

Update from GenoEx

B. van Doormaal

Speaker: Brian van Doormaal







GENESEX Parentage SNP Exchange (PSE)





Presented at the ICAR Annual Meeting Edinburgh, Scotland
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Background

- ICAR Board approved the introduction of two new services:
 - ➤ Genotype exchange services for parentage analysis, named **GenoEx-PSE**
 - Accreditation of DNA Data Interpretation Centres wishing to offer parentage analysis services (differing from accreditation of labs conducting DNA analysis using microsatellite- and/or SNP-based tests)
- Both were officially announced at the bi-annual meeting held in Chile in October 2016
- Presentation aim is to provide a status update on both services





GenoEx-PSE Overview

- Goal: To facilitate the international exchange/sharing of SNP genotypes for the sole purpose of parentage analysis
 - ➤ Parentage Verification
 - Parentage Discovery
 - Imputation of SNP associated with microsatellite STR (for subsequent parentage verification)
- Service targets organizations that have been responsible for parentage integrity (i.e.: breed societies) and/or those offering such services
- Currently includes dairy and beef cattle



GenoEx-PSE Signatories

- The original proposed Service Agreement Contract had three signatories:
- Service ICAR, as the legal subsidiary of ICAR responsible for its various services
- Interbull Centre, as the entity responsible for delivering the service and is legally part of the Swedish University of Agricultural Sciences (SLU)
- Service User, which is normally the Authorized Service User unless a Contracted Service Provider is designated



GenoEx-PSE Signatories

- Current proposal is a 2-party agreement involving the Interbull Centre and the Service User
- An agreement between ICAR and SLU has already existed for over 20 years
 - It describes the establishment and operations of the Interbull Centre
 - This agreement is currently being updated and will cover the provision of GenoEx-PSE services by the Interbull Centre on behalf of Service ICAR





SNP Groups for GenoEx-PSE

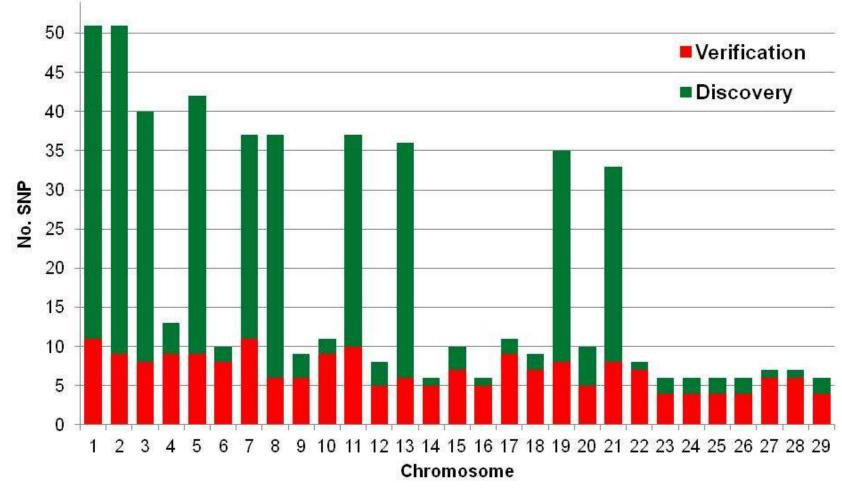
- Parentage Verification
 - ≥200 SNP recommended by ISAG
- Parentage Discovery
 - >200 ISAG SNP
 - ➤ Plus 354 other SNP (total 554) based on research in USA, Ireland, Australia
 - Reduced based on work in Canada
- Microsatellite Imputation
 - Additional 980 SNP deemed most useful for imputation to microsatellite SNP profiles to allow for parentage verification on that basis
 - ➤ Overall total of 1,534 SNP for possible exchange







Parentage SNP by Chromosome

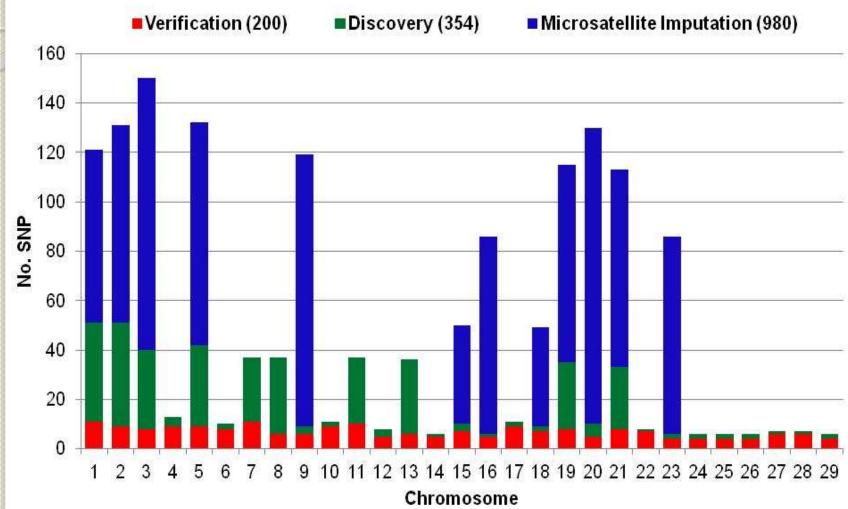








Parentage SNP by Chromosome





Data Exchange Rights & Options

- Only Service Users with a signed agreement can upload and download data
 - Each Service User has designated "population(s)" of animals under its responsibility, normally defined by country/region, breed(s), herdbook or not, etc...
- GenoEx-PSE will allow each Service User to download SNP genotypes in accordance to what they upload
 - ➤ Parentage Verification level = 200 SNP exchanged
 - ➤ Parentage Discovery level = 554 SNP exchanged





Service User Obligations

- Must upload all defined SNP genotypes routinely, at least 3X per year
- Must not use the SNP genotypes for purposes other than those related to parentage analysis
- Must not share data with third parties
- Must follow ICAR accreditation rules and regulations
 - ➤ Initially pass the process for ICAR Accreditation of DNA Data Interpretation Centres
 - Maintain the appropriate accreditation status





ICAR Accreditation for DNA Data Interpretation Centres

- Includes an initial application followed by a accreditation renewal every two years
- Initially to exclude Microsatellite Imputation
- Application form and fee submitted to ICAR
- Interbull Centre to provide the applicant with data file in standardized format for running through its parentage analysis system
- Applicant returns parentage analysis results to Interbull Centre with "pass" or "fail" status given to ICAR





ICAR Accreditation for DNA Data Interpretation Centres

- For any accreditation service, clear guidelines are required in order to identify "Pass"/"Fail"
- New ICAR DNA Working Group recently developed recommended "ICAR Guidelines for Parentage Verification and Parentage Discovery Based on SNP Genotypes"
 - ➤ Revised ISAG 2012 guidelines for Verification but require use of all 200 SNP not just "Core 100 SNP"
 - ➤ No international guidelines exist for Discovery
- Still pending:
 - > Feedback and support from ISAG
 - Formal ICAR Board approval



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Fees

- Interbull Steering Committee recommended an initial flat rate annual fee of 1 000 Euro per Service User for GenoEx-PSE
 - >To be reviewed over time
 - ➤ Difficult to predict the number of Service Users
 - ➤ Goal is for cost recovery for ongoing maintenance and staff time at Interbull Centre
- ICAR Accreditation for Interpretation Centres
 - ➤ICAR has set administration fee of 300 Euro per application (initial and bi-annual renewal)
 - Interbull Centre will wait to establish any fee for conducting data analysis on behalf of Service ICAR

Expected Timelines

- ICAR Accreditation is pre-requisite to being a GenoEx-PSE Service User
- Automated process for producing the data files for testing accreditation is being developed
 - Will need to have separate files for testing parentage verification versus discovery
- Official offering of accreditation service still a few months away
- Official GenoEx-PSE service soon after the next Interbull meetings in Estonia in late August







<u>GENESEX</u> Parentage SNP Exchange (PSE)



Thank You!