Abstract Title: Population Genomics and Public Health

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Abstract: Modern India is a region of remarkable cultural, linguistic, and genetic diversity with over 4,500 anthropologically well-defined groups. We have been studying various Indian/South Asian populations to understand the origin, migration, and impact of consanguinity and endogamy on our health and diseases. Our genetic studies provided evidence that the enigmatic tribal populations of Andaman Islands are the first modern humans, who migrated out of Africa about 65,000 years back. Subsequently, we demonstrated that the contemporary Indian populations have descend from two divergent groups: (1) Ancestral South Indians (ASI), and (2) Ancestral North Indians (ANI). These two founding groups have admixed during the past 2000 – 4000 years. Since then, almost all the populations of Indian subcontinent have been practicing endogamy. To assess the impact of endogamy, we have analysed samples from more than 2,800 individuals from over 275 distinct South Asian groups and found that 81 out of 275 groups, have a strong founder event than the one that occurred in both Finns and Ashkenazi Jews. Further, we went back to the populations that have strong founder event, and found some of the populations have high prevalence of population-specific diseases. Notably, Kallar population from Tamil Nadu has high frequency of Junctional Herlitz Epidermolysis Bullosa disease, characterized by vesicobullous skin lesions, oral mucositis, congenital heart disease, and premature death. Subsequently, exome sequencing found a homozygous 11 base pair deletion in the LAMB3 gene of the patients, whereas the parents were heterozygous for this deletion. Likewise, large number of population-specific diseases exist in endogamous populations. We are now analysing populations with strong founder event to understand the genetic basis of diseases, which would help us to provide prenatal and premarital counseling to avoid such diseases in the family/population in the future.

Area of expertise: population genetics, rare genetic diseases, cardiovascular diseases, mitochondrial disorders, male infertility and developmental sex disorders