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A Short Commentary on "VHL Gene Mutation Analysis of a Chinese Family with Non-Syndromic Pheochromocytomas and Patients with Apparently Sporadic Pheochromocytoma"

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VHL gene is a tumor suppressor gene encoding two different protein VHL (pVHL30 and pVHL19), which mutations are related to the pathogenesis of VHLD [1]. We have reported VHL gene mutation analysis of members in a Chinese family with non-syndromic PCCs and those with apparently sporadic pheochromocytoma (ASP).

Mutations Types and Locations

We have found three novel VHL gene mutations, including 2 missense mutations and 1 insertion mutation (H125P, R120T and 623 (ˆ TTTGTtG)), which have been located in exon 2 encoding β-domain of pVHL. H125P mutation has been detected in the 5 probands with non-syndromic of the Chinese family (5 patients and 15 relatives). R120T and 623 (ˆ TTTGTtG)) mutations have been detected in 3 among 41 ASP patients.

Genotype and Phenotype

These mutations have resulted in dysfunction pVHL affecting a series of processes, including cell-cycle regulation, mRNA stability, hypoxia-inducible gene expression and transcription, etc. [2]. The clinical phenotype in patients carrying mutations are associated mainly with VHL type 2B or type 2C: H125P and R120T could be associated with VHL type 2C and 623 (ˆ TTTGTtG) could be associated with VHL type 2B or type 2C.

Diagnosis and Treatment

The genetic analysis is helpful for early diagnosis and medical management of patients with VHLD, including timely diagnosis of non-syndromic PCCs, accurate genetic counseling and predictive testing, tumor resection, postoperative care and close follow-up based on gene-specific clinical protocols, which is also beneficial for research of the VHLD pathogenesis [3-5].

Recent Treatment Progress

The high throughput sequencing technology (second-generation sequencing technology) has achieved rapid development in recent year. By this technology, we can detect the familial pheochromocytoma-associated mutations (SDHB, SDHC, SDHD, VHL, MAX, TMEM127 and RET) precisely and rapidly, which can provide a more comprehensive gene-level treatment information.

According to the results of the high throughput sequencing detection, we can determine if it is single genetic disease or not. If patient with single genetic disease, he can accept the treatment by pre-implantation genetic diagnosis (PGD) and pre-implantation genetic screening (PGS) in order to prevent the mutations onset of offspring [6].

Declaration of Interest

The authors declare that there is no conflict of interest that could be perceived as prejudicing the impartiality of the research reported. And our research findings are true and reliable.

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