

## Spatial Genetic Heterogeneity Assessment - a regional case study

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**Introduction** Geographical genetics provides evidence about genetic heterogeneity among and within modern societies [1-2].

Assessing genetic heterogeneity within populations will contribute to (i) determination of parameters for genetic epidemiological studies, (ii) correlation of origin and disease prevalence in studies of human evolution, (iii) estimation of the influence of environmental, cultural and socioeconomic factors on the manifestation of disease, as well as (iv) prognosis about demand of pharmaceutical products.

Genetic landscape analysis can be defined as an interdisciplinary field which brings together genetics, evolutionary research, environmental science, sociology and socioeconomics by applying methods, models and theory from landscape ecology and geodesic disciplines of spatial pattern modelling. The genetic landscape approach opens promising insights into the understanding of high resolution regional genetic heterogeneity of human populations and its consequences today. Genetic Geographic Information Systems (GGIS) provide the tools necessary for genetic landscape analysis. The German population can be characterized as a fairly old one, large in size and inhabiting an extended and diverse geographical area. For the German population, a complicated geographical pattern of genetic variation is expected a priori, characterized by clines and genetic barriers.

Subjecting genetic information to clusters and ordinations, thus revealing genetic affinities that have unsurprising geographic, linguistic and cultural parallels can lead to models and analysis of spatial genetic heterogeneity within the German population. Study areas of interests are those containing a high density of probands and exhibiting contrasting socioeconomic, cultural, and ethnicity levels.

**Material and Methods** Target sample group is comprised of non affected probands, self-reported family ancestry as of German ethnicity within epidemiological studies.

Zip-code area defines the minimal spatial unit. Zip-code area provides a spatial reference of each proband without harming probands' anonymity. The case study involves a sample of controls (ca. 700 probands), genotyped for coding and neutral SNPs (ca. 200 SNPs), collected in the Southern Germany (Ausburg and surroundings). Besides genotypic information, sociocultural characterisation of the probands is incorporated to the analysis.

Spatial pattern is assessed by quantifying variation of allele frequencies in the geographical space. Effect of different type of loci is explored. Analysis is conducted separately for coding and neutral loci and under indifference of location of loci (merging loci from neutral and coding regions).

Landscape structure is analysed quantifying landscape indexes: patchiness, connectivity and fragmentation [3]. Spatial trends are identified investigating the similarity of values between pairs of localities [4].

Departs from random spatial distribution is attested. Furthermore, correlations between spatial pattern of allele frequencies and sociocultural parameters are quantified with Mantel test statistics [5].

**Results and Discussion** The presented methodology GGIS provides a framework for quantifying genetic heterogeneity within a defined geographical space.

Presently, enormous amount of genetic data is being generated (SNPs, STRs). Extending the cooperation between epidemiological centres will enable assessing the spatial genetic heterogeneity within German probands and estimating its impact within epidemiological research.

## References

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