

MONDAY

# COLLOQUIUM

Our origin, culture and consequences: genetic perspectives

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30 Mar 2026 (Monday) | 16:00 Hrs (Tea / Coffee 15:45 Hrs) | Venue: TIFRH Auditorium

Indian subcontinent is a region of remarkable cultural, linguistic, and genetic diversity with over 4,500 anthropologically well-defined groups. We have been studying various South Asian populations to understand their origin, affinities, and impact of endogamy. Our genetic study provided evidence that the enigmatic tribal populations of Andaman Islands are the first modern humans, who migrated out of Africa (*Science*, 2005). Subsequently, we demonstrated that the contemporary Indian populations have descend from two divergent groups: (1) Ancestral South Indians & (2) Ancestral North Indians (*Nature*, 2009), and these two founding groups have admixed during the past 2000 – 4000 years (*Am. J. Hum. Genet.*, 2013). Since then, almost all the populations in Indian subcontinent have been practicing endogamy. To assess the impact of endogamy, we have analysed 275 distinct South Asian groups and found that 81 groups have strong founder event than the one that occurred in both Finns and Ashkenazi Jews (*Nat. Genet.*, 2017). Further, we went back to the populations that have strong founder event, and found that they have high prevalence of population-specific diseases. Notably, one of the populations has high frequency of Junctional Herlitz Epidermolysis Bullosa disease, characterized by vesicobullous skin lesions, oral mucositis, congenital heart disease, and premature death. We performed exome sequencing and found a novel homozygous mutation in the LAMB3 gene of the patients, whereas the parents were heterozygous. CRISPR/Cas9 mediated knockout of the above mutation exhibits the same phenotype in mouse (C57BL/6NJ) (*unpublished data*). Our continuing effort is to identify recessive mutations in the populations with strong founder event, provide prenatal and premarital counselling, which would help in eliminating the pathogenic mutation(s) from the population.