■ S-449 ■

Thyrotoxic periodic paralysis associated with a mutation in the sodium channel gene SCN4A

가톨릭의대 인천성모병원 내분비내과

*이연희, 모은영, 김민지, 김은숙, 한제호, 문성대

Background: Thyrotoxic periodic paralysis (TPP) is rare complication of thyrotoxicosis with unknown etiology. TPP is acute attack of muscle weakness and hypokalemia with similarities to familial hypokalemic periodic parpalysis (FHPP). FHPP is autosomal dominant disease associated with mutations in CACNA1S(Ca channel, voltage dependent, L type, alpha 1S subunit) and SCN4A (Na channel, voltage-gated, type IV, alpha subunit). Similar clinical manifestations of TPP and FHPP suggest strong genetic predisposition. However TPP patients with mutations in CCNA1S and SCN4A were rare and only one TPP pedigree was reported to have SCN4A mutation. Case)A 28-year-old man admitted for numbness and weakness in lower limbs, distally symmetrically, which progressed proximally. He took large meal and alcohol. He had distal tremor, sweating and 5kg loss during 4weeks. Initial blood pressure was 130/80mmHg and heart rate 112beats/min. Physical exam revealed resting tremor on stretched hands without palpable goiter. Neurologic exam revealed lower limbs motor grade 0/5 and decreased deep tendon reflex. Initial K+ level was 2.0 mEq/L and ECG shows ST depression, U-wave, and high QRS Voltage. Initial diagnosis of hypokalemia periodic paralysis was kept and IV K+ 20 mEq/L with oral replacement was started. TSH was 0.05 μIU/L (0.34-5.6), free T4 2.05 ng/dL (0.58-1.64), T3 216.73 nmol/L (87-178), and TSH receptor Ab 7.53 (normal 0.3-1.75). Thyroid scan revealed increased uptake in thyroid. Patient was started propranolol 20 mg TID and Methimazole 20 mg/day and discharged with diagnosis of TPP secondary to Graves' disease. Two weeks later, he re-admitted for lower limbs weakness. K+ level was 2.6 mEq/L, corrected with oral replacement. After 1 year follow-up, the patient remains euthyroid and symptom free. **Result:** Direct automated sequencing of SCN4A exon 24 PCR product in proband revealed heterozygous C→A transition at Thr1646Asn. The same nucleotide change was found in his brother and mother. Father's PCR products had normal sequence. There were no mutations found in exon 11 and 30 of CACN1AS. **Conclusion:** We report novel mutation in SCN4A in patient with TPP whose family had been asymptomatic until developing thyrotoxicosis caused by Graves' disease.

■ S-450 ■

A case of infected thyroid cyst

대구가톨릭대학교병원 내과

*이준기, 정의달, 이지현, 손호상

A 33-year-old Korean female patient with no specific past medical record and no recent pregnancy history diagnosed right thyroid single benign follicular nodule by the method of thyroid ultrasonography (USG) guided fine needle aspiration (FNA) in the other medical clinics. After six months later, she just wanted to follow up in our medical center, and we checked her medical record and follow up with the thyroid nodule by thyroid USG-guided FNA. We found the USG with FNA follow result was right thyroid benign follicular nodule, and planned to follow up this nodule in six months later. After 1 month, she came again to outpatient clinic of our medical center, and she got the problem of neck swelling, neck pain with tenderness, and swallowing difficulty, but there was no fever. As we underwent laboratory work up, white blood cell (WBC) count was 10,700 (3,600-9600) /μL, C-reactive protein (CRP) was 93.9 (＜5) mg/L, erythrocyte sedimentation rate (ESR) 51 (0-20) mm/hr, thyroid stimulation hormone (TSH) 0.8 (0.4-4.7) μIU/ml, free T4 1.17 (0.8-1.9) ng/dL, T3 0.69 (0.8-2) ng/mL. Thyroid dynamic computed tomography (CT) revealed a 4.2×3.6×4.8 cm sized cystic mass involving right thyroid lobe with combined hemorrhagic fluid. And then we follow up the thyroid nodule by USG-guided FNA, and the result was the right thyroid nodule size abrupt increasing, peri-thyroidal soft tissue swelling, and cystic change with many neutrophils. In the same time, as we analyzed the fluid portion of the nodule, the fluid aspirated 9cc was dark-brown colored, and neutrophil count was 57800 /μL. So we concluded the thyroid nodule had cystic change and complicated infected thyroid cyst. We prescribed Cefixime 200mg/day per os (PO), and acetaminophen for pain control. After six weeks later, we followed up the patient, and we found that the symptoms had subsided and also neck swelling and tenderness had relieved as well. And we followed up by USG and CT, the size of the nodule with cyst decreased in a half before, estimated volume of cystic portion was 2.33 mL, compare to initially estimated volume 30.9 mL. Now she is following up the thyroid nodule annually on outpatient without complications or recurrence.