Clinical management as well as the genotypic and phenotypic analysis of 16 different hereditary MTC pedigrees from the mainland China

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Background: There is still a need of data on MEN (multiple endocrine neoplasia) for variant RET mutations based on risk for aggressive MTC in China.

Methods: Sixteen different pedigrees have been taken from China and the efficacy as well as genotype and phenotype were analyzed retrospectively.

Results: The gene mutations of 16 different MTC pedigrees (86 patients and 10 clinically asymptomatic RET mutation carriers) are as follows: Risk Level A: p.S891A (N=1), p.L790F (N=2); Risk Level B: p.C618R (N=3), p.C620R (N=4); Risk Level C: p.C634R (N=6), p.C634Y/D707E (N=1), p.C634Y/V292M (N=1), p.C634Y (N=2), p.C634S (N=1); Risk Level D: R982H (N=1); the mean age of MTC is 36.74±13.408y (14-77y). The incidence of PHEO is 32.56% (n=28), the bilateral rate of which is 100% with an average age of 39.09±9.451y (23-55y); the incidence of PTH is just 1.16% (n=1.22y); papillary thyroid carcinoma was found simultaneously in 2 cases; one esophagus involved, distant metastasises are found in 10.46% (n=9,15-69y: Liver (n=3), lung (n=3), bone (n=2), breast (n=1)); mortality rate is 6.98% (n=6). A female patient suffered from MTC, PHPT, PHEO, CLA and pituitary adenoma, fibroadenoma, back fibroma, knee vascular smooth muscle tumors and uterine fibroids simultaneously. Complete operation data of 22 patients was collected (9 cases were treated in our hospital, while the others were operated outside).

Conclusions: The clinical phenotypic differences still vary widely, even in the same RET gene point mutation, the presence of this phenomenon may be due to environmental or the joint influence of other related genes but the exact mechanism still needs further study.

Biography
Xiaohong Chen is MD and a Professor. In 2005, he has obtained his ENT Doctor degree and he has worked as Post-doctor in UCLA from 2009-2010. He is the Vice-Director of Otolaryngology, Head and Neck Surgery Department in Beijing Tongren Hospital, Capital Medical University, China. He is a Member of the Committee of Beijing Anticancer Association and the Commission of Chinese Anti-Cancer Association and Melanoma Committee. He is an expert in diagnosis and treatment on rare diseases of head and neck, operation design for maintaining function of important organs and cosmetic result of incision design in head and neck tumor surgery, retention function of eye of the patients with the malignant nasal cavity and paranasal sinus tumor, salvage or radical surgery for advanced tumor of head and neck and familial hereditary tumor.

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