Synopsis of Original Research Paper

Investigation and functional analysis on the responsible genes for oculocutaneous albinism

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Oculocutaneous albinism (OCA) is a group of autosomal recessive disorders characterized by reduced or absent biosynthesis of melanin in melanocytes of the skin, hair, and eye. OCA is caused by mutations in the genes associated with melanin synthesis. So far, 16 genes have been reported to be involved in OCA. We have investigated the mechanism of melanogenesis. Here, we describe the analysis of the OCA genes in Japanese OCA patients and the identification of ten these patients, one OCA1, one OCA2, and eight OCA4. Ten novel mutations (c.445delTA in *TYR* gene for OCA1, p.792V in *P* gene for OCA2, and p.Y49C, p.G89R, P.C229Y, p.T437A, p.T440A, p.G473A in SLC45A2 gene for OCA4) were detected. Furthermore, we analyzed an OCA patient who was a Moroccan origin Belgian boy with type 4 oculocutaneous albinism (homozygous p.H38R mutation in the *SLC45A2* gene). Functional analysis by transfection of the mutant OCA4 cDNA, p.D157N, or p.G188V which had been reported as high frequent mutations in Japanese OCA4 into *uw*-mutant melanocytes established from OCA4 model mice showed that these missense substitutions are pathologic, resulting in these variant being unable to produce melanin. The p.H38R mutation was also investigated for the function in melanogenesis, and revealed to have no activity for melanogenesis. This is the first report on patient with OCA4 identified among African origin ethnic groups.