

# Conservation in the 21<sup>st</sup> century, technological advances and their challenges

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Prof Nikica Šprem, University of Zagreb, Croatia Prof Elena Bužan, University of Primorska, Slovenia and their teams



DNA as evidence of distribution and vitality of endangered Balkan chamois – BalkCham IP-2016-06-5751

![](_page_1_Picture_4.jpeg)

Prof Massimo Scandura University of Sassari, Italy

![](_page_1_Picture_6.jpeg)

Dr Alexander Kopatz NINA, Norway

![](_page_2_Figure_0.jpeg)

### **Genetic** markers in Conservation

![](_page_3_Figure_1.jpeg)

Fig. 3. Use of different genetic markers across the most studied taxonomic orders. Note: cpDNA used in animal groups is from diet studies.

![](_page_4_Figure_0.jpeg)

Number of studies

Pérez-Espona et al. (2017) Biol. Cons.

#### Conservation&Management 2017 2016 2015 2014 2013 2011 2010 2009 -Microsatellite —mtDNA —SNP

![](_page_6_Picture_0.jpeg)

![](_page_7_Picture_0.jpeg)

### mtDNA

![](_page_7_Figure_2.jpeg)

Vilaça et al. (2014) J. of Biogeography – D-loop

Rodriguez et al. (2009) J. of Heredity - cytb

![](_page_8_Figure_0.jpeg)

### **mtDNA**

![](_page_8_Figure_2.jpeg)

![](_page_9_Picture_0.jpeg)

### **mtDNA**

![](_page_9_Figure_2.jpeg)

Hassanin et al. (2009) J. Mol. Evol.

![](_page_10_Picture_0.jpeg)

# mtDNA iDNA - eDNA

![](_page_10_Picture_2.jpeg)

![](_page_10_Picture_3.jpeg)

![](_page_10_Picture_4.jpeg)

# i/eDNA

#### Pros

- Low DNA quality
- Detectability of rare or elusive species
- With the right combination of primers can help screen the biodiversity in an area or the diet of the target species

#### Cons

- Careful screening of results needed
- Rely on availability of reference sequences (soon to improve)
- Bioinformatic skills and computational facilities needed
- High initial costs

# Applications

- Kinship analysis paternity testing
- Forensic identification
- Marker assisted breeding
- Population genetics
- Evolution
- Local adaptation
- Disease associated stu
- Analyses of specific tr

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# STR

#### Pros

- Biparentally inherited
- Codominant
- Neutral
- Individual specific
- Hybridization/migration
- High recombination
- Highly informative many alleles
- Recent demographic events
- Cheap
- Low DNA quality
- Availability of historical databases
- Availability of standard marker sets for several species

#### Cons

- Difficult to have comparable results across labs
- Not all STR are equally informative
- Complex mutation mechanism
- Labour intensive (multiplex help)
- Price will not decrease

# **SNPs**

Pros

- Biparentally inherited
- Individual specific
- Codominant
- Abundant
- Low to high recombination
- Present in both coding and neutral regions
- Automation
- Low genotyping error rates
- Comparable across labs
- Single locus/haplotypes
- Allow the identification of genes/regions under selection for specific traits without prior information

#### Cons

- Biallelic (higher numbers needed)
- Costly
- High quality DNA needed
- Some are still unmapped
- Ascertainment bias
- Computational power & bioinformatic skills

![](_page_15_Picture_0.jpeg)

#### The Sardinian wild boar is considered a dwarf form of the European wild boar

It was classified as a distinct subspecies

Sus scrofa meridionalis (Major 1883)

![](_page_15_Picture_4.jpeg)

confirmed by several morphometrical and genetic studies (De Beaux & Festa 1927, Groves 1981, Apollonio et al. 1988, Randi et al. 1989)

#### Main goal

Evaluating the genetic diversity of the Sardinian wild boar (WB) population with respect to three aspects:

DIFFERENTIATION

genetic differentiation of the Sardinian WB population from the continental WB populations and from the domestic pigs

Expectation: high levels of differentiation

INTROGRESSION

signs of genetic introgression from continental WB populations and from domestic pigs into the Sardinian WB

**Expectation:** high introgression (mainly from domestic stocks)

![](_page_16_Picture_8.jpeg)

genetic structure into subpopulations within the Sardinian WB population

**Expectation:** panmixia

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![](_page_18_Figure_0.jpeg)

![](_page_19_Figure_0.jpeg)

C2

All datasets consistently represent the Sardinian WB as a separate

![](_page_20_Figure_0.jpeg)

#### DIFFERENTIATION

![](_page_21_Figure_1.jpeg)

#### **INTROGRESSION**

![](_page_22_Figure_1.jpeg)

- Loss of resolution
- Overestimation of introgression

![](_page_22_Picture_4.jpeg)

![](_page_22_Picture_5.jpeg)

- Similar resolution
- Possible overestimation of introgression

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![](_page_22_Figure_9.jpeg)

![](_page_22_Figure_10.jpeg)

![](_page_23_Figure_0.jpeg)

 $r^2 = 0.525$ 

### **INTROGRESSION** 16 STRs 12k SNPs vs. 57% 0 •• • 13% . •

• Weak correlation between individual Q-values obtained with STRs and SNPs data

 $r^2 = 0.133$ 

![](_page_25_Figure_0.jpeg)

#### **POP STRUCTURE**

#### only Sardinian WB with Q<sub>Sar</sub>>0.9

200 SNPs

n=85

![](_page_26_Figure_4.jpeg)

Different inference of population structure

The selected panel of SNPs was likely biased towards interpopulation divergence and poorly informative about inner population structure

#### To summarise

#### DIFFERENTIATION

• All datasets detect a **high level of differentiation** of the Sardinian population

• In all cases the cluster analysis identified a **private cluster** for Sardinian wild boars

#### **INTROGRESSION**

- STRs detect a higher level of introgression than SNPs (overestimation)
- A **loss of resolution** is generally associated with reducing the number of markers, while a **bias** may derive from selection of SNPs

#### **POP STRUCTURE**

• If introgressed individuals are removed, a similar **genetic structure** is detected by STRs and 12k SNPs, but not with the selected panel of 200 SNPs

![](_page_28_Picture_0.jpeg)

![](_page_28_Figure_1.jpeg)

![](_page_28_Picture_2.jpeg)

Fulgione et al. (2016) Evol. Appl.

![](_page_29_Figure_0.jpeg)

Fig. 3 Examples of differentially introgressed ancestry blocks, their multilocus ancestry genotypes and frequencies in the sampled localities for blocks NE1 (A) and GL1.1 (B). Per cent exogenous ancestry is provided in parentheses. A general schematic for ancestry assignments is shown in the legend in the lower right box. Each row is an individual, separated into their population of origin. Tick marks in the chromosome position maps indicate the location of ancestry informative SNPs; genes found within the regions are shown above the plots.

![](_page_30_Figure_0.jpeg)

Åkesson et al. (2016) Mol. Ecol.

## 19-36 STR

![](_page_31_Figure_1.jpeg)

Pairing: 70% - 38% Breeding: 65% - 26%

Beneficial effect of migrants

![](_page_31_Figure_4.jpeg)

Fig. 2 Geographic distribution of the original wolf pairs and their descendants. The location of wolf territories in Scandinavia 2008-2012, illustrating wolf pairs that can be traced back to the 14 original pairs (circles) that started to reproduce in 2008. Squares and triangles represent pairs with first- (F1) and second-generation (F2) descendants, respectively, whereas diamonds symbolize pairs with both one F1 and one F2 descendant. Edge-lined symbols represent breeding pairs. Dark blue represents the original pairs with an immigrant and pairs composed of immigrant descendants with partners descending from inbred pairs other than the 12 in the study. Light blue symbolizes pairs composed by an immigrant descendant and a descendant from one of the 12 inbred families included in the study (i.e. born 2008). Yellow, orange and red, respectively, represent the inbred native pairs with medium (f = 0.2-0.3), high (f = 0.3-0.4) and very high (f > 0.40) inbreeding levels and their descendants.

Åkesson et al. (2016) Mol. Ecol.

![](_page_32_Picture_0.jpeg)

97 (1977-2015)

6,701,147 SNP

Low diversity, even among immigrants and founders. One pair of immigrants was full-sib  $\rightarrow$  inbreeding based on pedigree is an underestimation

Confirmed presence of homozygous chromosomes and of possible purging selection

## Management

Sweden is committed to maintain naturally occurring species at population sizes large enough to permit harbouring genetic variation for longterm persistence (Swedish Environmental Objectives, Government Proposition 2009/10:155)

![](_page_33_Figure_2.jpeg)

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# Moving towards the future

1617 Estimation of gene flow into the Scandinavian wolverine population

Oddmund Kleven, Robert Ekblom, Göran Spong, Gerhardus M. J. Lansink, Jouni Aspi, Scott Creel, Ilpo Kojola, Alexander Kopatz, Anni Koskela, Laura Kvist, Navinder Singh, Jonas Kindberg, Hans Ellegren, Øystein Flagstad

![](_page_35_Picture_3.jpeg)

![](_page_36_Figure_0.jpeg)

![](_page_36_Figure_1.jpeg)

Figure 4. Map showing the geographic localities for all samples included in the microsatellite analyses. Colours according to the genetic clustering produced using BAPS (K=4). Dashed lines indicate borders between the three inferred subpopulations (central Scandinavia, northern Fennoscandia and southern Finland).

96 SNP 92 autosomal, 1 mtDNA, 3 Y-chromosome

![](_page_36_Figure_4.jpeg)

Figure 8. Results from a spatial PCA plotted onto a map (run in the R package 'adegenet'). EV stands for eigenvector, where positive values indicate global structure and negative values local structure. Global structures exhibit positive spatial autocorrelation while local structures display negative spatial autocorrelation. This approach (in contrast to the algorithms used by the software Structure) does not use assumptions of Hardy-Weinberg equilibrium to delineate clusters. As can be seen in the figure, both EV1 and EV2 show a weak global structure overall, with the exception of the southern Finnish population.

![](_page_37_Picture_0.jpeg)

The choice of a set of markers is a **critical** point to any analysis, together with study design. What's the scope?

#### Old but gold

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Genome-wide genotyping technologies are attractive because of the **huge amount of information** they can quickly and ever more cost-effectively provide

However, **marker reduction** is often required because of problems like lack of independence, data handling, software and hardware limitations

![](_page_37_Picture_6.jpeg)

Caution should be taken when using such data for population inferences, by critically checking possible **bias** that cross-species application or non-random selection can generate in the dataset

![](_page_37_Picture_8.jpeg)

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