rule out any developmental anomaly. It revealed reduction in size of body of mandible, condyles, coronoid process and ramus on the right side of mandible with normal surrounding structures. Lateral cephalogram was taken which revealed proclined maxillary incisors with increased overjet. Impedance Audiometry revealed hearing sensitivity of right ear to be in normal range whereas severe loss of hearing was noted in left ear. Consultation with the Ophthalmologist was done, who suggested normal eye morphology with no abnormality. Ultrasonography of abdomen was done which revealed no abnormality.

Correlating the history, clinical findings and investigative findings a final diagnosis of Goldenhar Syndrome was given.

The patient was then referred to the Department of Orthodontics for his orthodontic treatment, where orthodontic brackets were fixed on his maxillary teeth with nickel-titanium wire [Figure 6].

DISCUSSION

Most of the patients reported within the OAV spectrum, a term proposed by Gorlin *et al.* (1990), are sporadic.[3] As similar to previously reported cases, our patient had marked right facial hypoplasia with the chin slightly deviated to the affected side.

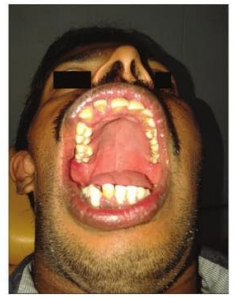

Figure 5: Intra-oral view showing high-arched palate

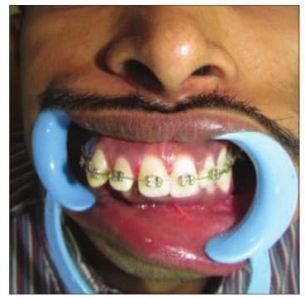

Figure 6: Patient started with orthodontic alignment

The association of epibulbar dermoids, preauricular fistulae, abnormalities of skin appendages and ocular malformations as a specific entity involving the first and second branchial arches was recognized by Goldenhar.[2] Pretragal fistulae, epibulbar dermoids and accessory auricular appendages are triad of Goldenhar Syndrome. The diagnosis is further substantiated if vertebral anomalies are present in addition to the triad. The diagnosis becomes more difficult if additional symptoms are present.

Cranial nerve involvement has been considered part of the OAV spectrum and the facial nerve involvement is cited most commonly.[25] Congenital ophthalmoplegia associated with Goldenhar Syndrome was reported in only one instance, concerning two patients with paralysis of one or more extraocular eye movements on neurologic examination. A third patient presented with unilateral agenesis of trochlear and abducens nerves and corresponding brain stem nuclei demonstrated at autopsy.[26] Brain stem involvement detected by magnetic resonance imaging in patients with Goldenhar Syndrome has been also reported; however, the stem lesions are very severe, resulting in a poor general prognosis.[27]

Some other etiologic factors include maternal vasoactive medication use (especially in conjunction with smoking) in the first 10 weeks of gestation, primidone, retinoic acid and thalidomide embryopathy and maternal (preexisting or gestational) diabetic embryopathy.[28]

In the 2nd month of embryonic development, aberrant fusion of the lateral portions of the maxillary and mandibular swellings results in macrostomia, usually associated with skin tags and pits between the comer of the mouth and the tragus.[29]

The prevalence of Goldenhar Syndrome in Indian population is very low. A study was taken up to understand the prevalence of this syndrome in children below the age of 14 years with hearing loss. Out of 1073 children, this syndrome was observed only in 1 (0.09%) case.[30]

The effect of Goldenhar Syndrome is more evident as the child grows, because of delays in the growth and the development of the affected areas. The lack of development of the jaws can cause breathing problems, as well as dental malocclusion which requires multidisciplinary approach.[5] Timing of the reconstruction plays an important role in the treatment. Primary reconstruction typically consists of a cleft repair, corrections of colobomas and ear deformities and removal of dermoids and preauricular tags. [31] The complex treatment is focused not only on dental care, articulation and hearing but also on the prevention and treatment of the psychosocial aspects of the malformation. Treatment requires constant follow-up and reassessment of the results.[23]

The study of this condition is still controversial because the symptoms and the physical features may vary greatly in range and severity from case to case.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

WHAT HAVE WE FOUND IN OUR PATIENT ?

- Unilateral facial hypoplasia
- Microstomia
- Pre and post auricular skin tags with external ear malformation
- Hypoplasia of left supraorbital ridge
- Scoliosis

ACKNOWLEDGEMENT

- Department of Cardiology, DMCH
- Department of ENT, DMCH
- Department of Radiology and Imaging, DMCH
- Department of Respiratory Medicine , DMCH
- Department of Neurology , DMCH

MEDICINE UNIT 5


THANK
YOU