Update from GenoEx

B. van Doormaal

Speaker: Brian van Doormaal
Parentage SNP Exchange (PSE)

Brian Van Doormaal
Chair, ICAR DNA Working Group

Presented at the ICAR Annual Meeting
Edinburgh, Scotland
15 June 2017
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
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

- Verification (200)
- Discovery (354)
- Microsatellite Imputation (980)

Chromosome:

- No. SNP
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 an 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
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
Thank You!

Parentage SNP Exchange (PSE)