2010년도 대한내분비학회 5월 학술집담회

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- 증례발표 -

제목 : 양측 부신피질증식증으로 인한 쿠싱증후군
소속 : 중앙대학교병원 내분비내과
발표자 : 이재희, 이석홍

50세 여자가 3년 전부터 몸이 부었으며, 고혈압으로 약물치료해 왔다. 1년전부터 전신부종이 악화되어 개인의원에 방문하였으며, 복부 초음파검사에서 신장암 의심되어 본원으로 의뢰되었다. 복부 전산단층촬영에서 각각 5 cm 이상의 양측 부신종양이 발견되었고, 호르몬 검사에서 24시간 소변 VMA, metanephrine, 혈장 aldosterone-renin ratio은 정상이었다. 24시간 소변 유리 cortisol이 상승하였으며, 저용량 dexamethasone 억제 검사에서 양성이었다. 추가 검사를 통해 ACTH 비의존성 쿠싱증후군으로 진단하였으며, 기능성 병변의 위치를 확인하기 부신정맥 채혈을 시도하였으나 접근이 용이하지 않았다. 복강경으로 좌측 부신절제술을 시행하였고, 병리소견 결과 부신피질증식증으로 확인되었다. 이후 시행한 저용량 dexamethasone 억제 검사에서도 고코티솔혈증 지속되어 추가적으로 복강경하 우측 부신 일부를 제거하였다. 수술 후 환자의 증상은 호전되었으며, 현재 외래에서 prednisolone과 fludrocortisone 처방으로 추적 진료 중이다.

제목 : Idiopathic Familial Hypoparathyroidism
소속 : 가천의대 내분비내과
발표자 : 엄영실

Hypoparathyroidism (HP) is characterized by low PTH levels, hypocalcemia, and hyperphosphatemia. HP is a rare complication seen most frequently after thyroidectomy, though idiopathic HP does occur. Although the incidence of HP has decreased substantially due to the progress of surgical techniques, it is still frequently encountered in clinics. The primary symptom of HPis spasm due to hypocalcemia, which until recently was treated with calcium and vitamin D. However, PTH replacement could be a more effective and physiologic treatment. Heterozygous mutations in pre-pro-PTH or the calcium-sensing receptor (CaSR) cause some forms of autosomal dominant HP (AD-HP). Furthermore, homozygous mutations in glial cells missing B (GCMB) have been implicated in autosomal recessive HP (AR-HP). In most other HP patients, however, the molecular defect remains undefined. We experienced two patients (proband and her son) with hypocalcemia potentially caused by a missense mutation of the *related* gene. The proband, aged 41, showed hypocalcemia and HP from infancy. She had been diagnosed as having idiopathic HP and had been treated with calcitriol. She gave birth to a male infant at age 27 years. Her sonwas found to have hypocalcemia (Ca, 6.6mg/dl), without seizure or tetany, when he was 14 months old. Our objectives are to determine the genetic defect in the affected members of family with AD-HP and define the underlying disease mechanism. Mutational analysis of the genes encoding pre-pro-PTH, CaSR, and GCMB is performed using PCR-amplified genomic DNA of the probands and other available members of family.

- 연구세미나 강의 -

제목 : Transcriptional control of hepatic glucose homeostasis
소속 : 성균관대학교 의과대학 분자세포생물학교실
발표자 : 구승회

Mammalian liver is critical in the maintenance of glucose homeostasis. Fasting hormones glucagon and glucocorticoid are responsible for activating hepatic gluconeogenesis via modulation of TORC2/CRTC2 and PGC-1a-dependent transcriptional programs. We have previously shown that CREB coactivator TORC2/CRTC2is a major regulator for hepatic glucose production by controlling key gluconeogenic gene expression including PEPCK and G6Pase. Families of AMPK-related kinases were shown to be involved in this process by catalyzing serine 171 phosphorylation of TORC2/CRTC2, whichpromotes its cytoplasmic retention and subsequent degradation process. Uncontrolled activation of TORC2/CRTC2 by insulin resistance has been attributed to the hyperglycemia and the progression of type 2 diabetes.
In the first part of the talk, we will discuss a novel role of TORC2/CRTC2 to promote insulinresistance by enhancing expression of LIPIN1, a mammalian phosphatidic acid phosphatase using mouse models. In the second part, we will also discuss a role of ER-bound factor, CREBH, in the control of hepatic gluconeogenesisas a critical component of a CRTC2-dependent transcriptional machinery.