Adult E-Poster

EP_A157

OSTEOPOROSIS IN ACROMEGALY: A PARADOXICAL COMPLICATION WITH MULTIFACTORIAL MECHANISMS

https://doi.org/10.15605/jafes.040.S1.165

Sarah Firdausa,[1,2] Luki Kusumaningtyas,[1,3] Imam Subekti,[1] Tri Juli Edi Tarigan,[1] Dicky L Tahapary[1]
[1]*Division of Endocrinology, Metabolism, and Diabetes, Department of Internal Medicine, Cipto Mangunkusumo Hospital, Faculty of Medicine, University of Indonesia*
[2]*Division of Endocrinology, Metabolism, and Diabetes, Department of Internal Medicine, Faculty of Medicine, Universitas Syiah Kuala, Banda Aceh, Indonesia*
[3]*Division of Endocrinology, Metabolism, and Diabetes, Department of Internal Medicine, Gatot Subroto Central Army Hospital, Jakarta, Indonesia*

INTRODUCTION/BACKGROUND

Acromegaly results from prolonged exposure to elevated levels of growth hormone (GH) and insulin-like growth factor-1 (IGF-1), which contribute to increased bone turnover. Despite IGF-1's known anabolic effects on bone, patients with acromegaly paradoxically face a higher risk of developing osteoporosis and vertebral fractures. This case series highlights the significance of early evaluation of bone health in managing acromegaly.

CASE

We evaluated four patients with confirmed acromegaly—three females and one male—ranging in age from 27 to 56 years, who underwent bone mineral density (BMD) testing via dual-energy X-ray absorptiometry (DXA). Half of the patients were diagnosed with osteoporosis, one had osteopenia, and one had normal BMD with borderline values.

A 27-year-old male, diagnosed with acromegaly at the age of 13, exhibited severe osteoporosis with lumbar Z-score –4.3, hip Z-score –3.6, and radius Z-score –3.3, and also exhibited panhypopituitarism and skeletal deformities. A 56-year-old postmenopausal female, diagnosed at 41 years, had osteoporosis with spinal T-score –2.7 and radius T-score –2.9. A 34-year-old female, diagnosed at age 29 and with secondary amenorrhea, had osteopenia (radius Z-score –2.2) despite near-normal lumbar and hip values. A 34-year-old male, diagnosed at 33 with hypogonadotropic hypogonadism, had overall normal BMD, though his radius showed a borderline Z-score of –0.5. Longer disease duration and hormonal deficiencies appeared to correlate with lower BMD, especially in trabecular-rich regions.

While GH and IGF-1 stimulate bone formation, chronic excess may disrupt bone remodeling balance, leading to increased resorption, deterioration of trabecular structure, and higher cortical porosity. Local IGF-1 resistance and hypogonadism further impair bone integrity. These changes contribute to bone fragility even when BMD appears normal, suggesting that skeletal damage may precede densitometric findings.

CONCLUSION

Osteoporosis is a frequent but underrecognized complication in acromegaly. Bone fragility may develop early due to increased bone turnover, trabecular deterioration, and hypogonadism—despite normal or elevated BMD.

EP_A158

UNVEILING THE UNEXPECTED: A RARE PARAOVARIAN PARAGANGLIOMA MASQUERADING AS AN ADNEXAL MASS

https://doi.org/10.15605/jafes.040.S1.166

Eng Seng Lim,[1] Hwee Ching Tee,[2] Jin Hui Ho[2]
[1]*Department of Internal Medicine, Hospital Queen Elizabeth II, Kota Kinabalu, Sabah, Malaysia*
[2]*Diabetes and Endocrinology Unit, Department of Internal Medicine, Hospital Queen Elizabeth II, Kota Kinabalu, Sabah, Malaysia*

INTRODUCTION/BACKGROUND

Paragangliomas are rare neuroendocrine tumors that arise from extra-adrenal paraganglionic tissue, typically associated with the autonomic nervous system. While they are most commonly found in the adrenal medulla (as pheochromocytomas) or along the sympathetic and parasympathetic chains, their occurrence in the paraovarian region is extremely rare.

CASE

We present the case of a 42-year-old female with a history of left external iliac and common femoral vein thrombosis, who presented with progressive abdominal distension over the past six months. She was normotensive and exhibited no constitutional symptoms, features of catecholamine excess, or compressive symptoms. Blood investigations revealed an elevated CA125 level while other tumor markers were within the normal range. A Computed Tomography (CT) scan of the thorax, abdomen, and pelvis revealed a large intra-abdominal cystic mass measuring 15.2 × 20.8 × 24.5 cm, likely originating from the left ovary with significant mass effect.

The patient underwent an extrafascial hysterectomy with bilateral salpingo-oophorectomy (EHBSO), left pelvic lymph node dissection (PLND), omentectomy, appendicectomy, and adhesiolysis. Histopathological examination of the left

ovarian tumor and fallopian tube favored a diagnosis of extra-adrenal paraganglioma as the immunohistochemical staining was positive for S-100, synaptophysin, and chromogranin. Retrospectively, the tumor was likely a non-functioning paraganglioma, as the patient underwent surgery without complications.

CONCLUSION

Paraovarian paraganglioma is an exceptionally rare entity that presents significant diagnostic challenges due to its atypical location and non-specific clinical features. This case highlights the importance of considering paraganglioma in the differential diagnosis of adnexal masses as the perioperative management may differ.

EP_A159

46,XY DSD WITH RETAINED MÜLLERIAN STRUCTURES AND GENDER TRANSITION IN ADULTHOOD: A STEPWISE DIAGNOSTIC APPROACH

https://doi.org/10.15605/jafes.040.S1.167

Sarah Firdausa,[1,2] Jerry Nasarudin,[1,3] Nur Rusyda Kuddah,[1] Wismandari Wisnu,[1] Em Yunir[1]

[1]Division of Endocrinology, Metabolism, and Diabetes, Department of Internal Medicine, Cipto Mangunkusumo Hospital, Faculty of Medicine, University of Indonesia

[2]Division of Endocrinology, Metabolism, and Diabetes, Department of Internal Medicine, Faculty of Medicine, Universitas Syiah Kuala, Aceh, Indonesia

[3]Division of Endocrinology, Metabolism, and Diabetes, Department of Internal Medicine, Fatmawati Hospital, Jakarta, Indonesia

INTRODUCTION/BACKGROUND

Disorders of Sex Development (DSD) are congenital conditions marked by atypical chromosomal, gonadal, or anatomical sex development. A structured diagnostic approach—starting from phenotype assessment through to chromosomal and molecular studies—is essential, particularly in 46,XY DSD where clinical presentations may vary widely. This report discusses a young adult with delayed-diagnosed 46,XY DSD who transitioned to male gender, analyzed through a stepwise framework.

CASE

A 20-year-old individual, assigned female at birth, presented with progressive virilization since early adolescence. The patient had no breast development or menstrual history. Instead, a deepened voice, facial and body hair, spontaneous erections, and wet dreams were reported. The patient urinated from an orifice beneath the clitoral area in a squatting position.

Physical examination revealed masculine features, gynecomastia, and clitoromegaly measuring approximately 3 cm in length. A bifid scrotum resembling labia majora was observed, with no palpable testes. Tanner staging was M2P4.

Hormonal analysis showed hypergonadotropic hypogonadism: LH 29.7 mIU/mL, FSH 47.8 mIU/mL, testosterone 21.1 nmol/L, estradiol 27.5 pmol/L. Karyotyping confirmed a 46,XY complement. MRI revealed bilateral gonads in the inguinal canals suspected as testes, and a uterine-like structure between the bladder and rectum. No ovaries or prostate were identified. FISH and SRY gene sequencing were performed; SRY was positive, and no pathogenic variants were found.

The presence of virilized phenotype, retained Müllerian structures, and undescended testes in a 46,XY individual suggests a disorder in androgen action or synthesis. While partial androgen insensitivity syndrome (PAIS) or 5α-reductase deficiency are possible, definitive diagnosis awaits further molecular studies such as SRD5A2 or AR gene sequencing.

CONCLUSION

This case illustrates the complexity of evaluating 46,XY DSD and emphasizes the utility of a stepwise diagnostic algorithm. Clinicians should remain vigilant to consider rare etiologies in late-presenting cases and provide multidisciplinary, gender-affirming care tailored to the patient's identity and needs.

EP_A160

REFINING THE DIAGNOSIS: A CASE REPORT ON THE ROLE OF FISH IN DETECTING SUBTLE MOSAIC KLINEFELTER SYNDROME

https://doi.org/10.15605/jafes.040.S1.168

Ardy Wildan,[1] Sarah Firdausa,[1,2] Em Yunir[1]

[1]Division of Endocrinology, Metabolism, and Diabetes, Department of Internal Medicine, Cipto Mangunkusumo Hospital, Faculty of Medicine, University of Indonesia

[2]Division of Endocrinology, Metabolism, and Diabetes, Department of Internal Medicine, Faculty of Medicine, Universitas Syiah Kuala, Aceh, Indonesia

INTRODUCTION/BACKGROUND

Mosaic forms of Klinefelter syndrome (KS) can pose a diagnostic challenge, particularly in patients with a normal male phenotype and unremarkable hormonal profiles. While conventional karyotyping is a widely used first-line tool for detecting chromosomal abnormalities, its sensitivity