## **UGC MINOR RESEARCH PROJECT SUMMARY**

## GENETIC EFFECTS OF CONSANGUINITY IN THE HOSPITAL POPULATION OF PALAKKAD DISTRICT, KERALA.

## Approved letter No: 2228-MRP/15-16/KLCA011/UGC-SWRO dated 25.4.2016 and Sanction letter No: 2228-MRP/15-16/KLCA011/UGC-SWRO dated 31.3.2016

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Consanguinity study was carried out in 3 hospital populations in Palakkad, Kerala. Data collected from the inpatients and outpatients of three hospitals in Palakkad using structured questionnaire. Hospitals were visited once in a month for data collection and this was repeated for six successive months. Data collected from a total of 2316 patients, of which 535 were from District Hospital, Palakkad, 1276 from Hospital for Women and Children and 505 from Community Health Centre, Nemmara.

The data classified based on social hierarchy and traditions into Nair, Chetti and Sc-St groups. Nair is a forward community and Chetti is an immigrant community of Kerala, while different Sc-St groups forming the socially lower strata and following almost same traditions are grouped together.

The patterns of different types of consanguineous unions in these three social groups are studied. The measures of consanguinity such as the frequency of inbreeding and the mean coefficient of inbreeding have been analysed. The frequency of inbreeding ranged from 10.04% in Nair to 26.95% in Chetti, and the corresponding measure of mean coefficient of inbreeding ranged from  $0.005\pm0.001$  to  $0.0159\pm0.0013$ .

The patterns of related marriages observed in this study include uncle-niece, first cousin, one and half cousin, and second cousin and the highest frequency of close-kin pattern observed is first cousin type in all three groups. Under first cousin type the matrilateral cross cousin sub-type showed highest frequency in all three groups. The uncle-niece pattern was observed only in Chetti and Sc-St groups.

Regarding socio-economic correlates that are influencing consanguinity the factors such as education level, occupation level and family income level of spouses were investigated. Consistent declining trend of consanguinity with increase in level of education was observed in the husbands and wives of the three studied groups and this negative correlation was found to be statistically significant. Regarding the level of occupation a steady subsiding trend on the rate of inbreeding was observed in the three studied groups but this association was found to be statistically significant only in Nair community. Considering the level of family income as a correlate of consanguinity, a constant diminishing trend in the rate of consanguinity was observed in Nair and Sc-St group, while the correlation was found to be significant only in Sc-St group. However, in Chetti community which is an immigrant group with highest frequency of consanguinity, it was observed that the rate of consanguinity remain unaltered by changes in occupation status and family income.

Temporal changes of inbreeding in the three groups were assessed to determine the direction of time trend, and were done by analysing the year of birth, year of marriage and age at marriage of spouses. Consanguinity by year of marriage showed a steady subsiding trend in all three groups (p<0.01). Rate of inbreeding by age at marriage revealed that higher rate of related are happening in cases where the age at marriage is low and in all three studied groups the correlation is found to be statistically significant (p<0.01). While analysing the year of birth of spouses it was found that higher percentage of close-kin marriages were observed in older generations while the occurrence is less in younger generations and this was found to be statistically significant in Nair and Sc-St groups (p<0.01).

Examining the geographical correlates, it was found that highest frequency of related marriages are occurring when the marital distance is zero, and as marital distance increase its occurrence found dwindling. This association was statistically significant in all three groups studied. Considering the place of residence, it was observed that in Nair community the rate of inbreeding is higher in rural area and less in urban places and was found to be statistically significant (p<0.01). However, in other two groups no such trend was observed.

Effect of inbreeding on couple fertility, mortality and morbidity was subjected to detailed study. Mean fertility rates are found to be invariably higher in the consanguineous categories of the three studied groups compared to their non-consanguineous counterparts. However this differential was found to be statistically significant only in Sc-St group (p=0.002).

However, in present study this increase in mean fertility rate is found to be statistically significant only in the consanguineous couples of the SC-ST group.

Mortality by consanguinity refers to the loss progeny from the stage of pregnancy up to the prereproductive stage and hence was studied in three stages such as pre-natal mortality (mortality before birth), post-natal mortality (after birth) and total mortality. Prenatal mortality was found to be constantly higher in consanguineous groups of all three groups studied and this was statistically significant (p<0.01). The total post-natal mortality was also observed to be higher in consanguineous couples of the three studied communities compared to non-consanguineous couples (p<0.01). The total mortality in consanguineous category ranged from 14.65 (Nair) to 16.52 (Chetti) and is constantly higher in consanguineous groups and this differential is observed to be statistically significant (p<0.01).

The morbid conditions detected among the inpatients of the hospitals in the present study are many, which can be classified as physical, sensory, mental, infectious and systemic. As the number of cases belonging to each of the medical defects/ diseases was not large enough, a discrete disease / abnormality wise analysis was not possible; and hence the pooled data comprising all the morbid elements together was analysed. Morbidity rates in consanguineous categories exhibited a statistically significant higher differential in all three studied groups.

In view of the high level of genetic risk effects of inbreeding noticed in all three studied communities, a few tangible measures of genetic counselling are proposed, aimed at bringing down the consanguinity rates, and thereby minimising the associated risk effects.

- (a) Literacy promotion is advised to bring down the rate of consanguinity.
- (b) Implementation of multi-pronged genetic counselling programmes.
- (c) Availing the modern medico-genetic methods and facilities for terminating at risk pregnancies to avoid defective children being born.