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Of Alps and Men: On Social and Y-Chromosomal Markers from Tyrol, Austria



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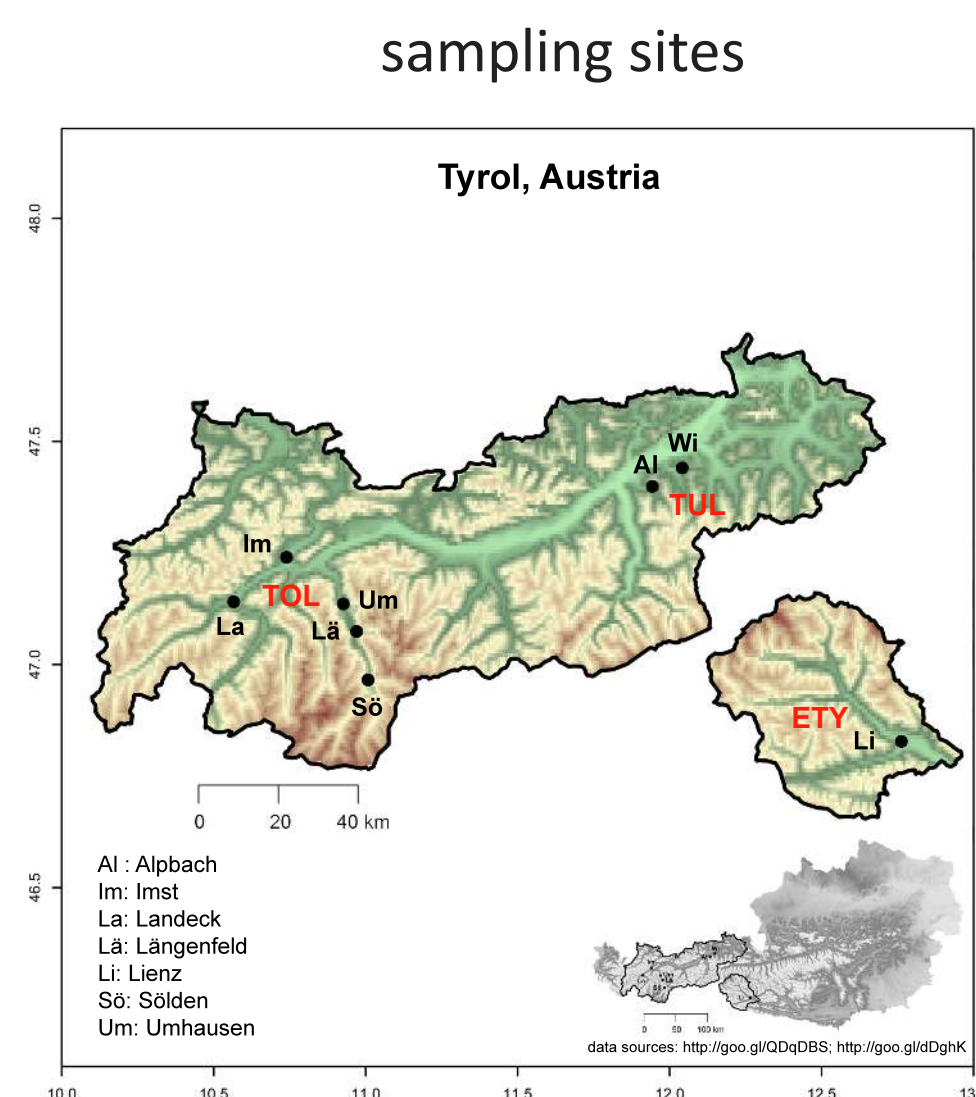
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Background Alpine landscapes impede human dispersal and both migration and permanently habitable areas are largely restricted to mountain valleys. The complex topography results in complex patterns of human settlement characterized by communities with differing degrees of connection and size, even at the microgeographical scale. This is likely to leave its mark on the local genetic make-up, as successful movement of individuals between communities forms a basis of genetic diversity. On top of that, social factors, such as inheritance law fostering patrilocality or authoritarian restrictions in access to marriage and reproduction, can form barriers with low permeability to gene flow and will support non-random mating. In general, larger and/or more connected communities will be less susceptible to the interplay of these different factors, whereas small and isolated groups are at a higher risk of becoming subject to the effects of genetic drift and losing genetic diversity.

On this background, we set out to characterize potential differences and commonalities among small, in historical terms rather isolated settlements and larger, more connected municipalities from different parts of Tyrol. Genealogical metadata, as reported by the voluntary study participants on themselves, their fathers, and paternal grandfathers, was used to delineate familial migration histories and to estimate the degree of patrilocality. Y-chromosomal single nucleotide polymorphisms (Y-SNPs) and short tandem repeat markers (Y-STRs) provided a molecular genetic perspective on male population composition and family names (surnames) served as cultural markers of ancestry.

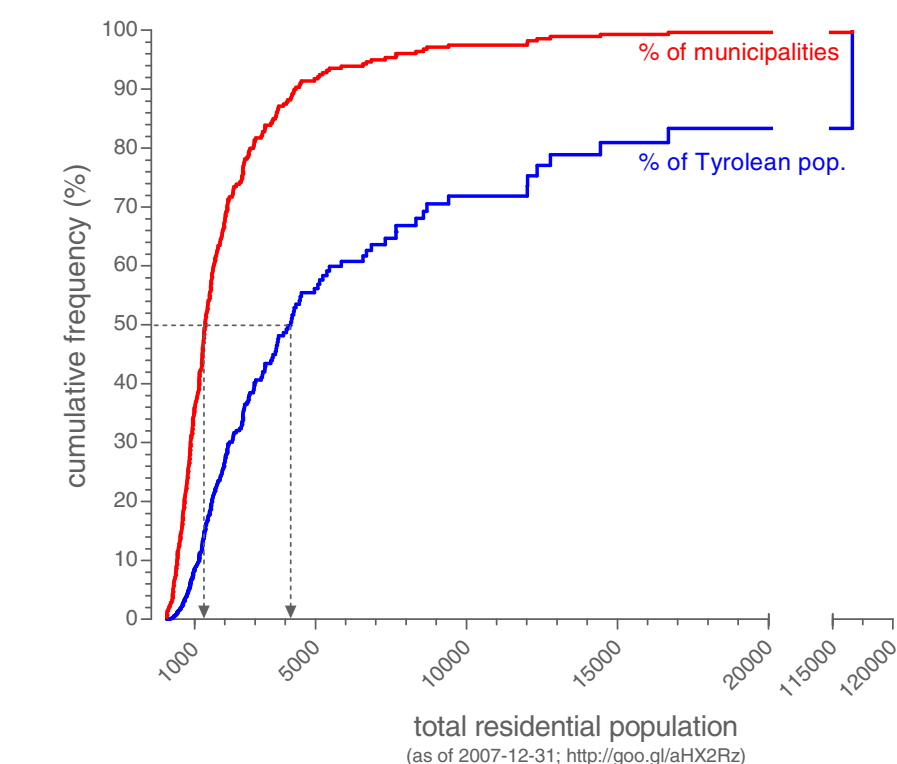
sampling



DNA samples of 1006 men from eight municipalities located in three NUTS3 regions were collected during routine blood drives of the Red Cross. These municipalities were chosen on basis of population and sample size, plus their differing degrees of urbanization. Genealogical data was utilized for filtering-out all but one close paternal relatives (up to first cousins) from inferred kinship groups. Here, matching Y-SNP haplogroup assignments served as a required plausibility criterion and the kin-filtered dataset ($n = 923$) was used in all downstream analyses.

Tyrolean municipality population sizes

A mix of differently sized settlements within a limited geographical range is characteristic for Tyrol. About 50% of its total population lives in communities comprising ≤ 4000 individuals, but the presence of more urbanized centers of settlement is typical as well.

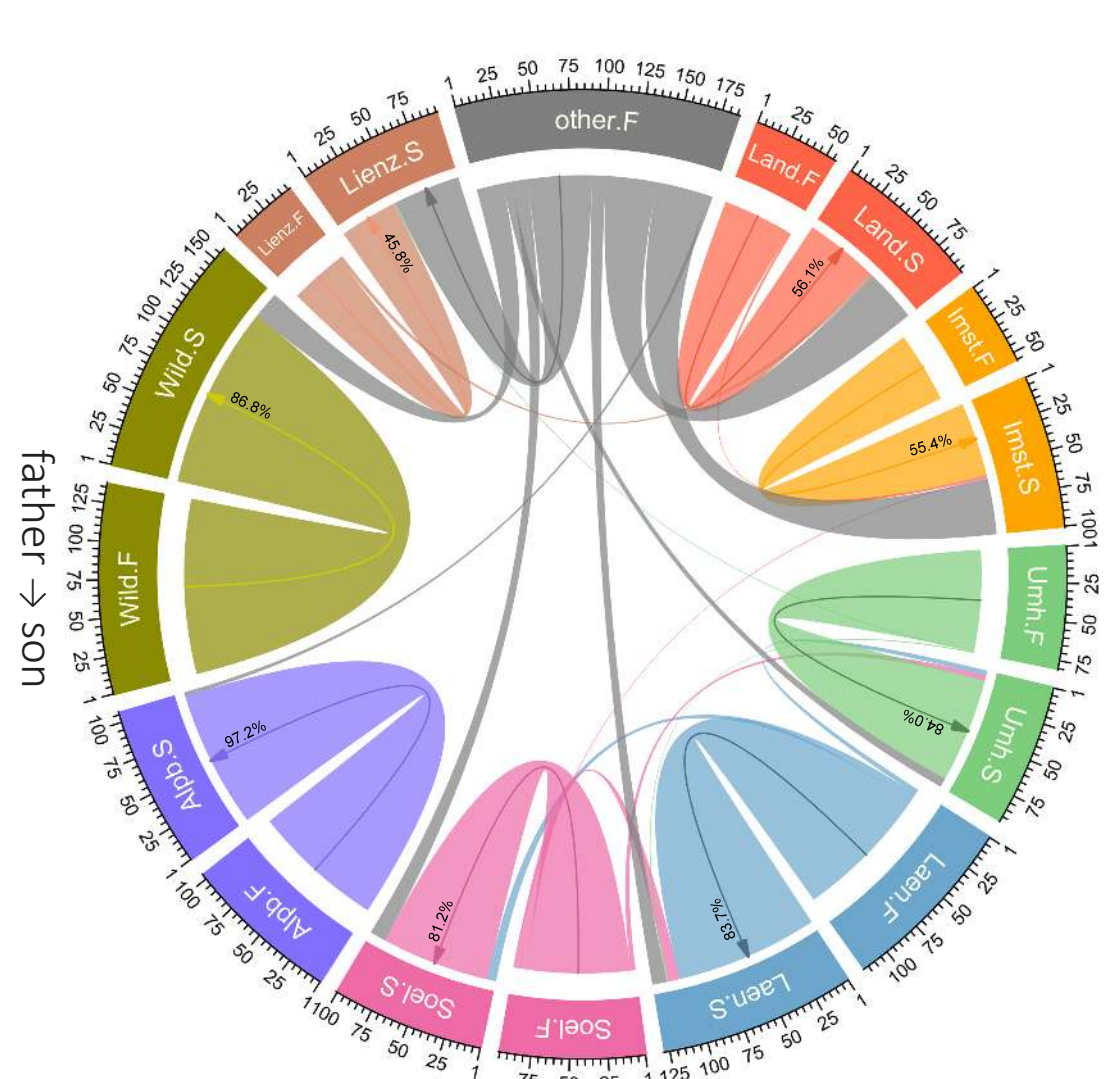
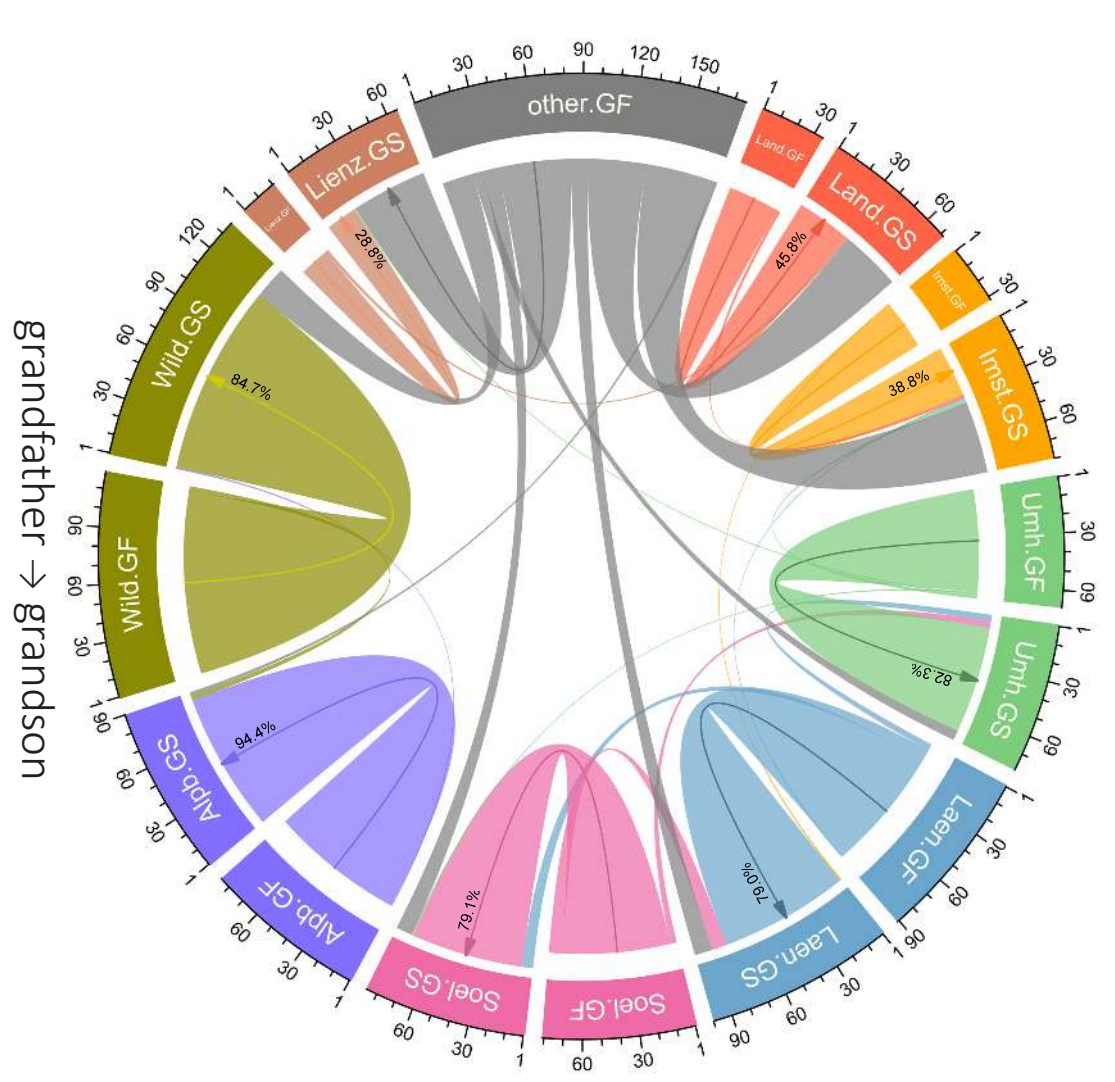


basic sampling statistics

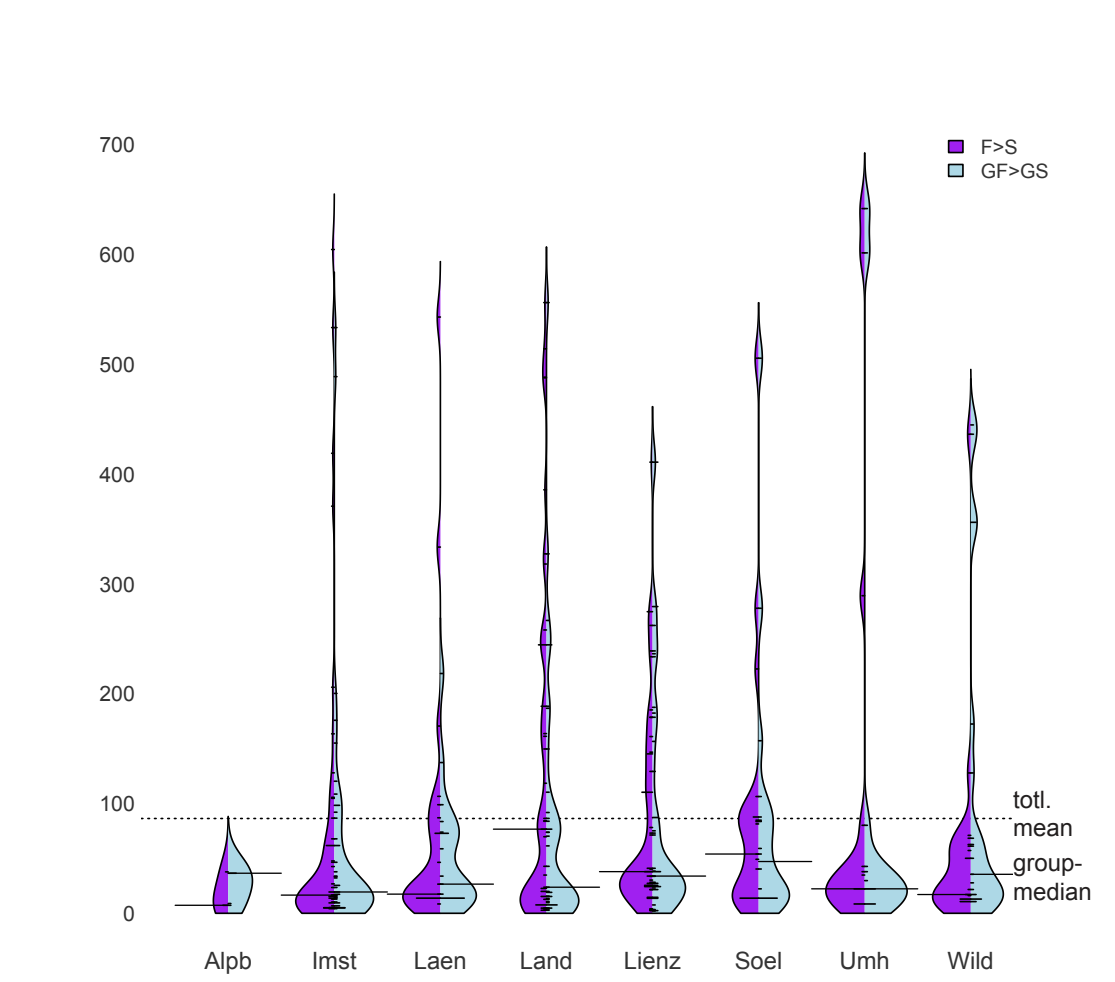
donor	municipality	NUTS3	region	total pop.	sample size	% of pop. filtering	% of pop. filtering
Imst	town	AT333	Osttirol	675,120	534	45.9	114
Landeck	town	AT334	Oberland	796,763	372	46.6	111
Längenfeld	town	AT334	Oberland	803,849	465	49.4	116
Lienz	town	AT334	Oberland	103,805	154	53.2	112
Soled	village	AT334	Oberland	1178,455	247	48.3	141
Umhausen	village	AT334	Oberland	1350,380	123	49.4	114
Wild	village	AT335	Unterland	508,424	213	50.1	179
Alpbach	village	AT335	Unterland	970,291	132	50.6	119
total				47,559	22,924	48.6	1006

patrilocality

The degree of patrilocality (percentages are given inside the ribbons illustrating the migrational movements) differed markedly between samples from small villages and towns. On basis of our data, male intergenerational migration among the studied municipalities appeared to be rare, even between directly neighbouring communities.

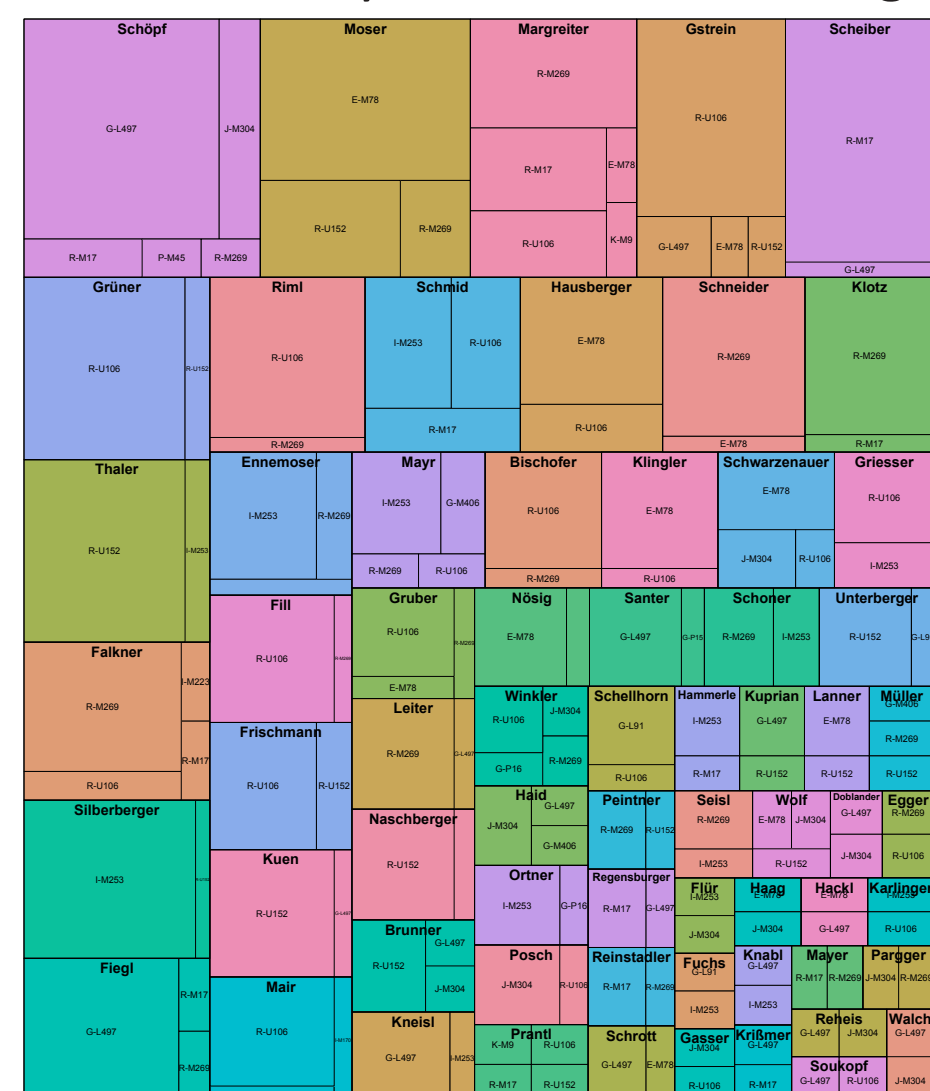


The typical intergenerational travelling distances were short (km, migrants only)

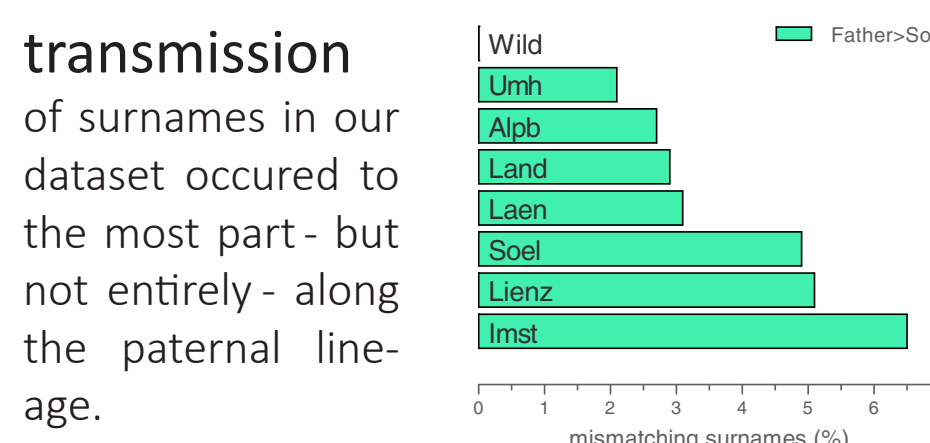


surnames

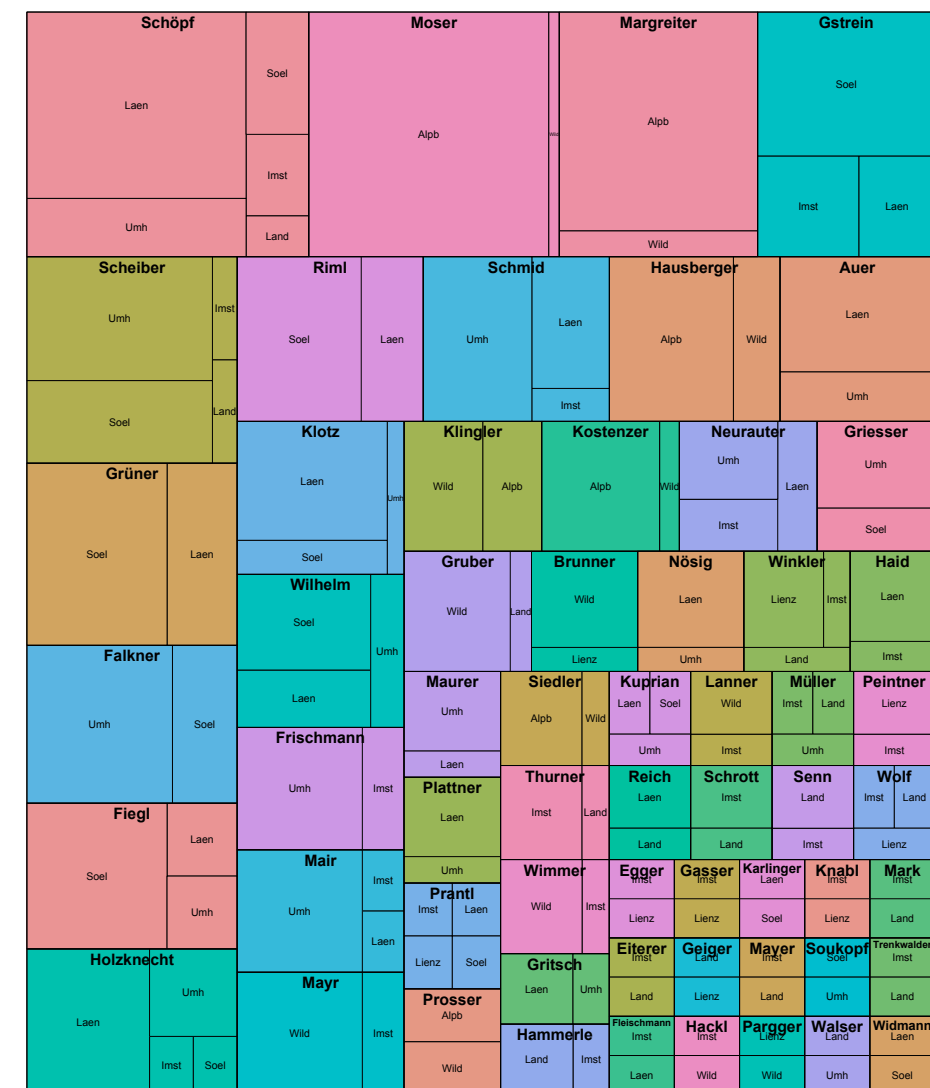
donor's family names and Y-SNP hgs



All of the non-unique family names in our dataset mapped at least to two different underlying Y-SNP haplogroups. This is likely to reflect multiple potential origins of these names, but also adoption of the mother's maiden name or false paternity need to be considered here.



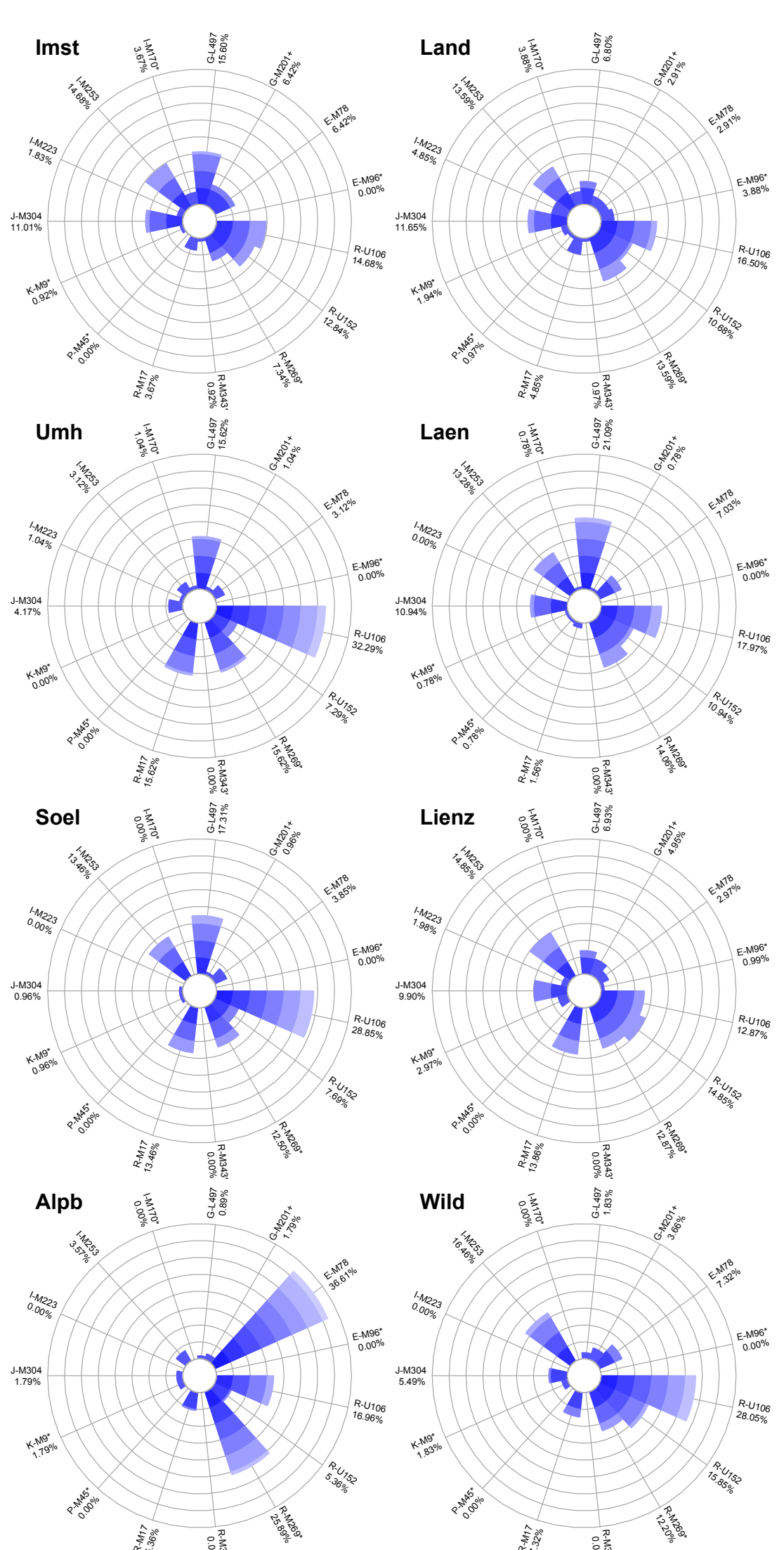
family names and municipalities



The majority of the non-unique family names in our dataset appeared to be region-typic, since most of the surname-sharing across municipality borders was observed within NUTS3 regions. Some of the names, however, seemed to be almost community-specific (e.g. Moser and Margreiter for Alpbach).

Y-SNPs

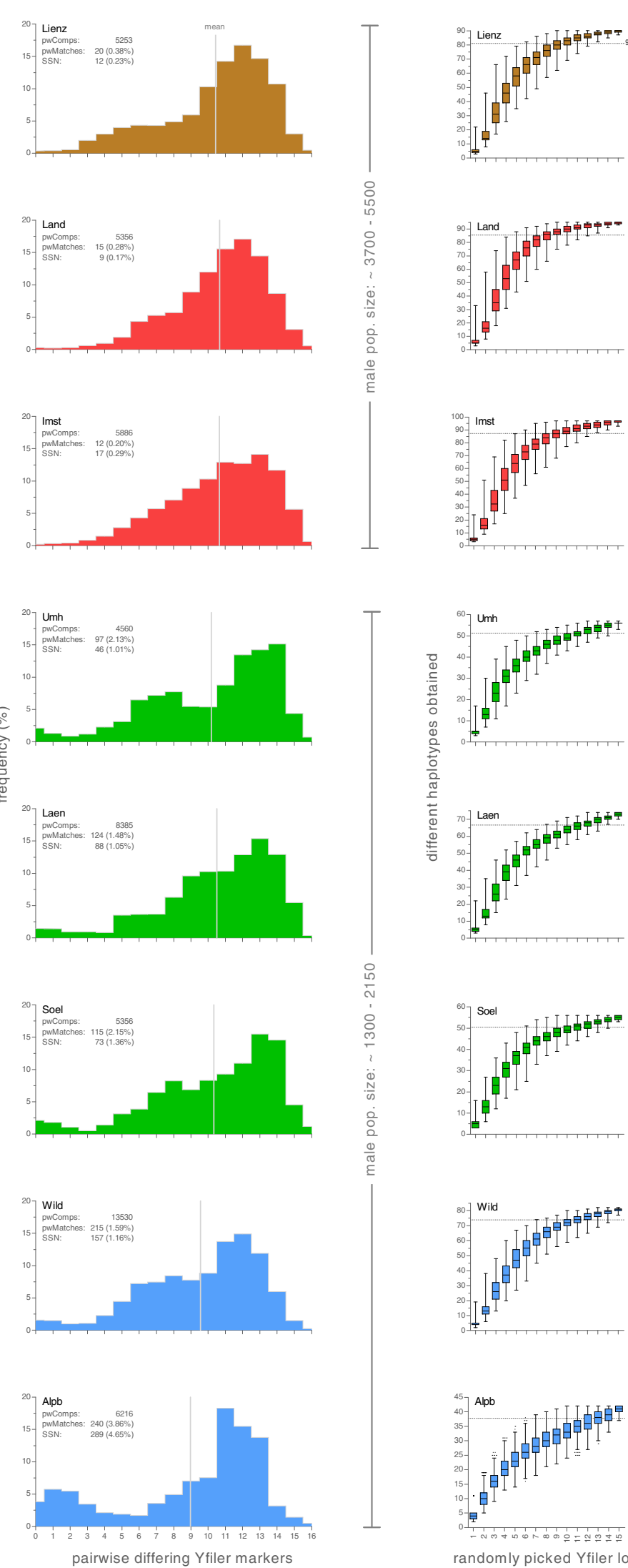
haplogroup composition



Y-chromosomal SNP haplogroup frequency spectra obtained in this study revealed differences among the NUTS3 regions addressed. For instance, in the Tiroler Oberland haplogroup G Y chromosomes were encountered at elevated frequencies and haplogroup E-M78 reached an impressive share of 36.6% in Alpb while being rather rare in the other communities.

Y-STRs

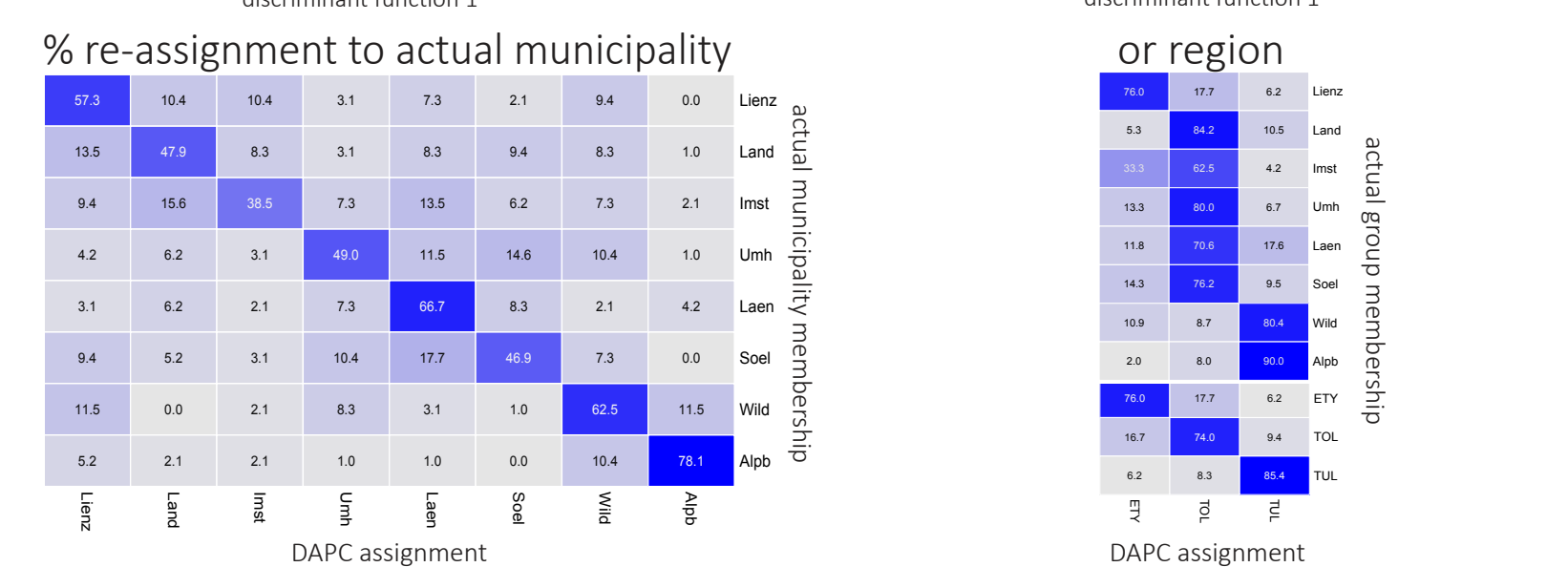
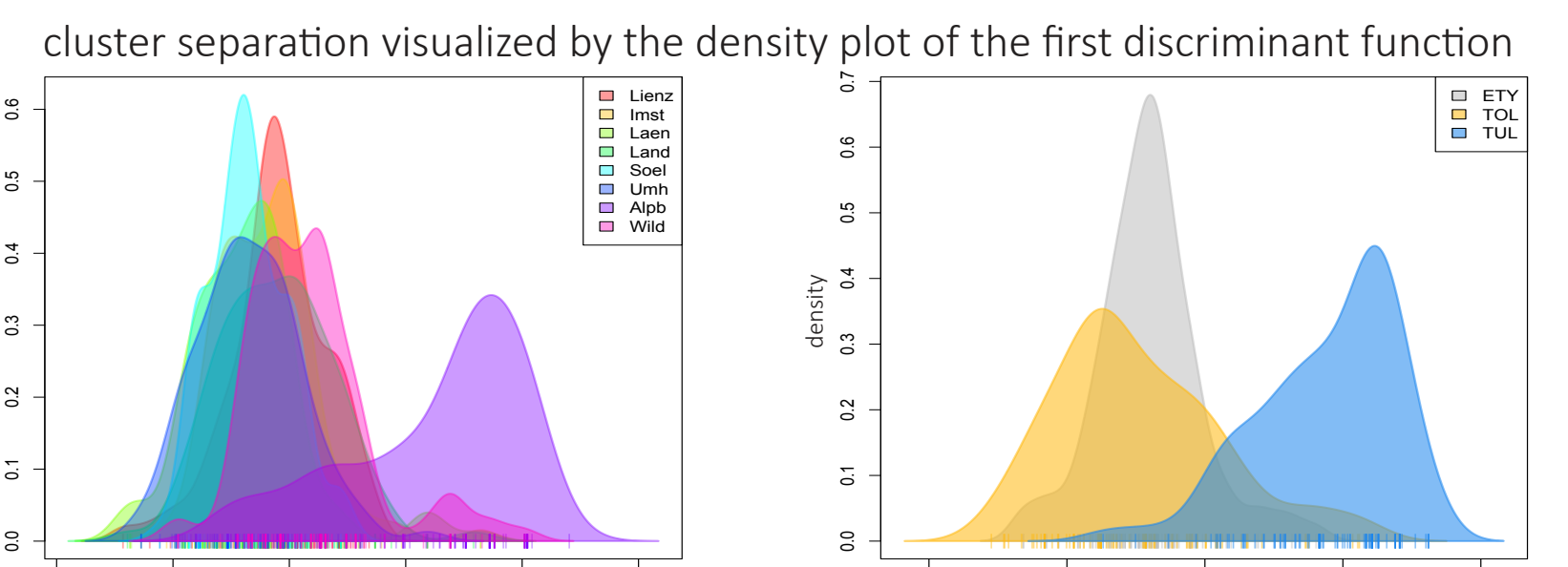
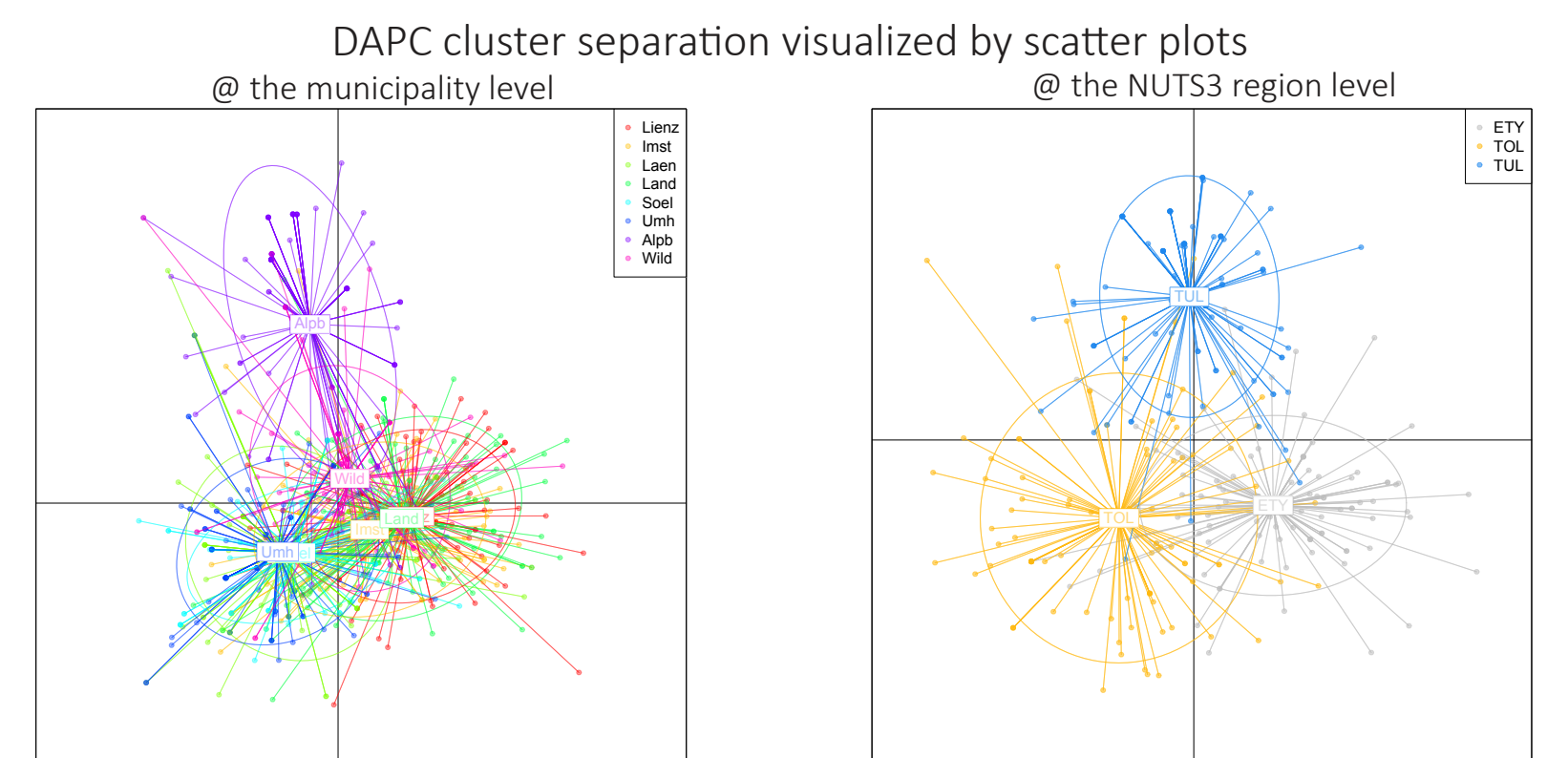
pairwise mismatch distribution spectra and genotype accumulation curves of Yfiler profiles



Despite kin-filtering, the small villages differed from the three towns by higher proportions of pairwise matching or similar Yfiler haplotypes, suggesting higher proportions of (cryptic) relatives in these settlements. This also became manifest in genotype accumulation curves not reaching the plateau within 16 loci. This observation points toward marker-wise under-sampling of the small communities.

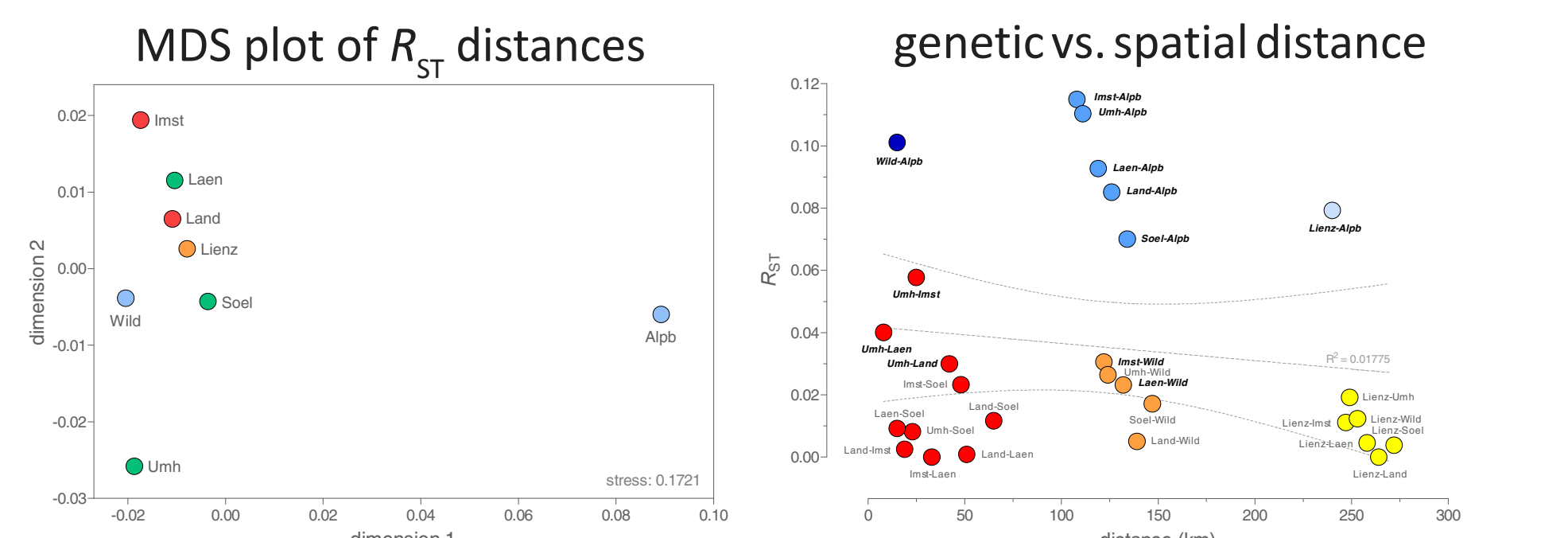
population structure - exploratory clustering

discriminant analysis of principal components (DAPC)



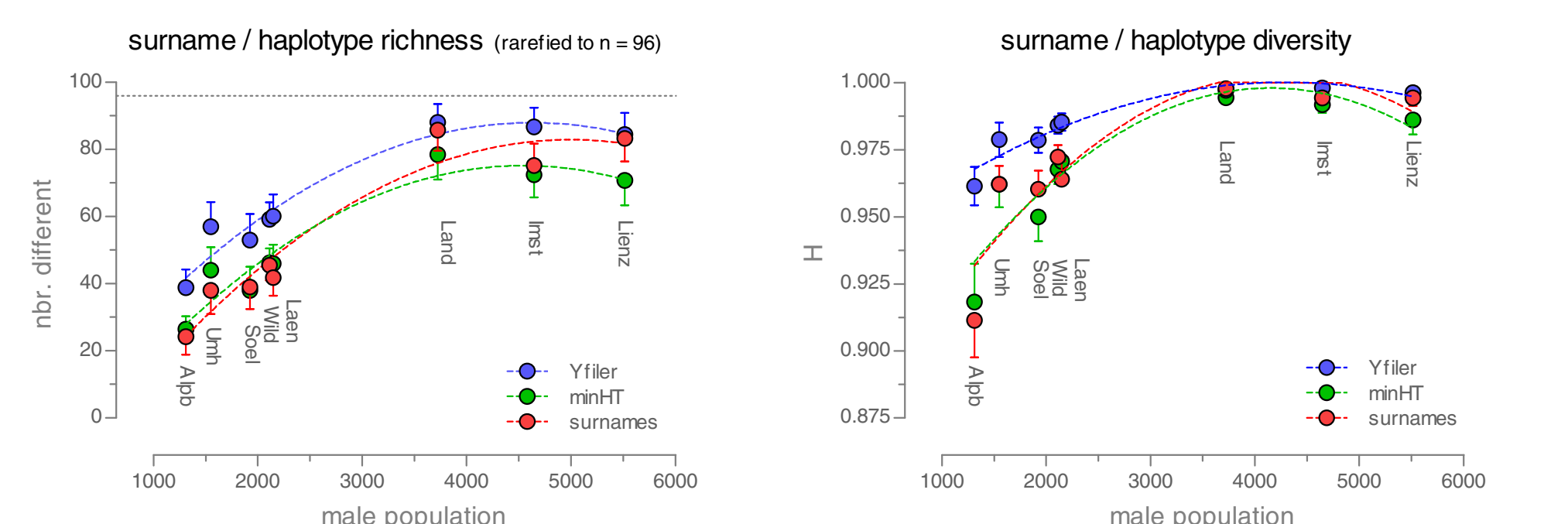
DAPC is a multivariate approach allowing for inferences regarding population structure without prior knowledge of the latter. For the Tyrolean sample DAPC provided evidence for population substructuring. Particularly for Alpb, Laen, and Wild re-assignment of individuals to their actual groups occurred at considerable proportions. A more clear-cut picture was obtained when performing the analyses at the resolution level of NUTS3 regions (ETY: Osttirol, TOL: Tiroler Oberland, TUL: Tiroler Unterland).

population structure - genetic distance

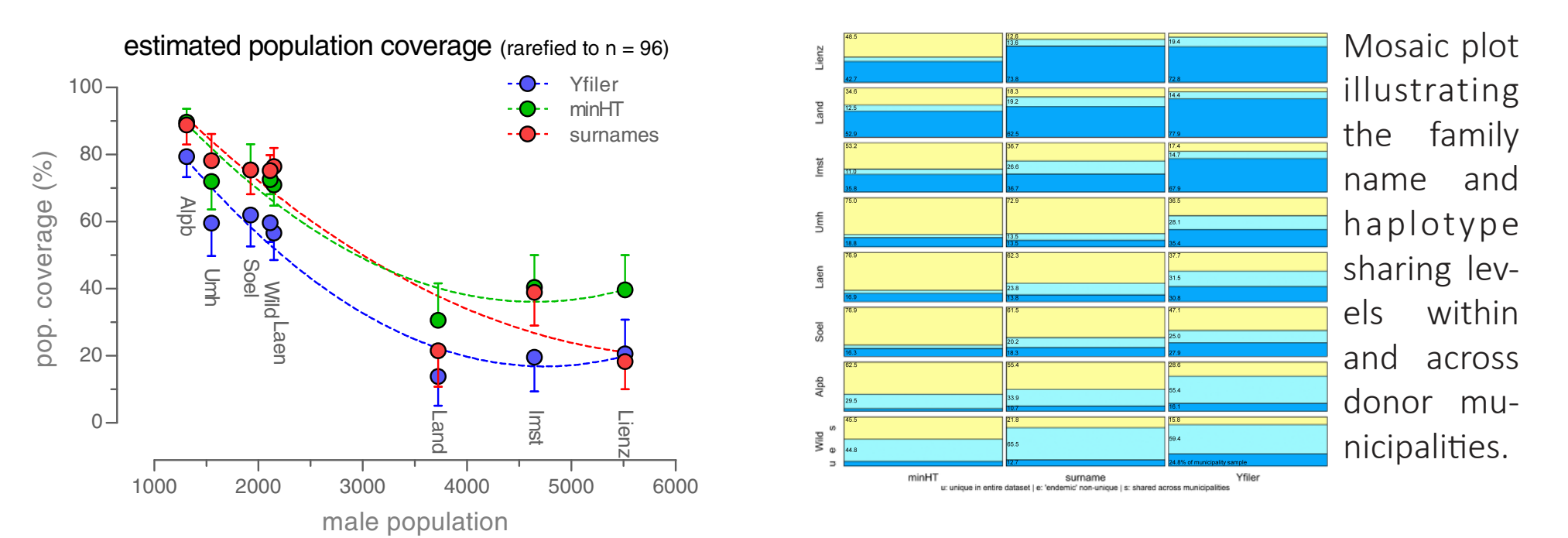


Analysis of R_{ST} distances corroborated the evidence for population substructuring obtained by DAPC. Overall, our results did not show significant correlation between spatial and genetic distances. However, there was negative correlation of these two parameters within sub-sets of our dataset (Alpb and Wild vs. TOL municipalities; Lienz vs. all the others, but also within the group of TOL municipalities; statistically significant R_{ST} values are indicated by italicised boldface municipality-pair designations).

Y-STRs and surnames - a comparison



Sample size standardization by random resampling allowed for direct population comparisons: **a)** in the small villages, but not in the three towns, surname- as well as minimal and Yfiler haplotype richness increased and the estimated population coverage decreased with increasing population size. **b)** the surnames showed a level of 'performance' similar to that of the minimal haplotype (minHT). **c)** inclusion of additional, informative Y-STR markers will be required to avoid under-sampling and to ameliorate the dependency of H on population size, as observed for the samples from small villages.



Mosaic plot illustrating the family name and haplotype sharing levels within and across donor municipalities.

Ethics Statement This study was approved by the "Ethics Commission of the Medical University of Innsbruck" and all participants gave written informed consent.

Note Part of the Lienz and haplogroup G data had been previously published: Niederstätter *et al.* PLoS ONE 7 (2012) e41885; Berger *et al.* FSIG 7 (2013) 624-631.

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