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The role of dual-energy X-ray absorptiometry in the detection of secondary skeletal involvement in a female patient with Gardner syndrome and HIV infection

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Abstract: gardner syndrome is a rare genetic disorder, considered a phenotypic variant of familial adenomatous polyposis, which is associated with an aberration in the adenomatous polyposis coli (APC) gene. It is characterized by a combination of intestinal (colonic polyps) and a wide spectrum of extracolonic manifestations, including osteomas, desmoid tumors, epidermal cysts, hypertrophy of the retinal pigment epithelium, follicular thyroid cancer, and others. This article describes a clinical case of atypical bone changes in a female patient with Gardner syndrome, HIV infection, chronic post-hemorrhagic anemia, and hypothyroidism in a state of medical compensation. Gardner syndrome was confirmed at the age of three by endoscopic and morphological examination of multiple adenomatous polyps in the esophagus, stomach, duodenum, and colon. HIV infection was diagnosed at the age of 13.5 years, for which the girl receives antiretroviral therapy (ART). At the age of 17, she was referred for dual-energy X-ray absorptiometry (DXA), which revealed areas of localized increased bone mineral density in the projection of the upper third of the skull bilaterally. Subsequent skull radiography in the anteroposterior projection identified these changes as pronounced bilateral hyperostosis, while magnetic resonance imaging (MRI) of the brain further visualized diffuse thickening of the skull bones, emphasized at the vault level, with no pathological changes in the brain parenchyma. Thus, the detected hyperostosis had no clear clinical manifestations but became an incidental diagnostic finding during a routine dual-energy X-ray absorptiometry scan. The described case also illustrates the need for a multidisciplinary approach to managing patients with comorbid pathologies, as only the collaboration of doctors from different specialties allows for the correct interpretation of the results and determination of the optimal tactics for further examination and treatment. The detected hyperostosis in an adolescent with HIV infection and Gardner syndrome may likely be attributed to severe chronic anemia, which may have both diagnostic and prognostic significance. This case highlights the potential utility of X-ray absorptiometry that can serve not only as a method

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for quantitative assessment of bone mineral density but also as an additional tool for detecting skeletal structural changes that require further verification using other diagnostic imaging methods.

Key words: Anemia, Dual-Energy X-Ray Absorptiometry, Gardner Syndrome, Hyperostosis, Diagnosis.

Introduction

Gardner syndrome (GS) is an autosomal dominant phenotypic variant of familial adenomatous polyposis (FAP), characterized by the presence of adenomatous polyps in the colon, which have a high risk of malignant transformation. Clinically, it often manifests as diarrhea, spasms, pain and/or rectal bleeding, constipation, and vomiting [1].

GS also involves extracolonic manifestations, including desmoid tumors, osteomas, epidermoid cysts, hyperdontia, odontomas, dental cysts, and bilateral pigmented congenital hypertrophy of the retinal pigment epithelium [2].

Dual-energy X-ray absorptiometry (DXA) is traditionally used for the quantitative assessment of bone mineral density (BMD) [3]. However, in rare cases, it can also aid in suspecting morphological skeletal changes. The impact of HIV infection and long-term antiretroviral therapy (ART) on bone health is an additional, well-documented risk factor for reduced BMD [6, 7].

Aim

To present the role of dual-energy X-ray absorptiometry (DXA) in the primary detection of structural skull bone changes in a female patient with Gardner syndrome and comorbid pathologies.

Materials and Methods

BMD assessment was performed using dual-energy X-ray absorptiometry on an OsteoSys PRIMUS densitometer, in accordance with the recommendations of the International Society for Clinical Densitometry (ISCD). The scanned areas included: total body, lumbar spine (L1-L4), and the proximal femur and neck of the left femur [3].

BMD was evaluated considering sex, age, height, ethnicity, and Tanner stage of sexual development. To confirm the detected structural changes in the skull bones, the patient underwent skull radiography in the anteroposterior projection, as well as magnetic resonance imaging (MRI) of the brain to exclude secondary intracranial pathology.

Case Description

A 17-year-old female patient presented with intestinal bleeding since the age of 18 months, for which she was regularly examined at her local hospital, where colonic polyps causing the bleeding were detected. The recurrent bleeding led to chronic post-hemorrhagic anemia, requiring regular blood transfusions for correction.

The first surgical intervention, removal of rectal polyps, was performed at the age of two. Due to worsening condition, she was referred to the National Children's Specialized Hospital "Ohmatdyt" at the age of three, where Gardner syndrome was suspected and verified based on colonoscopy and pathohistological examination of the detected neoplasms. Further examination revealed multiple polyps in the esophagus, stomach, duodenum, and colon with a high risk of malignant transformation and progression of anemia. At the time of diagnosis, the parents refused the recommended scope of further surgical treatment.

During a re-hospitalization at "Ohmatdyt" in 2014, with parental consent, a colectomy and loop ileostomy were performed, with stoma closure one year later. After the last surgical intervention, the volume of bleeding decreased to occasional single streaks of blood in the stool. Due to chronic post-hemorrhagic anemia, she received multiple blood transfusions according to clinical guidelines, with constant monitoring of necessary hematological parameters.

In 2020, due to frequent episodes of pneumonia, HIV infection was suspected and verified. Given the negative test results of the parents, the probable cause of infection was blood transfusions. The patient receives standard antiretroviral therapy (ART). During her last hospitalization, other comorbidities were identified: multinodular goiter and medically controlled hypothyroidism at the time of examination. The family history was unremarkable; there was no family history of Gardner syndrome.

The patient was referred for a planned DXA scan to assess BMD, as she had been receiving ART for four years and had several comorbid pathologies known to be risk factors for osteoporosis [6].

The DXA results showed that BMD indicators for the total skeleton, lumbar spine, proximal femur, and femoral neck were within the normal range for her age (Figure 1, A, B, C).

When analyzing the total body DXA scan, a localized bilateral increase in bone density was localized in the upper third of the skull, Z-score > +5.0 SD (Figure 2).

Given the patient's diagnosis of Gardner syndrome, skull osteomas were suspected [8]. To verify this, the patient underwent skull radiography

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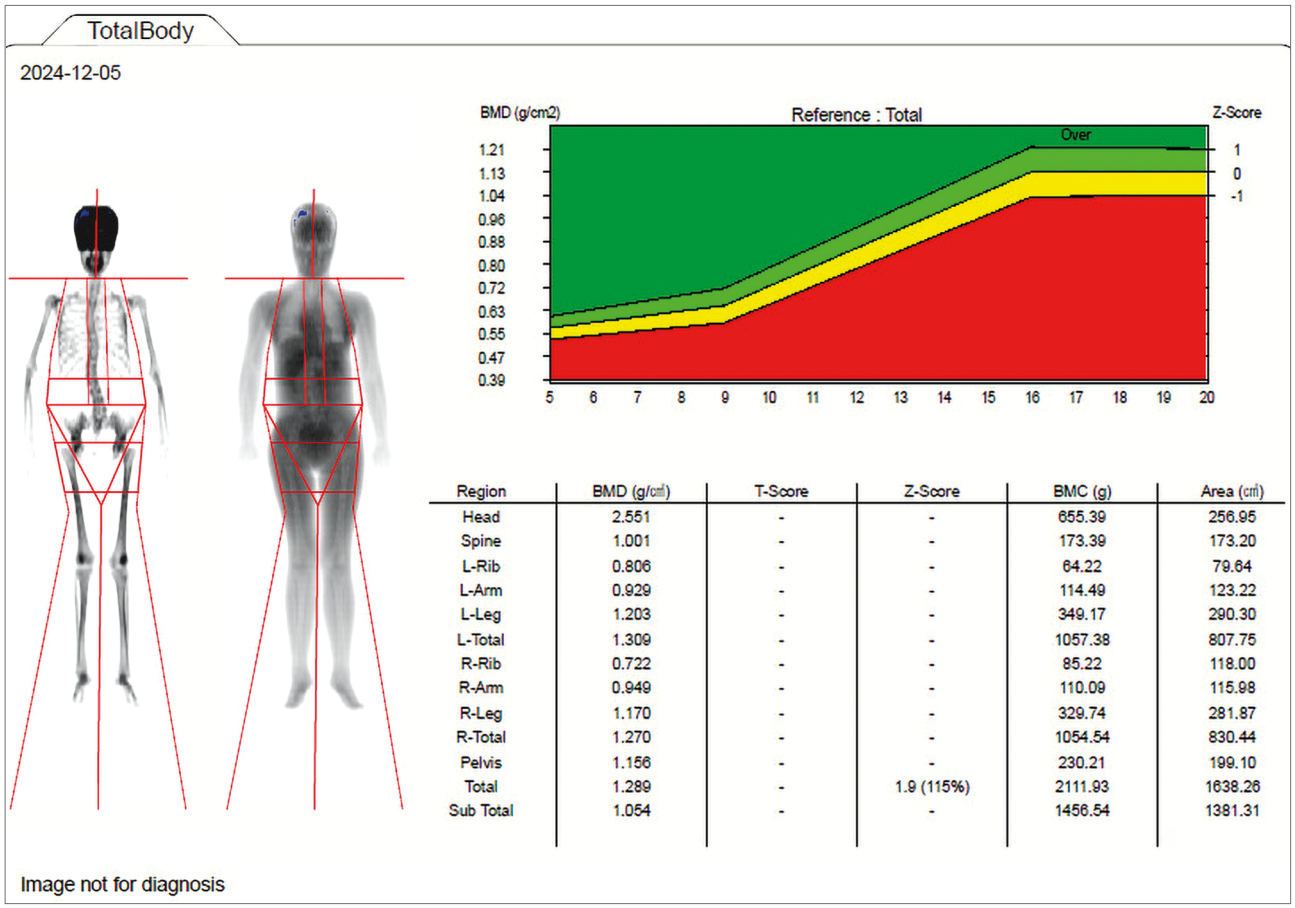
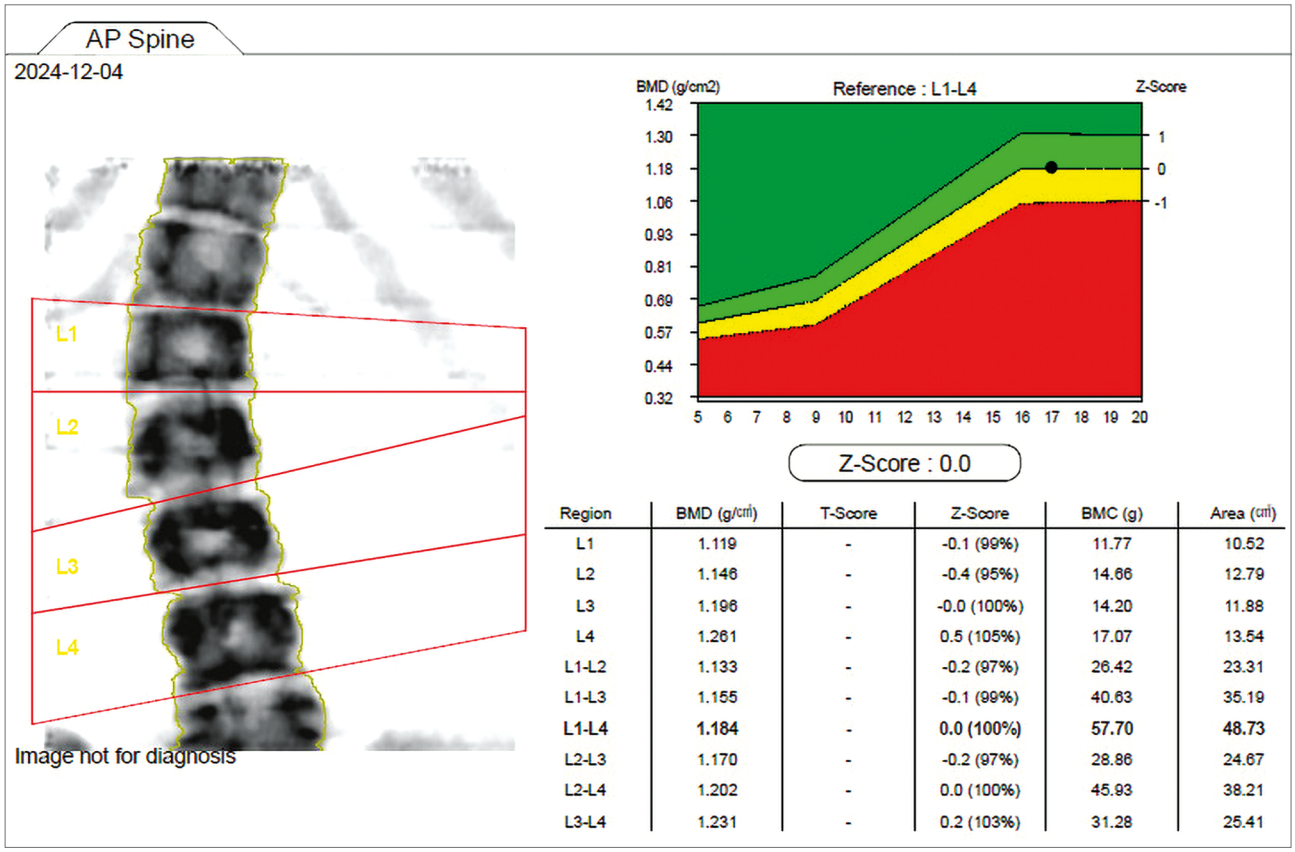


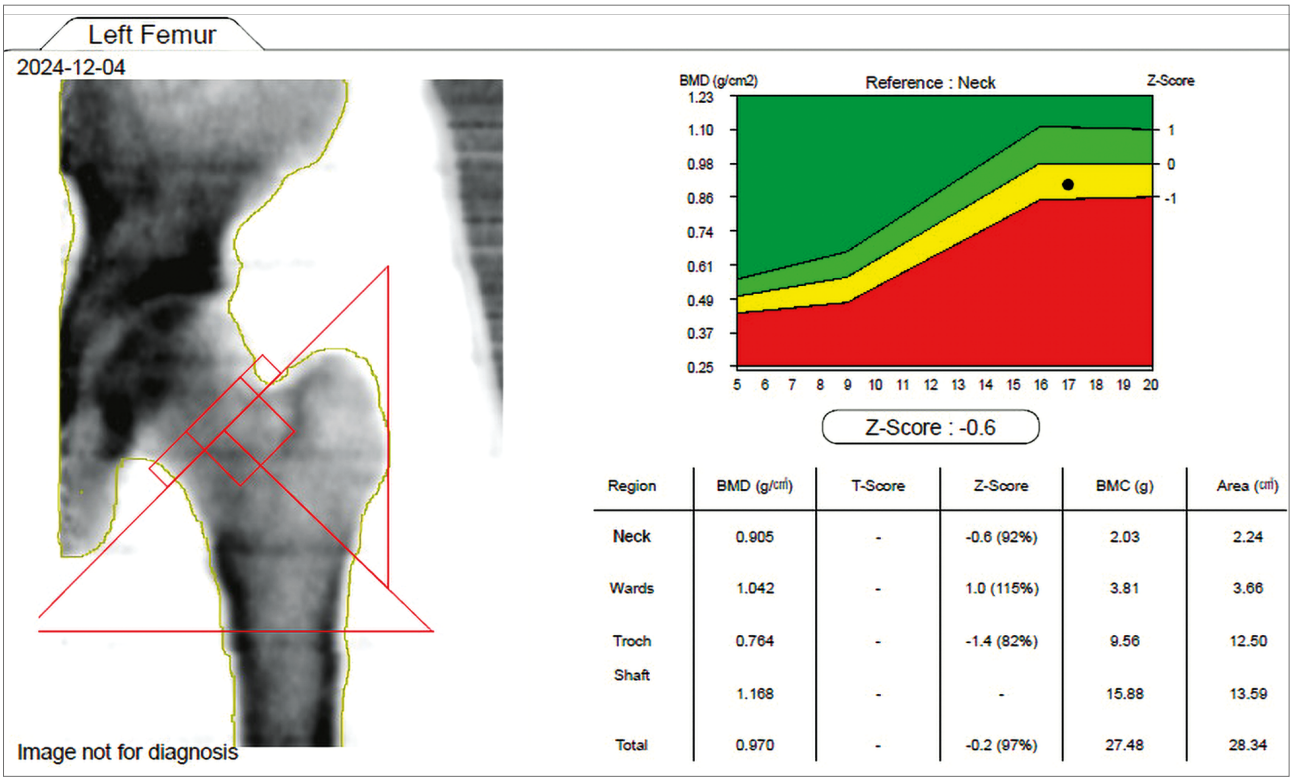
Region	BMD (g/cm²)	T-Score	Z-Score	BMC (g)	Area (cm²)
Head	2.551	-	-	655.39	256.95
Spine	1.001	-	-	173.39	173.20
L-Rib	0.806	-	-	64.22	79.64
L-Arm	0.929	-	-	114.49	123.22
L-Leg	1.203	-	-	349.17	290.30
L-Total	1.309	-	-	1057.38	807.75
R-Rib	0.722	-	-	85.22	118.00
R-Arm	0.949	-	-	110.09	115.98
R-Leg	1.170	-	-	329.74	281.87
R-Total	1.270	-	-	1054.54	830.44
Pelvis	1.156	-	-	230.21	199.10
Total	1.289	-	1.9 (115%)	2111.93	1638.26
Sub Total	1.054	-	-	1456.54	1381.31

A



Region	BMD (g/cm²)	T-Score	Z-Score	BMC (g)	Area (cm²)
L1	1.119	-	-0.1 (99%)	11.77	10.52
L2	1.146	-	-0.4 (95%)	14.66	12.79
L3	1.196	-	-0.0 (100%)	14.20	11.88
L4	1.261	-	0.5 (105%)	17.07	13.54
L1-L2	1.133	-	-0.2 (97%)	26.42	23.31
L1-L3	1.155	-	-0.1 (99%)	40.63	35.19
L1-L4	**1.184**	-	**0.0 (100%)**	**57.70**	**48.73**
L2-L3	1.170	-	-0.2 (97%)	28.86	24.67
L2-L4	1.202	-	0.0 (100%)	45.93	38.21
L3-L4	1.231	-	0.2 (103%)	31.28	25.41

B



C

Fig. 1. DXA bone mineral density scan regions: A – total body, B – lumbar spine, C – proximal left femur

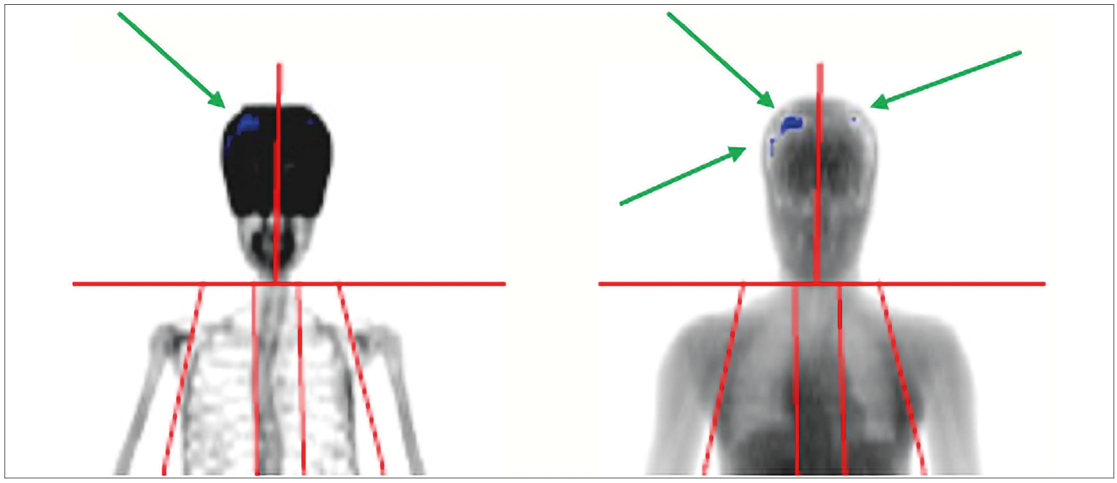


Fig. 2 Regions of increased BMD of the skull bones, detected by DXA

in the anteroposterior projection, which revealed hyperostosis in the frontoparietal regions of the skull, more pronounced on the right (Figure 3).

To exclude secondary intracranial pathology, an MRI of the brain was performed during the next planned hospitalization 3 months later (Figure 4). The MRI confirmed diffuse thickening of the cerebral skull bones, more pronounced in the vault and occipital bone, without involvement of the brain structures in the pathological process.

At the time of writing this case report, the patient's condition is stable. She continues to take ART on an outpatient basis and is under the dynamic observation of local physicians. No additional treatment or examinations were prescribed.

Discussion

The literature describes cases where extracolonic manifestations of Gardner syndrome, such as osteomas, as well as dental and jaw anomalies (retained teeth, supernumerary teeth, odontomas), were the first manifestation preceding the appearance of intestinal polyps [8, 9].

From a genetic perspective, mutations in the APC gene within Gardner syndrome cause impaired

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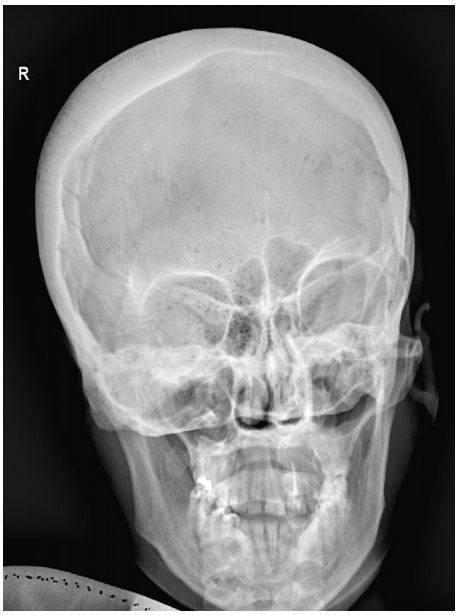

Fig. 3 X-ray of the skull, frontal view

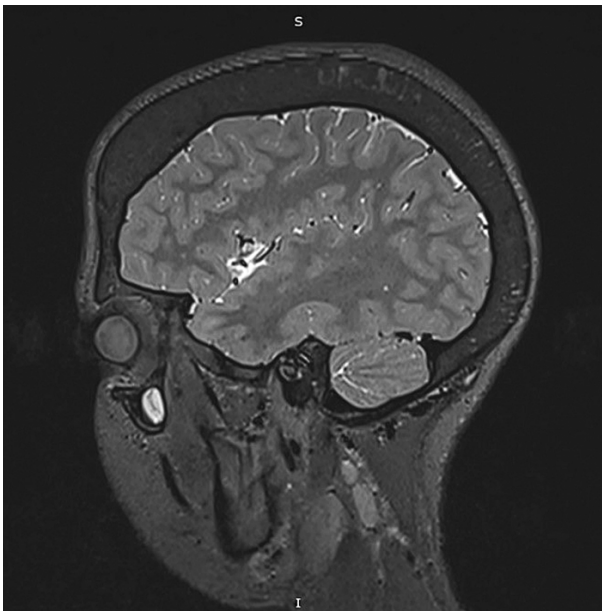

Fig. 4. MRI of the brain, sagittal view

control of cellular proliferation, manifesting as intestinal and extracolonic features, including the aforementioned bone changes [1, 10].

However, in the context of this case, the detected diffuse skull thickening without the classic bone manifestations of Gardner syndrome (such as dense, circumscribed osteomas) indicates that the structural changes, considering the patient's medical history, this process was initiated by severe chronic post-hemorrhagic anemia from an early age [11].

The literature describes that severe, prolonged anemia leads to bone marrow hyperplasia, stimulating the expansion of the diploic space of the skull bones, often resulting in calvarial thickening [12, 13]. This finding is crucial, as chronic anemia itself can cause significant calvarial changes that mimic other pathologies [14].

Conclusions

Dual-energy X-ray absorptiometry, which is used in clinical practice for the quantitative assessment of bone mineral density, provided important incidental findings in this case, leading to the detection of localized morphological changes in the form of hyperostosis of the skull bones.

This report illustrates that densitometry scans may occasionally reveal morphological skeletal changes and emphasizes the importance of a comprehensive, multidisciplinary approach to analyzing the obtained results, taking into account anamnestic, clinical, imaging, and laboratory data.

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Patient consent. Written informed consent was obtained from the patient and her legal representatives for the publication of this case report and accompanying images."

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Роль двоенергетичної рентгенівської денситометрії у діагностиці вторинного ураження скелета у пацієнтки з синдромом Гарднера та ВІЛ-інфекцією

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Анотація: синдром Гарднера є рідкісним генетичним захворюванням, що відноситься до фенотипових варіантів сімейного аденоматозного поліпозу, що пов'язують з аберацією гена аденоматозного поліпозу товстої кишки (APC). Він характеризується комбінацією кишкових (поліпи товстої кишки) та широкого спектру позакишкових проявів, серед яких: остеоми, десмоїдні пухлини, епідермальні кісти, гіпертрофія пігментного епітелію сітківки, фолікулярний рак щитоподібної залози тощо. У статті описано клінічний випадок нетипових кісткових змін у пацієнтки з синдромом Гарднера, ВІЛ-інфекцією, хронічною постгеморагічною анемією та гіпотиреозом у стадії медикаментозної компенсації. Синдром Гарднера підтверджено за результатами ендоскопічного та морфологічного дослідження множинних аденоматозних поліпів стравоходу, шлунку, дванадцятипалої та товстої кишок у віці трьох років. ВІЛ-інфекцію виявили у віці 13,5 років, у зв'язку з чим дівчинка отримує антиретровірусну терапію (АРТ). У 17 років її було скеровано на проведення двофотонної рентгенівської денситометрії, при оцінці результатів якої виявлено ділянки локалізованого підвищення мінеральної щільності кісткової тканини у проекції верхньої третини черепа обабіч. Подальша рентгенографія черепа в прямій проекції підтвердила знайдені зміни у вигляді вираженого двобічного гіперостозу, а магнітно-резонансна томографія (МРТ) головного мозку додатково візуалізувала дифузне потовщення кісток черепа з акцентом на рівні склепіння без патологічних змін з боку паренхіми мозку. Таким чином, виявлений гіперостоз не мав чітких клінічних проявів, проте став випадковою діагностичною знахідкою під час планового проведення двофотонної рентгенівської денситометрії. Описаний випадок також ілюструє необхідність мультидисциплінарного підходу до ведення пацієнтів із коморбідними патологіями, оскільки лише взаємодія лікарів різних спеціальностей дозволяє коректно інтерпретувати отримані результати та визначити оптимальну тактику подальшого обстеження та лікування. Виявлений гіперостоз у підлітка з ВІЛ-інфекцією та синдромом Гарднера слід розглядати як наслідки тяжкої хронічної анемії, що може мати як діагностичне, так і прогностичне значення. Представлений випадок демонструє, що рентгенівська денситометрія може виступати не лише методом кількісної оцінки мінеральної щільності кісткової тканини, а й додатковим інструментом виявлення структурних змін скелета, які потребують подальшої верифікації за допомогою візуалізаційних методів діагностики.

Ключові слова: гіперостоз, двоенергетична рентгенівська денситометрія, синдром гарднера, анемія, діагноз.