<< 11월 학술집담회 증례발표 >>

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제 목: Nonclassical 21-Hydroxylase Deficiency
소 속: 이화여자대학교 의학전문대학원 내분비내과
발표자: 심은진

15세 여자가 월경이 없어서 왔다. 임신 40주에 제왕절개로 출생하였고 출생체중은 2.7 kg이었다. 초등학교 시절 같은 나이의 아동에 비해 키가 컸으나 6학년 이후 거의 자라지 않았다. 혈압 130/70 mmHg, 신장 154 cm, 체중 60.5 kg였다. 유방이 거의 발달되지 않았고, 어깨 근육이 발달되어 있었으며 외부 생식기의 이상은 없었고 음모와 액와모의 발달은 정상이었다. LH 2.8 IU/L FSH 7.0 IU/L, E2 17 pg/mL, TSH 2.7 uIU/mL, prolactin 19.9 ng/mL 였고 ACTH 자극검사 소견은 표와 같다. CYP21A2 유전자의 compound heterozygote (c.[515T>A] +[293-13A>G] (p.Ile172Asn + splicing mutation))였다. Nonclassical 21-hydroxylase deficiency로 취침전 dexamethasone 0.5 mg 복용 4개월 후 월경이 있었다.

ACTH Stimulation Test	Basal	30min	60min	90min	120min	치료 후
Progesterone(ng/mL)	10.94	71.11	51.65	61.48	50.62	1.73
17-OHP (ng/dL)	24917	89462	75050	91748	78283	181
DHEA (ug/dL)	24.60	>30	>30	>30	>30	26.5
Testosterone (ng/dL)	275	276	283	252	263	27.8
Cortisol (ug/dL)	6.1	7.8	8.6	9.2	9.7	0.1
Aldosterone (pg/mL)	378.08	549.85	612.97	506.61	764.49	245.68
ACTH (pg/mL)	44					24
Renin (ng/mL/hr)	3.08					4.51

⇧
Dexamethasone 0.5 mg 복용

제 목 : Mutational Analysis of NF-1 Gene in a Family of Neurofibromatosis type 1 accompanied by Pheochromocytoma
소 속 : 가천의대 길병원 내분비내과
발표자 : 이현승

Neurofibromatosis type 1 (NF1) is one of the most common autosomal dominant inherited disorder affecting nervous system and is associated with mutations in the *NF1* gene, which is located on chromosome sub-band 17q11.2and contains 57 exons spanning approximately 300 kb of genomic DNA. NF1 is caused by a loss of function mutation of the *NF1* gene resulting in inactivation of neurofibromin, as a tumor suppressor, which encodes a GTPase activating protein (GAP) involving in the negative regulation of Rasactivity. GAP related domain, which is encoded by exons 20-27a, and cysteine-serine rich domain are well known to be mostly important functional domains in neur ofibromin .In spite of many genetic analysis of NF1 related pheochromocytoma, there still remains the question of whether pheochromocytoma is a true component of NF1. We found a novel germline mutation of *NF-1* gene (c.7907+1G>A) in a 31-year-old female patient with NF-1 accompanied by pheochromocytoma and genetic analyses of her family members showed that this mutation leads to skipping of exon 45 during *NF1* mRNA splicing. As this novel germline mutation falls into neither GAP related domain nor cysteine-serine rich domain but into C-terminal region of *NF1* gene, which is relatively not well known for its function, this suggests that there must be essential function in the C-terminal region of *NF1* gene, especially in the development of NF-1 related pheochromocytoma. Therefore, we should necessarily consider the mutation of C-terminal regionin *NF1* gene when we encounter the patient with NF1 accompanied by pheochromocytoma.