FGFR-Related Disorders in Thai Patients

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 SfoI. For Apert syndrome, we PCR amplified exon 8 of FGFR2 gene and digested with MboI and BglII. 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.