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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, *CHSI* for Chediak-Higashi syndrome, *GSI-3* for Griscelli syndrome, have been characterized. An optimized strategy for the genotyping of more than 300 Chinese OCA patients was implemented. Over 70 previously unreported alleles in several OCA genes including *TYR*, *OCA2*, *SLC45A2*, *SLC24A5* and *HPS1* were identified. It was found that the mutational spectrum is population specific in Chinese (different from Caucasian and Japanese). The abnormal melanosomal localization of several commonly occurred alleles of *TYR* and *SLC45A2* in Chinese OCA patients was characterized. The melanosomes in the skin melanocytes of these OCA patients was examined and found that more immature 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*. The melanosomal localization in multiple HPS melanocytes was further investigated. 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 this study will be translational and significant for gene diagnosis and prenatal diagnosis of OCA in China.

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Evolution of surgical management of vitiligo: From tissue grafts to cellular grafts

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Vitiligo commonly known as leukoderma is an acquired disorder of pigmentation due to loss of melanocytes from basal layer of epidermis. Although vitiligo is a disease of unknown etiology, therapy and prognosis have improved remarkably during the past few decades after unraveling most of the histological, biochemical, immunological and molecular events occurring within and around the pigment cell. Surgical attempts to treat a disease that appeared initially as an exclusive medical condition began in the middle of 20th century and different approaches and refinement have been successfully reported since then. A number of surgical treatments are available including autologous thin thiersch grafting, suction blister epidermal grafting, autologous minipunch grafting, autologous cultured melanocyte grafting, autologous non-cultured epidermal cell suspension grafting and many more. Tissue grafting techniques are very simple and effective methods of vitiligo surgery requiring no laboratory setup. However, it has few drawbacks as it requires a large amount of donor skin and postoperative hyperpigmentation and perilesional halo are more common. Cellular grafting techniques are more refined methods of vitiligo surgery requiring minimal donor skin and covering maximal recipient area. Excellent color match, minimal postoperative discomfort and higher patient satisfaction rates are other advantages of cellular grafting techniques. But in recent times, cellular grafting techniques are becoming more favorable because of less sophistication, easy availability, lower costs and better outcomes. Among the various cellular grafting techniques, non-cultured epidermal cell suspension is the most widely practiced method. Soon we hope to formulate guidelines regarding the preferred method of vitiligo surgery for a particular type of vitiligo.

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