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- 증례 발표 -

제목 : Nonclassic 11 β Hydroxylase Deficiency with Thyrotoxic Periodic Paralysis: A Case Report
소속 및 발표자 : 조선의대 내분비내과 김진화

11 β hydroxylase deficiency (OHD) is one of the main causes of congenital adrenal hyperplasia (CAH). There have only been a few reported cases of non-classic 11 β OHD, which is a milder form of OHD. Moreover, the concomitant presence of non-classic 11 β OHD and another endocrine disease has not been previously reported. We present here a case of non-classic 11 β OHD combined with thyrotoxic periodic paralysis (TPP). To the best of our knowledge, this is the first such case to be reported in the English medical literature.

A 23-year-old male presented with paralysis of both lower extremities. His blood pressure was 170/100 mmHg. He had hypokalemia (1.9 mEq/L) and elevated serum levels of free T4 (3.48 ng/dL) (normal: 0.7-1.8 ng/dL) and T3 (530.2 ng/dL) (normal: 60-190 ng/dL) with a low TSH level (0.01 IU/mL) (normal: 0.25-4 IU/mL), which confirmed a clinical presentation of TPP. Abdominal computed tomography (CT) demonstrated hyperplasia of both adrenal glands. The basal- and ACTH-stimulated serum 11-deoxycortisol levels were 138 ng/mL and 739 ng/mL, respectively (normal: 0-120 ng/mL). The ACTH-stimulated serum cortisol levels were low. These results established the diagnosis of non-classic 11 β OHD. The results from molecular genetic analysis demonstrated hypofunctional genetic polymorphisms of the CYP11B1 gene, which were c.128G>A and c.595+12G>A.

It is currently unclear whether these two diseases dependently affect each other. This case may suggest a possible association of non-classic 11 β OHD with TPP. Increased levels of thyroid hormone may play an etiologic role in the development or aggravation of the symptoms of undisclosed 11 β OHD.

Key Words: non-classic 11 β hydroxylase deficiency, congenital adrenal hyperplasia, thyrotoxic periodic paralysis

제목 : Three cases of CNS Lymphoma involving hypothalamus and pituitary gland

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Primary central nervous system lymphoma (PCNSL) is an uncommon variant of extranodal non-Hodgkin lymphoma, it accounts for 1-2% of all malignant diseases of the central nervous system. Moreover, Primary pituitary lymphoma in immune-competent patients is very rare and has been described in less than 20 cases. We report three primary pituitary lymphoma cases in immune-competent patients.

Case 1: A 58-year-old woman presented with unusual behavior. A magnetic resonance imaging scan of the head was performed and revealed a hypothalamic and thalamic mass. Laboratory evaluation revealed hypopituitarism. Through performing a water deprivation test and a combined pituitary stimulation test, we diagnosed complete type central diabetes insipidus and panhypopituitarism. Primary CNS lymphoma was confirmed by a CT-guided stereotactic biopsy. The thalamic and hypothalamic masses were significantly decreased in size after chemotherapy.

Case 2: A 58-year-old woman presented to an ophthalmology clinic because of headache, visual disturbance, and dizziness. The visual field examination revealed temporal hemianopsia. The patient was referred to the department of neurology and brain magnetic resonance imaging was performed, which revealed a suprasellar mass. Laboratory evaluation revealed hypopituitarism and hyperprolactinemia. Through performing a water deprivation test and a combined pituitary stimulation test, we diagnosed complete type central diabetes insipidus and hypopituitarism and hyperprolactinemia. Primary CNS lymphoma was confirmed by a CT-guided stereotactic biopsy. The suprasella masses were significantly decreased in size after chemotherapy and radiotherapy.

Case 3: A 31-year-old man presented with fever of unknown origin. Laboratory evaluation revealed pan-hypopituitarism after other possible etiologies for fever were excluded. Through performing a water deprivation test and a combined pituitary stimulation test, we diagnosed complete type central diabetes insipidus and panhypopituitarism. Magnetic resonance imaging scan was performed and revealed a sellar mass. Primary CNS lymphoma was confirmed by a CT-guided stereotactic biopsy. The suprasella mass was significantly decreased in size after high dose MTX based chemotherapy.

제목 : Gene regulation of NFATc1 during osteoclast differentiation

소속 및 발표자 : 전남의대 약리학교실 김낙성

Osteoclasts are responsible for bone resorption. Receptor activator of nuclear factor κ B ligand (RANKL) induces osteoclast formation from hematopoietic cells via up-regulation of transcription factors, including NF- κ B, c-Fos, and nuclear factor of activated T cells (NFAT) c1. NFATc1, a key transcription factor, plays a role in regulating expression of osteoclast-specific downstream target genes such as tartrate-resistant acid phosphatase (TRAP) and osteoclastreceptor (OSCAR). c-Fos, a component of the AP-1 transcription factor, is well known to be an inducer of NFATc1 expression. The NF- κ B components p50 and p65 have been shown to mediate the induction of NFATc1 through their recruitment to the NFATc1 promoter. Thus, NF- κ B and c-Fos are important for the initial induction of NFATc1, which is then autoamplified by NFATc1 transactivation of its own promoter. Autoamplification of NFATc1 is required for the robust induction of NFATc1 and is regulated by a calcium-dependent pathway. In addition to the transcriptional regulation of NFATc1 during osteoclast differentiation, we discovered that M-CSF induces NFATc1 degradation through Cbl-mediated NFATc1 ubiquitination during late stage osteoclast differentiation. Furthermore, we found the post-translational modifications of NFATc1 regulated by RANKL and elucidated the factors involved in determining the state of post-translational modification of NFATc1.