<< 3월 학술집담회 증례발표 >>

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제목 : On Unrarity of Latent Cobalamin Deficiency : Diagnostic pitfalls based on 7 cases of latent Cobalamin deficiency & an adult type pernicious anemia
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Vitamin B12(Cobalamin=Cbl) 결핍 질환은, classic pernicious anemia 외에, 기타(~>80%) 경우 autoimmune endocrinopathy, metformin장기간 사용, 만성 위축성 위염, 위장관 절제술후 등등에서 유의할 진단,관리대상으로 관심이 고조되고 있다. 저자는 최근 1년간 지역병원에서 경험한 7예의 Latent Cbl deficiency와 adult pernicious anemia 1예의 임상양상을 요약 보고하며, 또한 screening test인 serum Cbl & folate 수치해석시 주의점,누락진단의 상황을 언급하고자 한다. 증례는 autoimmune hypothyroidism 2 예, classic adult pernicious anemia 1예, metformin 1g/day 3년간 복용한 당뇨병 1예, 위암 절제술 5-8년후의 4예이며,이중 1예에서 serum Cbl>2,000pg/mL이었으나, MMA assay 실패 & high homocysteine과, 치료 시도에서 확인된 "False high Cbl" 경우 이었다. 증세는 megaloblastic anemia 및 신경소견이 확인되는 pernicious anemia와, 위 절제후 3예, 나머지 4예는 경미하거나 무증상이었고, 이중 2예의 Hypothyroidism에서 type-A autoimmune gastritis가 중증 IDA와 함께 "Food-iron malabsorption"상태이었다 . Latent (mild, subtle, subclinical, undiagnosed, neglected) Cbl deficiency의 잠재적 위험성을 강조하며, Cbl assay kit의 제조사별 심각한 결과차이, " False high Cbl "의심시 고려할 사항에 대해 문헌 조사 하였다.

제목 : Calcimimetic Use in Primary and Secondary Hyperparathyroidism
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발표자 : 김경민, 구철영, 신동엽, 최한석, 이유미, 이은직, 임승길

Hyperparathyroidism (HPT) is characterized by inappropriately increased parathyroid hormone (PTH) concentration. HTP alter metabolism of calcium and phosphorous, and contribute soft tissue or vascular calcification, cardiovascular disease and bone loss. Primary hyperparathyroidism (PHPT) is characterized by incompletely regulated, excessive secretion of PTH from one or more parathyroid glands. The most common cause of PHPT is parathyroid adenoma. Secondary hyperparathyroidism (SHPT) is caused by hypocalcemia and hyperphosphatemia, common in patients with chronic kidney disease, affecting most of those who are receiving dialysis. The only definitive treatment for PHPT is parathyroidectomy and some cases of severe SHPT are also indicated for surgery. However, for those who do not undergo parathyroidectomy, calcimimetic agent is a novel medical therapy for HPT. It increases the sensitivity of the calcium sensing receptor to extracellular calcium ions, inhibiting the release of PTH and lowering PTH levels within a few hours after administration.

We experienced two HPT cases, one was PHPT with multiple endocrine neoplasia type I (MEN I) and the other was SHPT in dialysis patient. The first case is a 83-year-old woman who was admitted to the hospital because of stroke. She had two parathyroid adenomas and pancreatic neuroendocrine tumor. Her daughter and son also had hypercalcemia, therefore she was presumed to have MEN I. We could confirm the diagnosis by finding a point mutation in MENIN gene(D350V, exon 7) during further genetic analysis both in this patient and her daughter. As she had cardiac arrest due to sudden attack of ventricular tachycardia just after admission, she had received limited parathyroidectomy on only one left superior gland under local anesthesia considering high mortality of general anesthesia. PTH level was rapidly normalized within few hours from 312 pg/ml to 87.13 pg/ml and serum calcium level also declined to 10.2mg/dl (ionized calcium 5.0mg/dl) from 13.2mg/dl (ionized calcium 6.55 mg/dl). However, calcium and PTH gradually increased, so, calcimimetic agent, cinacalcet 90mg was started once daily. Cinacalcet was tapered and maintained 30 mg once a day and the effect has been sustained over 34 months with calcium and PTH level within normal range. The second case is a 34-year-old woman with end stage renal disease on continuous ambulatory peritoneal dialysis. She was referred to our endocrinology clinic for persistent hyperparathyroidism despite the use of calcitriol and phosphate binders for 1 year. Two years earlier total thyroidectomy was done due to incidentally found papillary thyroid cancer, therefore the re-operation on neck was hard to go into. So cinacalcet was administered 30mg once daily and serum PTH level decreased markedly from 499 pg/ml to 209 pg/ml.

Cinacalcet rapidly normalized serum calcium and reduced PTH in patient with PHPT and these effects were maintained with long term treatment. Moreover, in management of SHPT with chronic kidney disease, cinacalcet is also effective. Cinacalcet may be a new important therapeutic option for treatment of not only PHPT but also secondary cause.