

# GenoEx-PSE Service: SNP Included for Exchange

October 2016

## Background

ICAR has decided to introduce GenoEx-PSE, which is a SNP genotype exchange service for the purposes of parentage analysis in dairy and beef cattle breeds. The Interbull Centre will act as the organization offering the service and maintaining the required database of genotypes and related information. Organizations interested in subscribing to this service must first sign a GenoEx-PSE Service Agreement and receive ICAR accreditation as a DNA Data Interpretation Centre.

## SNP Included for Exchange

The GenoEx-PSE service involves the exchange of three groups of SNP to be used for parentage analysis as follows:

- Group A: SNP recommended by the International Society for Animal Genetics (ISAG) required for conducting SNP-based parentage verification and animal identification verification.
- Group B: SNP in addition to Group A for conducting parentage discovery analysis.
- Group C: SNP required to impute an animal's microsatellite (STR) profile for the purpose of parentage verification.

ICAR appointed a GenoEx-PSE Expert Group to discuss and make recommendations associated with technical details of the GenoEx-PSE service. One of the key areas required for the service is to define the list of SNP to be included in Groups A, B and C for the purpose of exchange within the GenoEx-PSE service. Table 1 below provides the total number of SNP in each group and their distribution by chromosome. An Excel file containing the complete list of SNP, and their associated details, is available upon request from ICAR and/or the Interbull Centre.

As recommended by ISAG, SNP included for Group A are those defined by GenoEx-PSE for the purposes of verifying that the reported/recorded parents qualify as such and these same SNP may also be used for animal identification verification by examining if a submitted tissue sample can be excluded as originating from a particular animal. The GenoEx-PSE Service User may elect to conduct such parentage analysis using only the 100 "Core" SNP or the combination of 200 "Core" and "Extra" SNP recommended by ISAG. All Service Users must upload to the GenoEx-PSE database the maximum number of these 200 SNP that they have for each animal, which may, in some cases, only include the 100 "Core" SNP within Group A.

A total of 675 SNP have been defined by the GenoEx-PSE Expert Group for inclusion in Group B for use, in addition to Group A SNP, when conducting parentage discovery analysis. The current list of recommended SNP in Group B have been derived based on research conducted in United States, Ireland and Australia including a wide variety of dairy and beef cattle breeds.

The GenoEx-PSE Expert Group has also recommended a list of 980 SNP for inclusion as Group C, which were defined based on research in Ireland and United States examining how to best impute an animal's microsatellite (STR) profile for the purpose of parentage verification. Such analysis is beneficial for cases whereby a SNP genotype for the animal and both of its parents are not available but those that are missing do have a microsatellite (STR) profile available.

Chromosome	Group A		Group B	Group C	Total
	ISAG-Core	ISAG-Extra	Added for Discovery	Added for MS Imputation	
1	7	4	40	70	121
2	3	6	42	80	131
3	7	1	32	110	150
4	2	7	22	0	31
5	6	3	33	90	132
6	3	5	17	0	25
7	5	6	26	0	37
8	5	1	31	0	37
9	0	6	19	110	135
10	5	4	24	0	33
11	4	6	27	0	37
12	2	3	21	0	26
13	4	2	30	0	36
14	4	1	20	0	25
15	3	4	14	40	61
16	1	4	21	80	106
17	4	5	24	0	33
18	4	3	23	40	70
19	4	4	27	80	115
20	2	3	16	120	141
21	2	6	25	80	113
22	3	4	20	0	27
23	2	2	16	80	100
24	3	1	24	0	28
25	2	2	18	0	22
26	3	1	17	0	21
27	3	3	14	0	20
28	4	2	16	0	22
29	3	1	16	0	20
<b>Total</b>	<b>100</b>	<b>100</b>	<b>675</b>	<b>980</b>	<b>1,855</b>

### Use of SNP for Parentage Analysis by Service Users

As described above, and outlined in Table 1, the GenoEx-PSE service involves the exchange of SNP defined in three groups. At the level of the Service User, it is not necessarily required that all SNP within any specific group be used for the parentage analysis services provided for any given population of animals. In fact, research in Ireland (see reference below) has demonstrated that three SNP are problematic for parentage analysis services in the populations of animals examined due to clustering issues. As a consequence, Service Users have the right to exclude the use of any specific SNP exchanged within the GenoEx-PSE service for its parentage analysis services at the national level, under the condition that the Service User is successful in receiving the ICAR accreditation for that level of parentage analysis.

#### Reference:

MCCLURE, M. C., MCCARTHY, J., FLYNN, J., WELD, R., KEANE, M., O'CONNEL, K., MULLEN, M. P., WATERS, S. & KEARNEY, J. F. Year. SNP selection for nationwide parentage verification and identification in beef and dairy cattle. In: KOWALSKI, Z., PETRENY, N., BURKE, M., BUCEK, P., JOURNAUX, L., COFFEY, M., HUNLUN, C. & RADZIO, D., eds. Proceedings, International Committee for Animal Recording Technical Series, June 2015 2015 Krakow, Poland. ICAR, Via Savoia 78, 00198 Rome, Italy, 175-181.

## **Microsatellite Imputation**

For interested Service Users, the GenoEx-PSE service allows for the exchange of 980 additional SNP (Group C) that have been defined as useful for the imputation of SNP genotypes to microsatellite (STR) profiles. The process of microsatellite imputation requires a population of genotyped animals for carrying out the imputation process with an appropriate imputation software, which may prohibit some Service Users from carrying out this area of activity. In addition, the accuracy of the imputation step affects the accuracy of the subsequent step of parent verification. Service Users interested in this area of activity are encouraged to share research results in an effort to improve the overall accuracy of this component of parentage analysis services that may be of significant interest for countries transitioning from the use of microsatellite (STR) profiles to SNP genotypes for parentage verification.

## **Handling the Evolution of SNP Included Over Time**

As technology improves and SNP genotyping panels evolve over time, it is expected that the list of SNP defined as Groups A, B and C for the GenoEx-PSE service will be modified over time. This may occur by either (a) adding to SNP to the listing lists, or (b) by removing some SNP currently included and adding other new SNP. Under either scenario, it is clear that the total number and the defined set of SNP to be exchanged within the GenoEx-PSE service must not permit a level of imputation accuracy to higher density genotypes for the estimation of genomic predictions, which is strictly prohibited within the terms of the GenoEx-PSE Service Agreement.

The GenoEx-PSE Expert Group has made the following recommendations for handling changes to the list of SNP defined as Group A, B or C:

1. SNP included for Group A shall evolve in a manner that is consistent with recommendations made by ISAG. This approach will maximize the level of international harmonization for dairy and beef cattle breeds.
2. The GenoEx-PSE Expert Group shall review and consider all relevant research and be responsible for making recommendations regarding the list of SNP for Groups B and C within the GenoEx-PSE service. Under most circumstances, new SNP shall be added to these groups to ensure accurate parentage discovery across the various populations of dairy and beef cattle breeds for which the Service Users are providing parentage analysis services. In general, only SNP deemed to be problematic for conducting the desired parentage analysis shall be dropped from Groups B and/or C.