


ID

- ❖ A 27-year-old woman
- ❖ From Bukan
- ❖ Source of history: patient, reliable
- ❖ She has been referred for endocrine evaluation after diagnosis of Carney complex

PI

- ❖ Six years ago: headache, vertigo & right-side hemiplegia → CVA was diagnosed
- ❖ Investigation for the CVA cause → cardiac myxomas were diagnosed (left atrium: 3.4 x 3 cm; right ventricle: 0.9 x 1 cm)
- ❖ In 1394, she underwent open heart surgery for myxoma removal.
- ❖ Two years later, she was evaluated again because of dyspnea & cardiac arrhythmia and recurrence of myxomas (left atrium: 3.2 x 2.7 cm; right ventricle: 1 x 1 cm) was documented.

PI

- ❖ In 1396, the second open heart surgery with incomplete resection was done. Metoprolol and Clopidogrel were prescribed.
- ❖ After one year, she was evaluated because of palpitation & EPS was done.
- ❖ In 1398, headache and nausea  myxoma recurrence (left atrium: 3 x 3 cm; right ventricle: 2 x 2 cm); in Shahid Rajaei hospital, open heart surgery was done for tumor removal and tricuspid valve repair.

In the general evaluation of the patient, some skin pigmentations and skin lesions were detected. Excisional biopsy was done.

دانشگاه علوم پزشکی تهران
بیمارستان فوق تخصصی پوست رازی

بخش باتولوژی / درمان باتولوژی / گزارش هیستوپاتولوژی

پارسیان رازی
صفحه ۱ از ۱

شماره	۹۸۰۷۹۸۵	عشقمات بیمار	[REDACTED]	باتولوژیست	[REDACTED]
شماره نمونه	۹۸-۶۸۷۵	آدرس بیمار	(۹۱۴۱۸۱۵۳۰۳)	تاریخ پذیرش	۱۳۹۸/۰۶/۳۰
پرونده	۶۱۶۸۴۶۷	نوع بیمه	بیمه همگانی شهرداری (سنواتی) (شماره=۲۹۲۰۲۷۶۶۳۱)	تاریخ جوابدهی	۱۳۹۸/۰۶/۲۷
شماره مدارک پزشکی	۹۸-۰۱۹۵۰۷	پزشک معالج	بدرام نور محمد پور	کد ملی	۲۹۲۰۲۷۶۶۳۱

[Clinical Impression]
Shawnomma -Melanocytic nevus -Other soft tissue tumor -Adnexal tumor - Lymphangioma

[Macroscopic Description]
specimen received in formalin, consists of a piece of tissue measuring 0.8*0.8*0.5 cm T.S in 2 blocks

[Microscopic Description]
Sections show dome shaped or mildly papillomatous lesion ,with mucinous matrix and some fibroblasts or spindle cells and scattered capillaries few thin epithelial strands are also present .

[Diagnosis]
Skin biopsy , nipple area :
-Cutaneous myxoma is compatible

Attending Dermatopathologist
Dr.J.TAGHIZADEH.MD.

Fellowship of DermatoPathology

Resident of Pathology

Genetic evaluation:

Code No.: 1542030	Lab No: S.3-carney1-98																			
Name: [REDACTED]	Birth: 27/10/22																			
Gender: [REDACTED]	Sample type: EDTA blood																			
Origin: Bokan	Contact address (Tel): 09141845302																			
Referring Physician: Dr. Kiyavar	Date of sampling: 98/05/13																			
Indication: Molecular testing of <i>PRKARIA</i> gene	Reporting date: 98/06/16																			
Clinical Features: Clinically diagnosed with carney complex disorder.																				
Referral Reason: This individual was referred for genetic diagnosis and molecular analysis of <i>PRKARIA</i> gene due to carney complex disorder.																				
Result	<table border="1"> <thead> <tr> <th rowspan="2">Gene</th> <th rowspan="2">Nucleic acid alteration</th> <th rowspan="2">Variant Location</th> <th rowspan="2">Amino Acid Alteration</th> <th colspan="3">Genotype</th> <th rowspan="2">Clinical Significance</th> </tr> <tr> <th>Index</th> <th>Mother</th> <th>Father</th> </tr> </thead> <tbody> <tr> <td><i>PRKARIA</i></td> <td>c.531_534delTGAT</td> <td>Exon 6</td> <td>p.Asp178Lysfs*27</td> <td>Het</td> <td>NA</td> <td>NA</td> <td>Pathogenic</td> </tr> </tbody> </table>	Gene	Nucleic acid alteration	Variant Location	Amino Acid Alteration	Genotype			Clinical Significance	Index	Mother	Father	<i>PRKARIA</i>	c.531_534delTGAT	Exon 6	p.Asp178Lysfs*27	Het	NA	NA	Pathogenic
Gene	Nucleic acid alteration					Variant Location	Amino Acid Alteration	Genotype			Clinical Significance									
		Index	Mother	Father																
<i>PRKARIA</i>	c.531_534delTGAT	Exon 6	p.Asp178Lysfs*27	Het	NA	NA	Pathogenic													
<small>Het.: heterozygote, Hom.: homozygote, Hem.: hemizygote, NA.: not available, VUS: variant with uncertain significance, [=]: Normal allele.</small>																				
Summary	Direct sequencing of the coding regions of <i>PRKARIA</i> gene showed a heterozygous pathogenic variant at position c.531_534delTGAT, presented at protein level as p.Asp178Lysfs*27.																			
Interpretation and Comments	<ul style="list-style-type: none"> c.531_534delTGAT variant in <i>PRKARIA</i> gene has been reported previously in patients with myxoma, intracardiac [1]. 																			

PI

1394

- CVA
- Cardiac myxoma
- Open heart surgery

1396

- Cardiac myxoma recurrence
- Open heart surgery

1397

- Palpitation
- EPS

1398

- Cardiac myxoma recurrence
- Skin myxoma
- Open heart surgery
- Genetic evaluation
- Referred for endocrine evaluation

PI

- ❖ In 1398, visit by an endocrinologist:
 - ❖ Headache , diplopia , seizure , depression (neg)
 - ❖ Snoring (neg)
 - ❖ Hirsutism (neg)
 - ❖ Acne (pos)
 - ❖ Easy bruising, skin thinning, hyperhidrosis, oily skin (neg)
 - ❖ Muscle weakness (neg)
 - ❖ Increased hand & foot size (neg)
 - ❖ Galactorrhea, libido change, irregular menstruation (neg)
 - ❖ Fracture, VTE (neg)

Pars hospital labs: (1398/09)

Wbc:7300	Ca:9.4	Urine sample:		LH:4.7 IU/L
HB: 11.8	Ph:4.4	Volume:600 cc/24 hr	Volume:700 cc/24 hr	FSH:3.5 IU/L
PLT:297000	Na:144	Creatinin:504 mg/24 hr	Creatinin :595 mg/24 hr	Prolactin:15.4 ng/ml
FBS:86	K:4.9	Cortisol:93 micgr/24 hr	Cortisol:25 micgr/24 hr	
BUN:11	AST:12	Serum cortisol:10.5 micgr/dl (Am)		
Cr:0.7	ALT:13	Serum cortisol:10.4 micgr/dl (Pm)		
Uric acid: 5.4	ALK-ph:41	ACTH: <5 pg/ml		
Cholestrol:137	TG:98	DHEAS:18.9 micgr/dl		
LDL:66	HDL:40			

Pars hospital lab(98/10)

Urine sample

Volume: 650 cc/24 hr

Creatinin: 788 mg/24hr

Cortisol: 76 micgr/24 hr

BMD(1398)

سنجش تراکم استخوان Bone Densitometry

Address: 2nd Floor, No.147, Ghaem Magham Ave. Across From
Tehran Clinic Hospital, Tehran, Iran
TEL: 021- 88750337 – 88750338

BONE DENSITOMETRY REPORT

Name: [Redacted] Date: 1398/09/17
Age: 24

Referring Physician: Dr. HADAEGH
The determination of BMD (Bone Mineral Density) was done by **DEXA** method by **Hologic Discovery Wi** (Dual energy X-ray Absorptiometry) for above patient on **L1 to L4** (lumbar vertebrae) and **Neck** of Femur.

Region	BMD (g/cm ²)	T score	Z score	AM (%)	ISCD Classification
Lumbar Spine (L1-L4)	0.885	-1.5	-1.4	85	NORMAL BMD
Femoral Neck (L)	0.804	-0.4	-0.4	95	NORMAL BMD

DIAGNOSIS BY ISCD DEFINITION:
NORMAL BMD

Telephone: 88750337 - 88750338

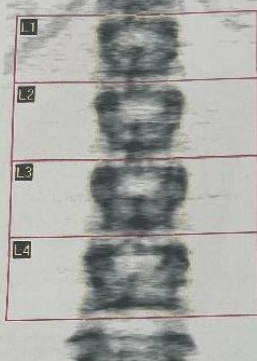
Name: SOLTANI, SOMAYEH
Patient ID: 98917-15
DOB: 12 January 1995

Sex: Female
Ethnicity: Caucasian

Height: 171.0 cm
Weight: 86.0 kg
Age: 24

Referring Physician: DR.HADAEGH

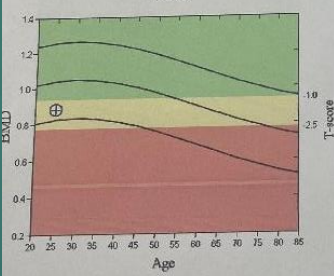
Scan Information:
Scan Date: 08 December 2019 ID: A1208190W
Scan Type: f Lumbar Spine
Analysis: 08 December 2019 15:50 Version 13.3.0.1 Spine
Operator:
Model: Discovery Wi (S/N 85352)
Comment:



DXA Results Summary:

Region	Area (cm ²)	BMC (g)	BMD (g/cm ²)	T-score	PR (%)	Z-score	AM (%)
L1	13.80	11.95	0.866	-1.1	87	-1.1	88
L2	15.52	13.63	0.878	-1.4	85	-1.3	86
L3	16.80	15.29	0.910	-1.6	84	-1.5	85
L4	18.86	16.62	0.881	-1.6	83	-1.6	84
Total	64.98	57.50	0.885	-1.5	85	-1.4	85

Total BMD CV: 1.0%
WHO Classification: Osteopenia
Fracture Risk: Increased



116 x 149
DAP: 2.1 cGy*cm²

Comment:

PARSIAN BONE DENSITOMETRY Center
ACROSS FROM TEHRAN CLINIC HOSPITAL NO.147
TEHRAN, IRAN


Telephone: 88750337 - 88750338

Name: SOLTANI, SOMAYEH
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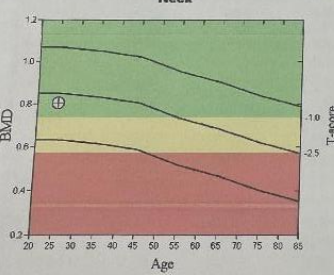


Scan Information:
Scan Date: 08 December 2019 ID: A1208190X
Scan Type: f Left Hip
Analysis: 08 December 2019 15:49 Version 13.3.0.1 Hip
Operator:
Model: Discovery Wi (S/N 85352)
Comment:

DXA Results Summary:

Region	Area (cm ²)	BMC (g)	BMD (g/cm ²)	T-score	PR (%)	Z-score	AM (%)
Neck	4.83	3.89	0.804	-0.4	95	-0.4	95
Troch	8.63	5.97	0.692	-0.1	98	-0.1	98
Inter	23.71	25.90	1.093	0.0	99	0.0	101
Total	37.18	35.76	0.962	0.2	102	0.2	102
Ward's	1.11	0.74	0.663	-0.6	90	-0.6	90

Total BMD CV: 1.0%
WHO Classification: Normal

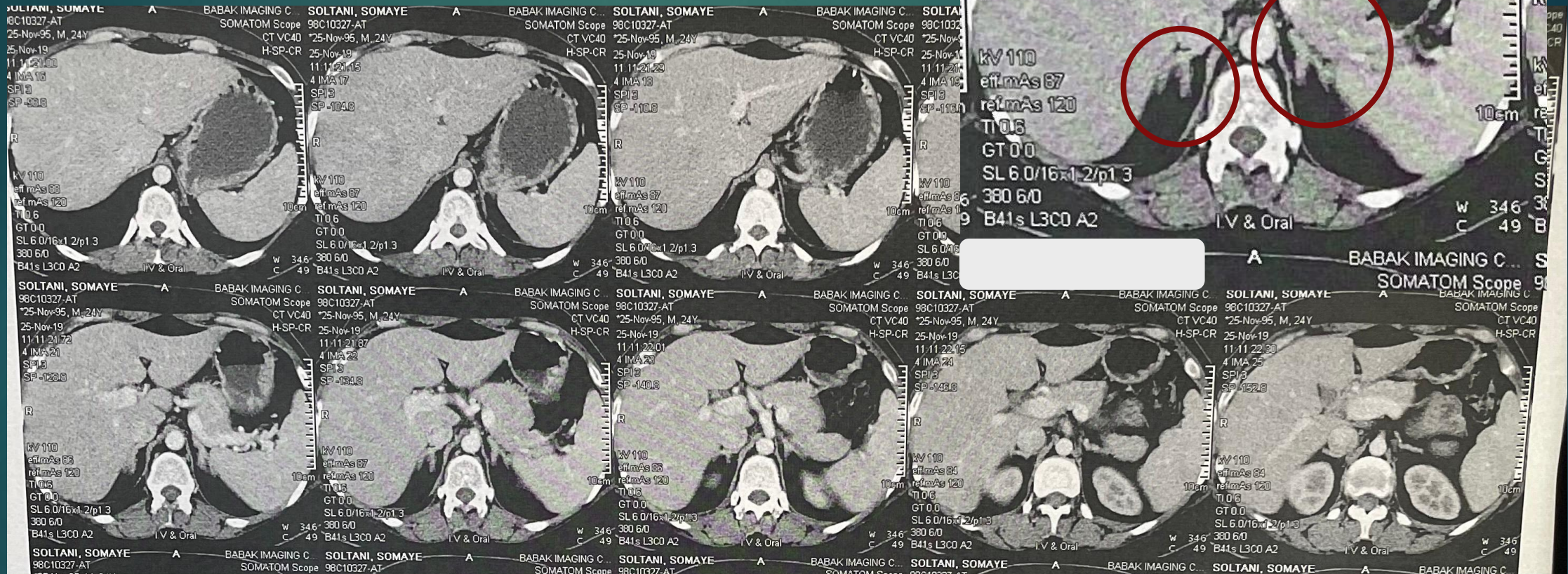


108 x 133
NECK: 49 x 15
DAP: 1.7 cGy*cm²

10-year Fracture Risk!
Major Osteoporotic Fracture: 1.7%
Hip Fracture: < 0.1%
Reported Risk Factors:
US (Caucasian), T-score(WHO)=-0.4, BMD=29.4
Input outside FRAX® limits. Adjusted to Age=40

Comment:

Abdominal CT



Pars hospital lab (99/3)



WBC:8800	AST :15	Urine sample	ODST:7.15 micgr/dl
HB:10.9	Alt :17	Volume:500 cc	ACTH:25 pg/ml
PLT:350000	Alk ph:55	Creatinin:720 mg/24 hr	Cortisol:7.18 micgr/dl(AM)
FBS:104	Chol:144	Cortisol: 64.7 micgr/24hr	Cortisol:6.52 micgr/dl (PM)
Na:142	HDL:44		
K:4.3	LDL:70		
	TG:100		

Pars hospital lab (1400/1)

Wbc:7800	Cortisol:15.5 micgr/dl (AM)	Urine sample
Hb :12.2	Cortisol:13.8 micgr/dl (PM)	Volume: 800cc
Plt:267000	ACTH: <5 pg/ml	Creatinin: 552 mg/24 hr
Fbs:100	DHEAS:33 micgr/dl	Cortisol: 254 micgr/24 hr
Cr:0.6		
Na :140		
K:4.7		

BMD(1400)

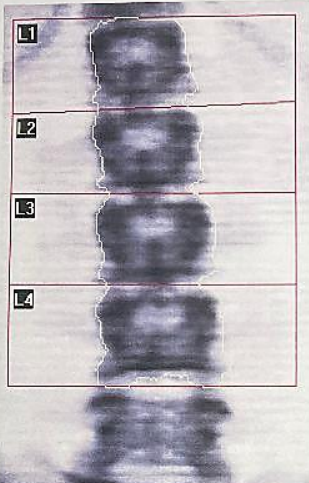
SHAHID BEHESHTI MEDICAL UNIVERSITY
TEHRAN

Telephone: 22432556-23031250 E-Mail: taleghanihospital@sbm.ac.ir Fax: 22432596

Sex: Female Height: 173.0 cm
Ethnicity: White Weight: 86.0 kg
Age: 27

Referring Physician:

Scan Information:
Scan Date: 26 May 2021 ID: A05262108
Scan Type: fe Lumbar Spine
Analysis: 26 May 2021 11:52 Version 13.3
Lumbar Spine
Operator: n-b
Model: Explorer (S/N 91819)
Comment:



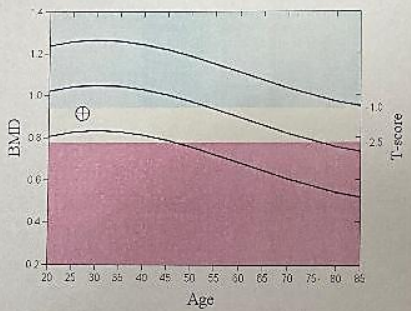
116 x 151
DAP: 4.2 cGy*cm²

DXA Results Summary:

Region	Area (cm ²)	BMC (g)	BMD (g/cm ³)	T-score	PR (%)	Z-score	AM (%)
L1	14.27	11.35	0.795	-1.2	86	-1.1	86
L2	14.35	13.16	0.917	-1.0	89	-1.0	90
L3	16.76	16.41	0.979	-1.0	90	-0.9	91
L4	19.58	18.48	0.944	-1.6	85	-1.5	85
Total	64.96	59.39	0.914	-1.2	87	-1.2	88

Total BMD CV 1.0%
WHO Classification: Osteopenia
Fracture Risk: Increased

Total



Comment:

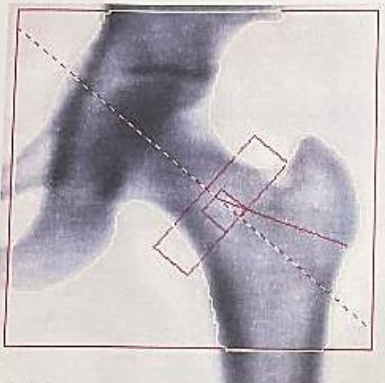
TALEGHANI HOSPITAL-NUCLEAR MEDICINE DEPARTMENT
SHAHID BEHESHTI MEDICAL UNIVERSITY
TEHRAN

Telephone: 22432556-23031250 E-Mail: taleghanihospital@sbm.ac.ir Fax: 22432596

Sex: Female Height: 173.0 cm
Ethnicity: White Weight: 86.0 kg
Age: 27

Referring Physician:

Scan Information:
Scan Date: 26 May 2021 ID: A05262107
Scan Type: fe Left Hip
Analysis: 26 May 2021 11:54 Version 13.3
Left Hip
Operator: n-b
Model: Explorer (S/N 91819)
Comment:



118 x 112
NECK: 49 x 15
DAP: 2.7 cGy*cm²

DXA Results Summary:

Region	Area (cm ²)	BMC (g)	BMD (g/cm ³)	T-score	PR (%)	Z-score	AM (%)
Neck	4.98	3.92	0.787	-0.6	93	-0.5	93
Troch	8.74	6.11	0.699	0.0	99	0.0	99
Inter	14.39	14.30	0.994	-0.7	90	-0.6	91
Total	28.11	24.33	0.866	-0.6	92	-0.6	92
Ward's	1.10	0.70	0.636	-0.8	87	-0.8	88

Total BMD CV 1.0%
WHO Classification: Normal

FRAX® WHO Fracture Risk Assessment Tool

10-year Fracture Risk!

Major Osteoporotic Fracture 1.3%
Hip Fracture <0.1%

Reported Risk Factors:
Turkey T-score(WHO)=-0.6, BMI=28.7
Input outside FRAX® limits. Adjusted to Age=40

FRAX® Version 3.01. Fracture probability calculated for an untreated patient. Fracture probability may be lower if the patient has received treatment.

Comment:

ne vs. White Female; Z-score vs. White Female. Source:BMDCS/NHANES

FH

- ❖ Father: HCV positive, HCC
- ❖ Brother: Bone histiocytosis - surgery

Physical exam

- ❖ Weight: 86 kg, height:167 cm, BMI: 33 kg/m²
- ❖ BP:115/70 mmHg
- ❖ Positive findings:
 - ❖ Moon face
 - ❖ Lentigo
 - ❖ Acne

Physical exam

- ❖ Breast: skin myxoma
- ❖ Open heart surgery scar on the chest wall
- ❖ Abdominal obesity is present.
- ❖ Muscle force: 5/5

Problem list

- ❖ A 27-year-old woman
- ❖ Known case of Carney complex
- ❖ Cardiac myxomas
- ❖ Cutaneous myxomas
- ❖ PPNLD
- ❖ **Desire for pregnancy**