Ray et al. Genome Biology 2010, 11(Suppl 1):P32
http://genomebiology.com/2010/11/S1/P32

Genome Biology

POSTER PRESENTATION Open Access

Comprehensive analysis of the molecular bases of OCA in Indians

Kunal Ray[1*], Mainak Sengupta[1], Moumita Chaki[1], Maitreyee Mondal[1], Swapan Samanta[2]

From Beyond the Genome: The true gene count, human evolution and disease genomics
Boston, MA, USA. 11-13 October 2010

Background

OCA is a group of autosomal recessive disorders characterized by hypopigmentation and abnormalities related to ocular development. Mutations in genes regulating melanin-biosynthesis cause four classical types of OCA (OCA 1-4). The clinical spectrum of OCA often depends on the pigmentation threshold of a patient, highlighting the importance of ethnic- specific SNPs. We aimed to understand the molecular bases of OCA in India, where it is one of the four major causes of childhood blindness.

Materials and methods

Blood samples were collected from OCA patients and family members, mostly from eastern and southern India. Seven pigmentation related genes were screened for variations. Relevant non-synonymous changes in tyrosinase (TYR) were functionally validated. Eighteen SNPs from three OCA genes were genotyped in 552 normal individuals covering various ethnic groups of India.

Results

Our data suggest that defects in *TYR* cause albinism in 58% (36/62) of the cases [1] (and unpublished data; see Figure 1). Functional assays with missense mutations proved that none of mutants are enzymatically active and are retained in the endoplasmic reticulum [1]. Screening of the remaining cases (43%) revealed *OCA2* to be the second common locus followed by *SLC45A2* [2] (Figure 1). Evaluation of SNPs in *TYR, OCA2* and *SLC45A2* in normal population suggested definitive bias for some of the SNPs towards specific populations.

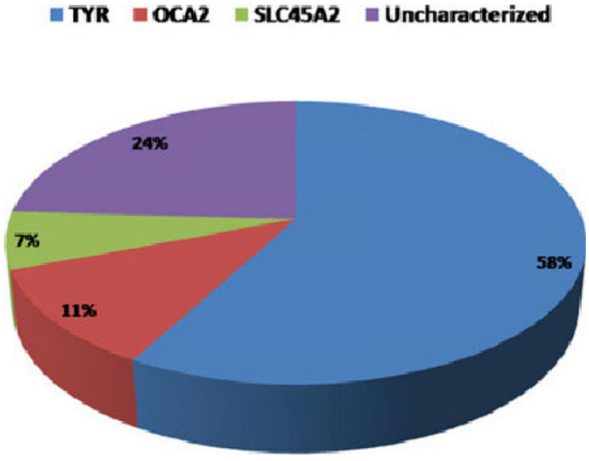

Figure 1 Relative contribution of OCA-associated genes towards OCA pathogenesis in India.

Conclusions

Our investigation suggests that ~58% of OCA in India belong to OCA1 category. ER retention is the major cause of lack of TYR activity in OCA1 patients. Information on allelic distribution of SNPs is important for cosegregation analysis of candidate genes in affected families.

Acknowledgement

SNP analysis in Indian population groups was done as part of a larger study to capture genomic variation among Indians by the Indian Genome Variation Consortium.

Author details

[1]Molecular and Human Genetics Division, Indian Institute of Chemical Biology (a CSIR unit), Kolkata 70032, India. [2]Department of Ophthalmology, Calcutta National Medical College and Hospital, Kolkata 700014, India.

Published: 11 October 2010

[1]Molecular and Human Genetics Division, Indian Institute of Chemical Biology (a CSIR unit), Kolkata 70032, India
Full list of author information is available at the end of the article

References

1. Chaki M, Sengupta M, Mondal M, Bhattacharyya A, Mallick S, Bhadra R, Consortium Indian Genome Variation, Ray K: **Molecular and functional studies on Tyrosinase among Indian Oculocutaneous Albinism type 1 (OCA1) patients.** *J Invest Dermatol* 2010.
2. Sengupta M, Mondal M, Jaiswal P, Sinha S, Chaki M, Samanta S, Ray K: **Comprehensive Analysis of the Molecular Basis of OCA in Indian Patients Lacking Mutation in Tyrosinase Gene.** *Br J Dermatol* 2010, Epub ahead of print.

doi:10.1186/gb-2010-11-S1-P32
Cite this article as: Ray *et al.*: Comprehensive analysis of the molecular bases of OCA in Indians. *Genome Biology* 2010 **11**(Suppl 1):P32.