

장골의 낭성섬유골염으로 발견된 원발성 부갑상선기능항진증 1예

동아대학교 의과대학 내과학교실1, 이비인후과학교실2, 핵의학교실3, 병리학교실4

\*박수경<sup>1</sup> · 김동균<sup>1</sup> · 박현수<sup>2</sup> · 강도영<sup>3</sup> · 홍숙희<sup>4</sup> · 김덕규<sup>1</sup> · 박미경<sup>1</sup>

원발성 부갑상선기능항진증은 혈청 칼슘의 측정이 용이해짐에 따라 어렵지 않게 진단할 수 있으며, 건강검진 등 혈청 칼슘에 대한 선별검사가 보편화됨에 따라 비교적 경미한 단계에서 발견되는 경우가 흔하다. 이에 따라 최근에는 낭성섬유골염(osteitis fibrosa cystica)이 동반되는 경우는 드물게 보고되고 있다. 낭성섬유골염이 있는 경우는 다발성 골수종 및 악성종양의 다발성 골전이 등의 감별이 필요하다. 저자들은 심한 고칼슘혈증과 함께 중증의 골병변이 동반된 원발성 부갑상선항진증의 증례를 경험하여 이를 보고하는 바이다. 왼쪽 골반뼈주위의 통증을 주소로 내원한 44세 남자 환자로, 골반 자기공명촬영에서 좌측 골반뼈에 형질세포종(plasmocytoma)이 의심되었고 전신뼈촬영에서 다발성 골병변이 관찰되었다. 이에 환자는 다발성 골수종에 대한 검사를 하였고, 좌측 장골에서 조직검사를 시행하였다. 하지만 조직검사에서는 낭성섬유골염이 의심되어, 혈청 부갑상선호르몬을 검사하였고 수치가 증가한 것을 확인하였다. 원발성 부갑상선항진증이 의심되어 시행한 경부 전산화단층촬영 및 부갑상선 MIBI-SPECT 에서 부갑상선 종양이 관찰되었고, 환자는 부갑상선 절제술을 시행후 부갑상선 선종으로 확진하였다.

A case of congenital adrenal hyperplasia mimicking Cushing syndrome

Division of Endocrinology and Metabolism, Department of Medicine, Samsung Medical Center,  
Sungkyunkwan University School of Medicine, Seoul, Korea

Mira Kang\*, You-Cheol Hwang, Hye Seung Jung, Kwang-Won Kim, Moon-Kyu Lee, Myung-Shik Lee

Congenital adrenal hyperplasia (CAH) is a common autosomal-recessive disorder. Steroid 21-hydroxylase deficiency (21-OHD) accounts for over 90% of CAH cases and presents as diverse manifestations from the salt-wasting to non-classical form due to a highly variable genomic mutation. In the case of simple virilizing or non-classical form, the symptoms related to the enzyme deficiency may be progressive so insidiously that it's not easy to identify the diseases. Impaired negative feedback by reduced cortisol causes an exaggerated rise of adrenocorticotrophic hormone (ACTH) levels, which stimulates the adrenal cortex to get hyperplastic, sometimes nodular or tumorous. The pathophysiology is basically a defect in biosynthesis of cortisol. Anyone, therefore, may not take into account CAH in the patient with hypercortisoluria. In thinking of incidentaloma with hypercortisoluria over 100 µg/24 h, we reach a tentative conclusion of Cushing syndrome. We report a case of 42 years old man with a left adrenal tumor and 226 µg/24 h of hypercortisoluria who was diagnosed as congenital adrenal hyperplasia. He visited to emergency room suffering from general weakness and dark urine. He was diagnosed as acute hepatitis A, based on positive anti-hepatitis A virus IgM. A 4 cm-sized left adrenal mass was incidentally identified on abdominal CT scan during the evaluation. His height was 152.5 cm and weight was 75.5 kg (Body mass index : 32.5). He had central obesity and acanthosis nigricans on the axilla which supported the hormonal problem such as Cushing syndrome. However, the 24-hour urine cortisol was decreased by over 90 % in the low dose dexamethasone suppression test. According to the high concentration of 17-hydroxprogesterone (300 µg/mL) and the precocious puberty at the age of 7 years, he was diagnosed as 21-OHD CAH hormonally and clinically. After one-year treatment with daily 0.25 mg of dexamethasone, the left adrenal tumor was substantially decreased. This implies that long term exposure to ACTH (about 35 years on the base of the clinical history) can develop benign adrenal tumors, and eventually cortisol might be produced abundantly by hypertrophied adrenal cortex.