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Genetic diagnosis of oculocutaneous albinism and functional studies of associated genes

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Oculocutaneous albinism (OCA) is a heterogeneous and autosomal recessive disorder with hypopigmentation in eye, hair and skin color. Six genes, *TYR, OCA2, TYRP1 SLC45A2, OCA5* and *SLC24A5*, have been identified as causative genes for non-syndromic OCA1-6 respectively. For syndromic OCA, at least 13 genes, HPS1-9 for Hermansky-Pudlak syndrome, CHS1 for Chediak-Higashi syndrome, *GS1-3* for Griscelli syndrome, have been characterized. We have implemented an optimized strategy for the genotyping of more than 300 Chinese OCA patients. We have identified over 70 previously unreported alleles in several OCA genes including *TYR, OCA2, SLC45A2, SLC24A5* and *HPS1*. We found that the mutational spectrum is population specific in Chinese (different from Caucasian and Japanese). We characterized the abnormal melanosomal localization of several commonly occurred alleles of *TYR* and *SLC45A2* in Chinese OCA patients. We examined the melanosomes in the skin melanocytes of these OCA patients and found that more immatured melanosomes were present in an OCA6 patient. Furthermore, the SLC24A5 protein was reduced in steady-state levels in mouse HPS mutants with deficiencies in BLOC-1 and BLOC-2. We further investigated the melanosomal localization in multiple HPS melanocytes. Our results suggest that SLC24A5 is required for melanosome maturation and is transported into mature melanosomes by HPS protein associated complexes (HPACs). The results of thins study will be translational and significant for gene diagnosis and prenatal diagnosis of OCA in China.

Biography

Aihua Wei got her M.D., Ph.D. from Capital Medical University in 2010. She works as an Assosiate Chief Physician in Beijing Tongren Affiliated Hospital of Capital Medical University. She is engaged in the clinic and research work on Genodermatosis, especially gene diagnosis and genetic consuling of albinism. She has published more than 10 articles about albinism in reputed journals.

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