

Overdiagnosis of osteoporosis in a patient with short stature and partial growth hormone insensitivity due to misinterpretation of Dual-energy x-ray absorptiometry (DEXA)

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Bone densitometry is currently one of the mainstays in the evaluation of systemic bone diseases. The most frequently assessed densitometric parameter is areal bone mineral density (BMD), measured by dual energy X-ray absorptiometry (DEXA) and expressed as g/cm². However it takes into account only the bone surface and not the depth of the bone which is being measured.

As a result the findings may represent an underestimation in patients with small bones such as in adults with short stature due to growth hormone insensitivity syndrome (GHIS) caused by mutations in the GH receptor gene or its downstream mediators. Volumetric bone density (BMAD) expressed as g/cm³, has been considered to be a more accurate estimate of bone density in these patients.

CASE REPORT

A 47-yr-old woman was admitted to our clinic for investigation of premenopausal osteoporosis. She had performed a DEXA due to bone pains, which revealed severe osteoporosis (lumbar spine T-score:-2,9, femoral neck T-score:-3).

MEDICAL HISTORY

- Normal birth size
- No history of growth retardation
- No metabolic diseases

PHYSICAL EXAMINATION

- short stature (1,47 m)
- normal BMI
- thin lips
- small chin



LABORATORY TESTS

WBC	7,8 K/μl	Glucose	87 mg/dl
RBC	4,3 M/μl	Urea	0.7 mg/dl
HBC	12,8 g/dl	Cr	27 mg/dl
HCT	38,2 %	Chol.	191 mg/dl
PTL	320 K/μl	SGOT	24 IU/ml
		SGPT	21 IU/ml
		ALP	61 IU/ml

HORMONE MEASUREMENTS

TSH	1,017 μU/ml	PTH	48,3 pg/ml	<ul style="list-style-type: none"> ➤ Vitamin D insufficiency Normal calcium metabolism ➤ Gonadotropin values indicate perimenopause 	DATE	11/2014	12/2014	01/2016		<ul style="list-style-type: none"> ➤ Findings support a diagnosis of partial GH insensitivity syndrome ➤ IGF-1 generation test is not diagnostic for mild defects ➤ Genetic test is pending
FT4	0,87 ng/dl	Ca	9,5 mg/dl		GH	3,54	16,7	6,09		
FSH	17,04 mIU/ml	Alb.	4,7 g/dl		IGF-1 (101-267 ng/ml)	118	103	104		
LH	25,15 mIU/ml	P	4,2 mg/dl		IGF-BP3 (3,4-6,7 μg/ml)			3,2		
E2	113 pg/ml	Mg	2,2 mg/dl							
ACTH	10,7 pg/ml	25-OH BIT.D	26 ng/ml							
CORT	21,7 μg/dl									

RADIOLOGICAL FINDINGS

X-ray	DEXA	BMAD
X-ray of lumbar and thoracic spine revealed no fracture	DEXA revealed severe osteoporosis (lumbar spine T-score:-2,9, femoral neck T-score:-3)	Estimated BMAD at the spine and femoral neck indicated osteopenia
		<p>Lunar</p> <p>L2-4 BMC, grams: 29.97 L2-4 area, cm²: 35.80</p> <p>Fem. neck BMC, grams: 2.95 Fem. neck area, cm²: 4.73</p> <p>Calculate</p> <p>L2-4 BMAD = $\frac{BMC}{area^{1.5}} = 0.122$ T-score = -1.94</p> <p>Femoral neck BMAD = $\frac{BMC}{area^2} = 0.106$ T-score = -2.32</p>

DIAGNOSIS

The patient was diagnosed with short stature due to partial growth hormone insensitivity syndrome and osteopenia. She was treated with vitamin D and calcium supplements.

CONCLUSION

The described case highlights the importance of a high degree of suspicion for diagnosis of partial GHIS in patients with short stature and osteoporosis. Appropriate bone mass estimation in these patients is crucial for therapeutic decisions because the low BMD found in them, using conventional DEXA densitometry, may be an artifact of the reduced bone size.

REFERENCES

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