Medicine beyond frontiers

The 42nd Annual Medical Congress

FP8

FGFR-Related Disorders in Thai Patients

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Objectives: To characterize clinical features and molecular defects of Thai patients with achondroplasia, Crouzon, Apert and Pfeiffer syndromes and to determine the frequency of an S249C mutation of the Fibroblast Growth Factor Receptor 3 (FGFR3) in Thai patients with cervical carcinoma.

Methods: DNA was extracted from peripheral blood. For achondroplasia, we PCR amplified exon 10 of *FGFR3* and digested the PCR products with *SfcI*. For Apert syndrome, we PCR amplified exon 8 of *FGFR2* gene and digested with *MboI* and *BglI*. For Crouzon and Pfeiffer syndromes, we PCR amplified exon 8 and 10 of *FGFR2* and exon 5 of *FGFR1* and sequenced the PCR products. For the S249C mutation of *FGFR3*, we nested PCR amplified segment of *FGFR3* and digested the products with *Fnu4H1*.

Results: We identified and molecularly characterized 3 Thai patients with achondroplasia. All of them had the G380R mutation of the *FGFR3*. Three patients with Crouzon syndrome had different mutations; C278F, S347C, and S351C mutations of the *FGFR2*, each. Two patients with Apert syndrome had S252W while the other two had P253R of the *FGFR2*. A patient with Pfeiffer syndrome had an A344P mutation of the *FGFR2*. None of the fifty Thai patients with cervical cancer were found to have the S249C.

Conclusion: Molecular defects of studied Thai patients with achondroplasia, Crouzon, Apert and Pfeiffer syndromes were successfully identified which may provide an efficient tool for prenatal diagnosis in these families. The S249C mutation of FGFR3 is uncommon in Thai patients with cervical carcinoma.