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# Comparison between Inbreeding Analyses Methodologies

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#### ABSTRACT

Surnames are widely used in inbreeding analysis, but the validity of results has often been questioned due to the failure to comply with the prerequisites of the method. Here we analyze inbreeding in Hallstatt (Austria) between the  $17^{h}$  and the  $19^{th}$  centuries both using genealogies and surnames. The high and significant correlation of the results obtained by both methods demonstrates the validity of the use of surnames in this kind of studies. On the other hand, the inbreeding values obtained ( $0.24 \times 10^{-3}$  in the genealogies analysis and  $2.66 \times 10^{-3}$  in the surnames analysis) are lower than those observed in Europe for this period and for this kind of population, demonstrating the falseness of the apparent isolation of Hallstatt's population. The temporal trend of inbreeding in both analyses does not follow the European general pattern, but shows a maximum in 1850 with a later decrease along the second half of the  $19^{th}$  century. This is probably due to the high migration rate that is implied by the construction of transport infrastructures around the 1870's.

Key words: Inbreeding, isonymy, genealogies, polyphiletism, migration, Hallstatt (Austria)

## Introduction

Surnames have been widely used as a simple method for calculating inbreeding since Crow and Mange established their methodology in 1965<sup>1</sup>. One of its main advantages is that it takes into account far inbreeding, as opposed to the values obtained from genealogies and dispensations. Many adaptations of this methodology and new methods based in surnames have been developed since<sup>2,3</sup>. However, critical voices emerged almost immediately<sup>4</sup>, especially in reference to the multiple origins of surnames and the overestimation of the inbreeding values that this might suppose. But more recently, some authors<sup>5-10</sup> asserted that results from the isonymy method, despite being sometimes overestimated, were informative enough in population genetics analysis. A good way to resolve this issue is to compare inbreeding obtained from isonymy with that obtained from other sources<sup>11,7</sup>.

The purpose of this work is twofold: first, to compare inbreeding values obtained with the isonymy method in

Hallstatt (Austria) with those directly obtained from the genealogies reconstructed from the demographic data, and second, to infer the degree of isolation of Hallstatt's population by way of its inbreeding levels. The availability of genealogical data allowed a time-wise analysis and the comparison of both methods so as to analyze whether surnames are a good source in inbreeding studies.

#### **Materials and Methods**

## **Population studied**

Hallstatt, one of the oldest settlements in Austria, is located at about 75 km south-east from Salzburg, in the province of Upper Austria, by Lake Hallstatt. Until the end of the 19th century it was *a priori* a rather isolated village, surrounded by mountains around 2000 m high, and it could only be reached walking through the mountains or sailing by boat across the lake. Communication with other populations improved with the construction of

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a road in 1875, which connected Hallstatt to Gosaumühle, and with the railway in 1877, allowing for more open population movement, especially between Hallstatt and the surrounding villages.

During the 18th and the 19th centuries, the population of Hallstatt ranged between 1500 and 2000 inhabitants, and, in contrast to other villages from the Salzkammergut region, it remained quite constant through these centuries. However, the number of inhabitants decreased all over the 20th century due to the high levels of emigration<sup>12</sup>. The lowest population level was achieved at the last census: in 2006 Hallstatt just had 923 inhabitants registered. The main economical activity since 1300 B.C. and during the analyzed period was salt mining, which attracted foreign workers to the supposedly »isolated« population.

People from Hallstatt were mainly Catholic until the 16th century. By that time, Lutheranism was introduced in Austria and during the Reformation it acquired many adherents among the miners and foresters of the Hallstatt region<sup>12</sup>. Afterwards, Hallstatt's population was split up into two communities, although Catholics have usually outnumbered Protestants<sup>13</sup>.

#### Sample

Data for the present study has been drawn from ecclesiastic sources and it derives from the records of the 13,211 baptisms, 3,042 marriages and 11,875 burials celebrated from 1602 to 1891 in the Catholic parish of Hallstatt. Until 1852 these records include both Catholic and Protestant events. Since that moment, there has been a complete split of both churches and the Protestant parish has its own books with its own records that have not been taken into account in the present work.

From all these records, a genealogical reconstruction has been done using software programs created *ad hoc*, obtaining 3,558 families that include 16,922 individuals.

## Data analyses

From the genealogies, the average coefficient of inbreeding ( $\alpha$ ) has been obtained following the Bernstein formula. With regards to the inbreeding from isonymy, as well as their two components, the random and the non-random ones, they have been obtained from the 3,042 registered marriages with the program ISO<sup>14</sup> following the Crow and Mange formula<sup>1</sup>.

To analyse the evolution along time of both inbreeding estimates and of isonymy method components we divided the study period in six sub periods of near 50 years each: 1602–1649, 1650–1699, 1700–1749, 1750–1799, 1800–1849 and 1850–1891.

Finally, to infer the relationship between both methods we obtained Pearson's correlation coefficient using the SPSS 12.0 software package.

## **Results and Discussion**

Table 1 shows the inbreeding values obtained both by isonymy and by genealogies, as well as the two components of inbreeding by isonymy. In both cases, general values observed are low for the geographical and temporal con $text^{15-19}$ , showing the existence of genetic flow to and from the population in spite of its apparent geographical isolation. Our results agree with Sjøvold<sup>20,21</sup>, who reported that in Hallstatt inbreeding was almost inexistent, since around 20% of population was from immigrant origin. The main reason of the low inbreeding levels is probably the great activity of the salt industry in this village, with a significant economical importance, which implies great immigration levels, with the subsequent population diversity and low endogamy. Most of these immigration processes were from surrounding areas<sup>20</sup>, implying the arrival of already present surnames to Hallstatt, and the presence of polyphyletism in the analyzed surnames.

One of the effects of this polyphyletism is an overestimation of inbreeding from isonymy, which always shows much higher values than those obtained from genealogies, and that are not attributable only to the far inbreeding that surnames take into account. This difference between pedigree and isonymy values has been seen in many other studies<sup>11,15,19</sup> and took Jorde<sup>22</sup> to propose that they be regarded as the lower and the upper boundaries, respectively, of the inbreeding estimate. Just in a

TABLE 1

NUMBER OF INDIVIDUALS ANALYZED BORN IN THIS PERIOD, NUMBER OF MARRIAGES REGISTERED, TOTAL (F), RANDOM (FR) AND NONRANDOM (FN) INBREEDING FROM ISONYMY AND  $\alpha$  IN HALLSTATT BETWEEN 1602 AND 1891

Periods	N of individuals	N of marriages	F (× 10 <sup>3</sup> )	$\mathrm{F_r}~(\!\!\!\times~10^3\!)$	F <sub>n</sub> (× 10 <sup>3</sup> )	α (× 10 <sup>3</sup> )
1602–1649	2257	373	1.41	1.87	-0.46	0
1650 - 1699	3046	480	0.01	2.30	-2.33	0
1700 - 1749	3767	618	2.03	2.50	-0.47	0.02
1750 - 1799	3202	638	3.13	3.39	-0.26	0.23
1800 - 1850	3348	612	5.75	3.44	2.32	0.93
1851 - 1891	1302	321	2.34	2.27	0.07	0.14
All periods	16922	3042	2.66	2.16	0.50	0.24

similar analysis, Gagnon and Toupance<sup>7</sup> find not only the same trend but also similar values of inbreeding obtained from genealogies and from isonymy in the early Québec population. However, the good fit these authors find is due to two biases that compensate each other: polyphyletism and the unbalanced sex ratio of the pioneer immigrants. Whereas in our study it appears that the sex ratio of the first generations analyzed was balanced and we could observe just the increasing of inbreeding due to polyphyletism.

Another possible reason for the low values of inbreeding is the considerably high percentage of illegitimate children in the region<sup>12</sup>. The levels of illegitimate births were between 5 and 10% at the 18th century and higher than 10% in the 19th century. The highest values were achieved by 1850, when the percentage of illegitimate born children in Hallstatt was as much as 30%. Children born in this situation never appear as a descendent of a consanguineous relationship, so the inbreeding values could be underestimated.

Analyzing temporal evolution of inbreeding from isonymy, it is apparent an increase since the second half of the 17<sup>th</sup> Century, with a maximum in the middle of the 19<sup>th</sup> Century. This is the same tendency of the nonrandom component, so the increase of inbreeding in most of the analyzed years was related to a tendency in the population to favor this kind of marriages.

Most of the inbreeding analyses in European populations show a general pattern for inbreeding, with an increase along the second half of the 19th century and the beginning of the 20<sup>th</sup>, related with an increase of the nonrandom component, and a later decrease<sup>16,18</sup>. This trend is usually considered to be related to social and economical factors. Hallstatt's inbreeding does not follow this trend, but rather predates it and shows the highest values in the first half of the 19th century and a decrease in the second half. This earlier decrease of inbreeding coincides with the construction of the road and the railway that connected the village with other populations faster and easier than previously. These infrastructures allowed first an increase of the arrival of workers from the outside of the region (including an important arrival of Italian men) who took part on their construction, and also an increased possibility of emigration for Hallstatt's inhabitants, as the reduction in the number of births and marriages registered in

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In the analysis of the trend of the inbreeding obtained from genealogies, the former periods show values of 0, due to the fact that the genealogical database cannot account for the remote links of kinship in the first periods. But since the 18<sup>th</sup> century a great similarity with the trend observed from isonymycal analysis can be observed: an increase until 1850 and a later decrease. To test the significance of this similarity between the two methods, a Pearson's correlation test was performed. The result shows a positive, strong and statistically significant correlation between both inbreeding statistics ( $\mathbf{r} = 0.923$ ; p<0.01). This correlation is also high and significant ( $\mathbf{r} = 0.995$ ; p<0.01) when only the four last periods are taken into account (the former periods, with an inbreeding of 0, are ignored).

## Conclusions

The high correlation obtained between the values of inbreeding resulting from isonymy analysis and those calculated from genealogies study, confirm the validity of surnames analyzing consanguinity trends in time. Therefore, surnames are confirmed as a good tool for the analysis of the evolution of inbreeding, and the results obtained with this methodology have to be considered as consistent as those obtained from dispensations or genealogies.

However, our results show a clear overestimation of the inbreeding values obtained with the surnames' analysis, mainly due to the effect of the polyphyletism. This effect is probably the most difficult aspect to control in the analysis of inbreeding from the surnames, especially in populations like Hallstatt, with short-distance migration processes. Thus, surnames have to be used carefully in inbreeding analysis, taking into account the microevolutive processes that the population has suffered and knowing the limitations of the method.

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#### USPOREDBA METODOLOGIJA ANALIZE SROĐIVANJA

## SAŽETAK

Prezimena se naširoko koriste u analizama srođivanja, ali valjanost rezultata često je upitan zbog nepoštivanja preduvjeta metode. Ovdje ćemo analizirati srođivanje u Hallstattu (Austrija) između 17. i 19. stoljeća pomoću rodoslovlja i prezimena. Visoka i značajna korelacija rezultata dobivenih pomoću obje metode pokazuje valjanost uporabe prezimena u ovoj vrsti istraživanja. S druge strane, dobivene vrijednosti srođivanja ( $0,24 \times 10^{-3}$  u analizi genealogije i  $2.66 \times 10^{-3}$  u analizi prezimena) su niže od onih u Europi u tom razdoblju i za ovu vrstu populacije, pokazujući lažnost prividne izolacije stanovništva Hallstatta. Vremenski trend srođivanja u obje analize ne slijedi europski opći uzorak, ali pokazuje svoj maksimum u 1850. godini, a trend pokazuje tendenciju pada kroz drugu polovicu 19. stoljeća. To je vjerojatno zbog visoke stope migracije koji podrazumijeva izgradnjom prometne infrastrukture oko 1870-ih godina.