



IRISH CATTLE BREEDING FEDERATION

SNP Quality Control, Parentage, and Genetic Disease Risk Management in Ireland.



M. C. McClure, M. Mullen, S. M. Waters, F. Kearney, J. McClure, P. Flynn, and R. Weld



Outline

- 1) Quality control
- 2) Parentage
- 3) Genetic Disease Management

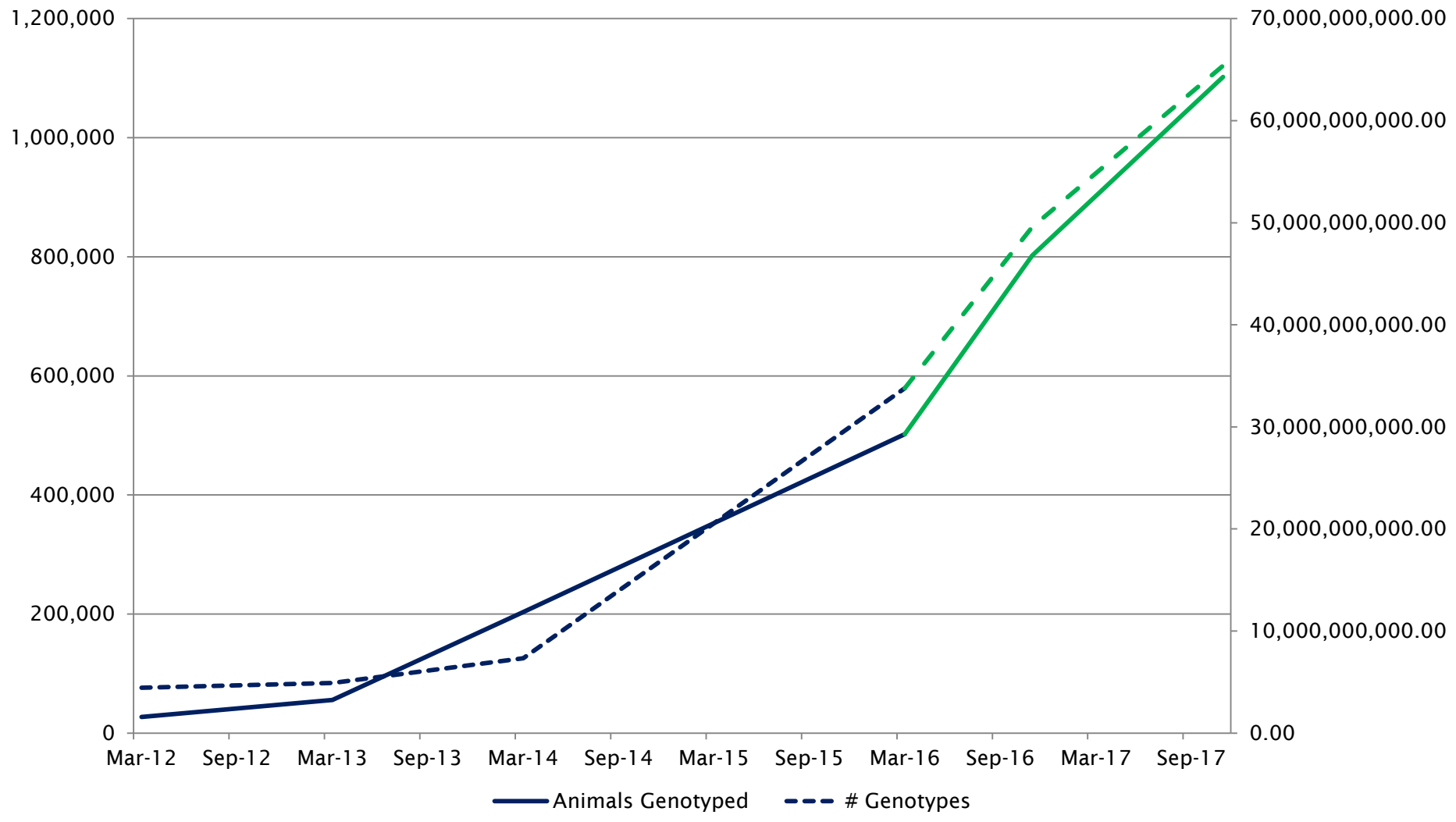
IRELAND



GENOTYPES



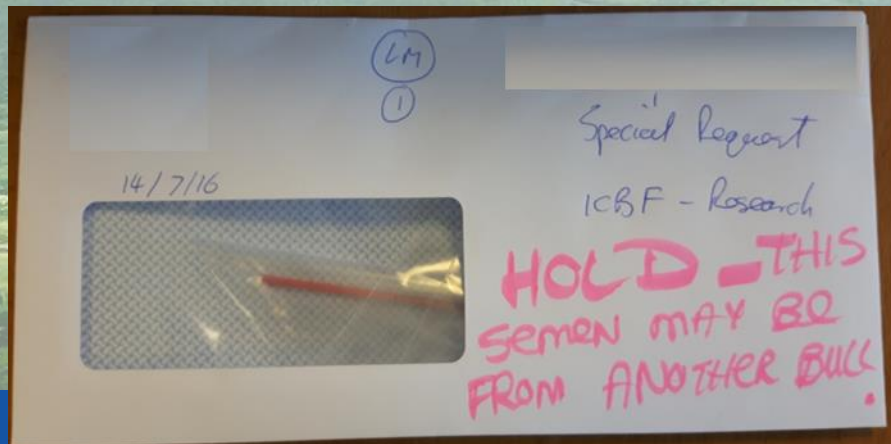
ICBF Genotype Growth

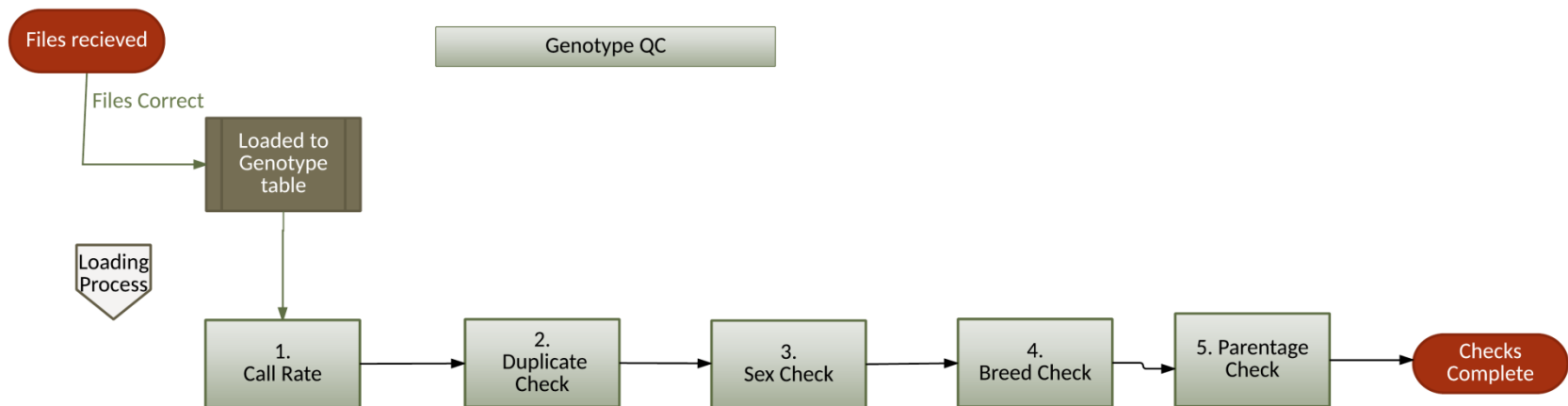


The “One in a Million Error” Will Occur

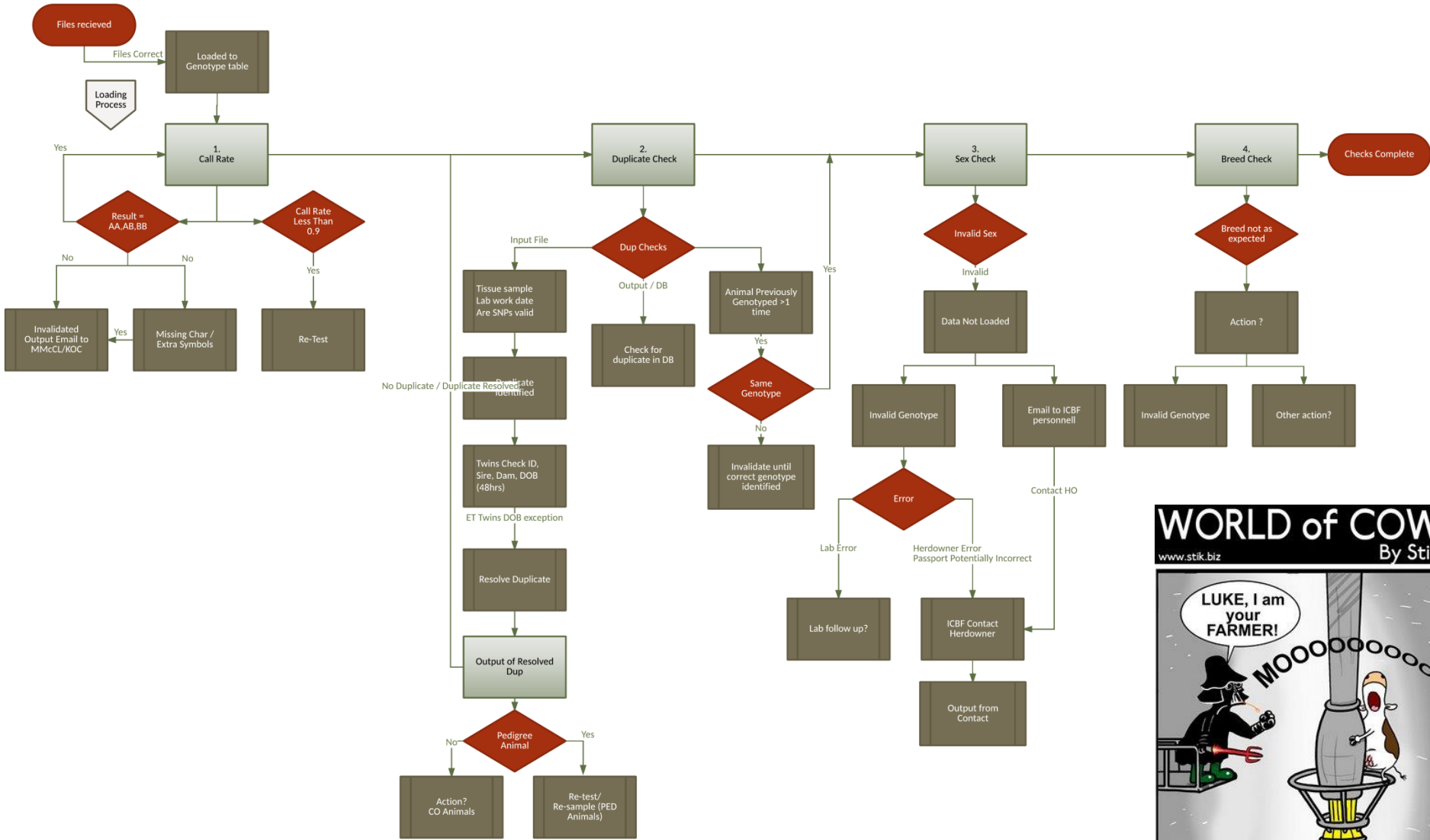
Did this DNA actually come from that Animal

- Farm Errors
- Lab Errors
- AI Errors





Genotype QC



What Does ICBF Do With a Genotype?



Male? Or Female?



Imputation
SNP + MS



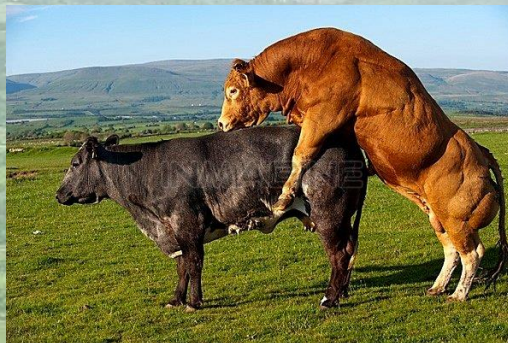
Breed Composition



?



Matchmaker



Carrier Status



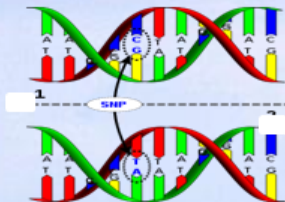
Brachyspina
Agerholm et al., 2006

Parentage

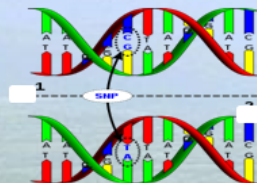
Every Sample

SNP validation
SNP prediction

800 SNP



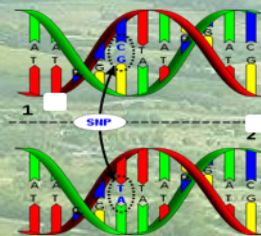
910 SNP



Pedigree animals

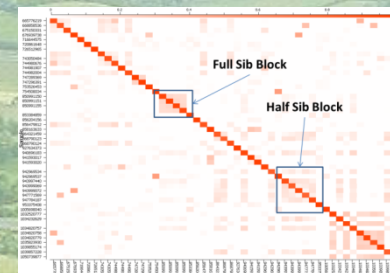
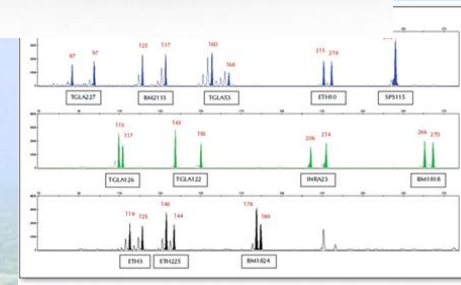
MS based validation

6900 SNP



As Needed to Suggest

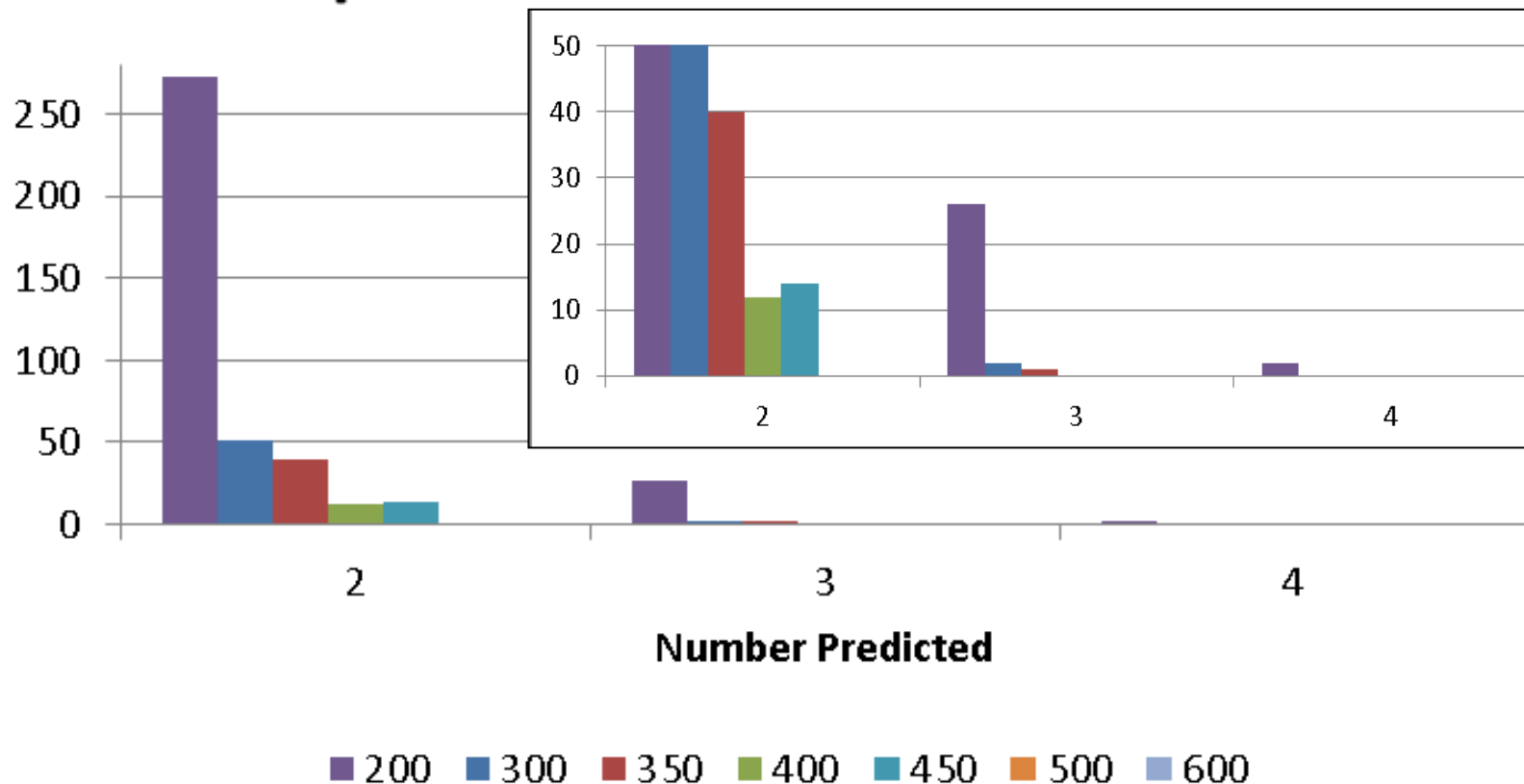
GRM analysis



Parentage SNP

ISAG100/200 not enough

>1 predicted Sires at X SNP levels

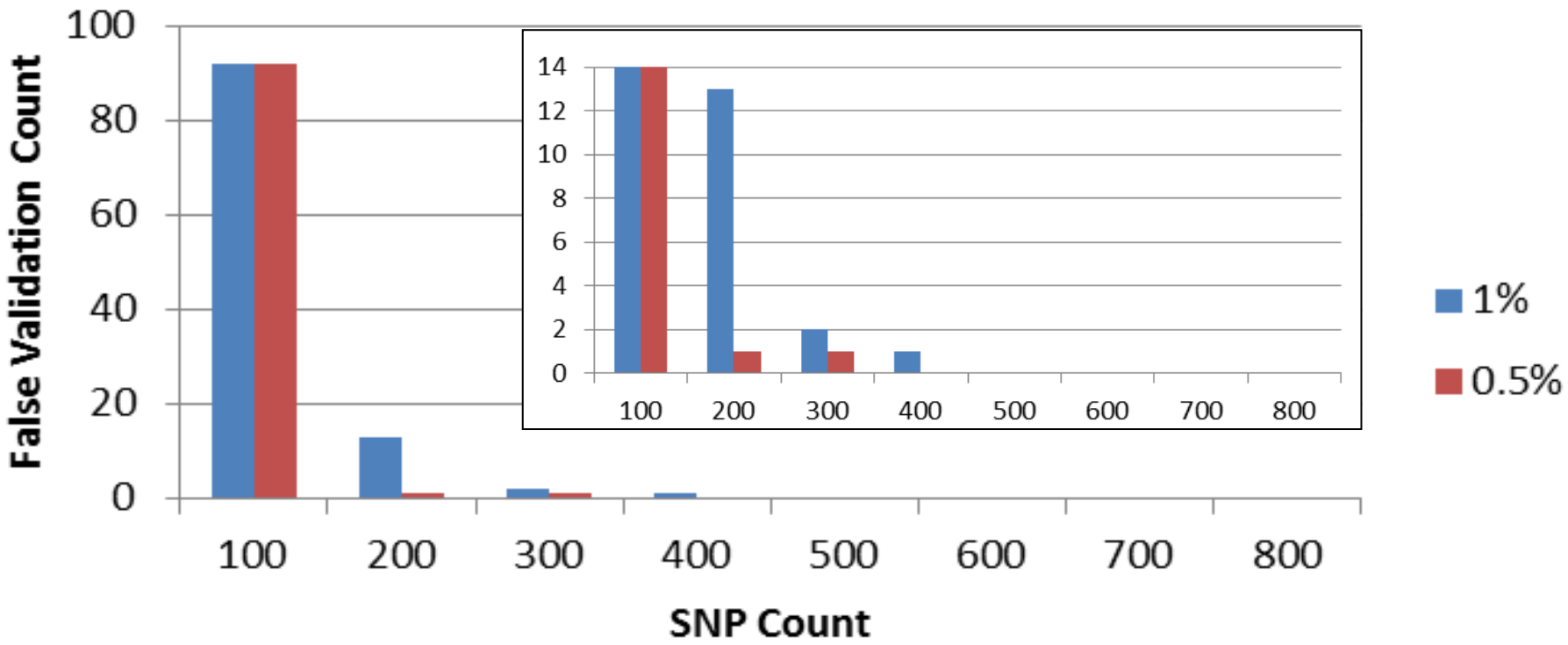


Parentage predicted on 9,000 animals against 60,000 reference, <1% misconcordance

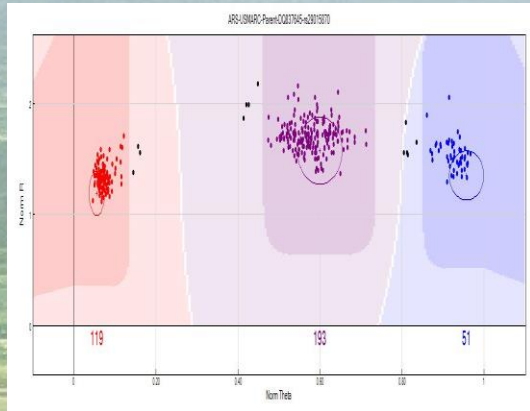
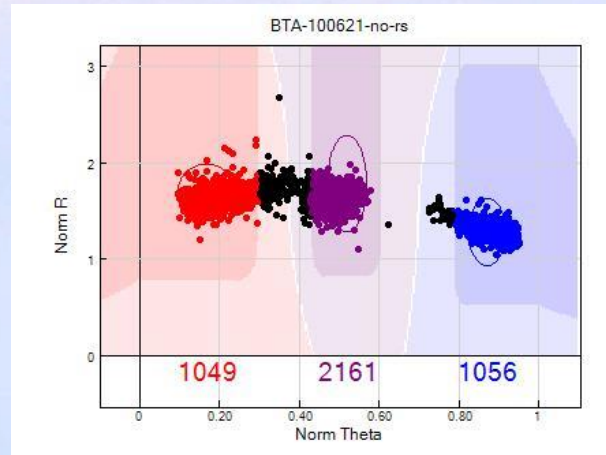
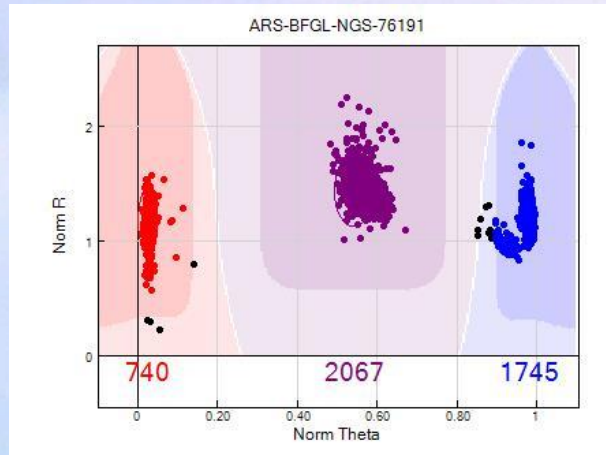
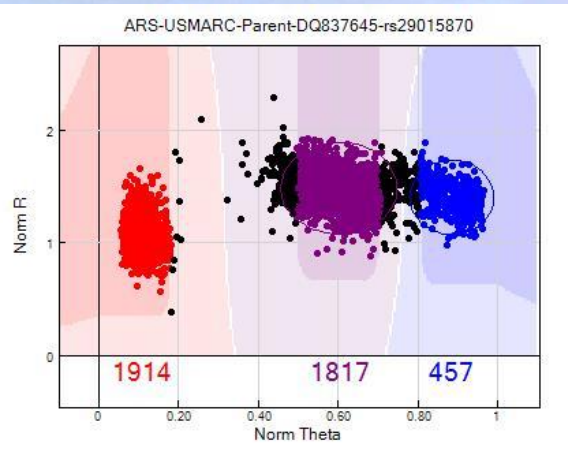


Can the **Listed Sire** wrongly validate with low SNP #?

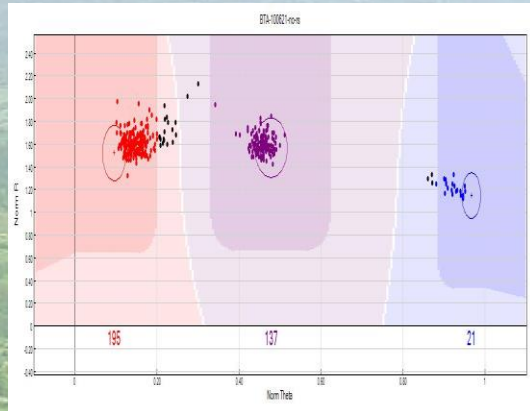
False Parentage Validations at Low SNP Levels.
26,857 Animals Whose Listed Sire Failed at 800 SNP



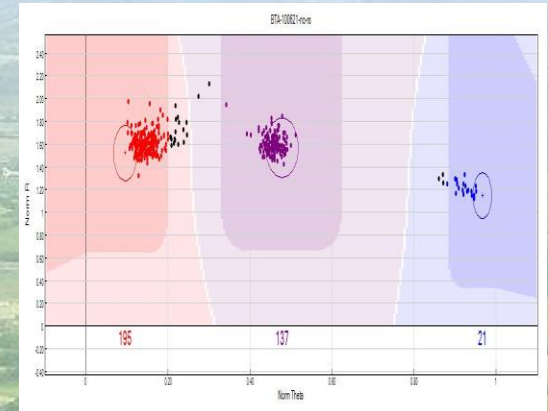
ISAG200 Panel SNP Cluster Issues



50k



LD



LD

What Does ICBF Do With a Genotype?



Male? Or Female?



Imputation
SNP + MS



Breed Composition



?



Angus



Limousin



Dexter



Saler

Matchmaker



Carrier Status



Brachyspina
Agerholm et al., 2006

Ireland Disease Testing IDB chip 213 Disease/Trait Probes

86 validated: 25 Lethal, 25 Unwanted,
13 Meat, 9 Beneficial, 9 Milk, 5 Color
82 awaiting sequence validation



Brachyspina
Agerholm et al., 2006

IDB SNP CHIP
INTERNATIONAL DAIRY & BEEF
SNP CHIP

Designed in association with the Irish Cattle Breeding Federation (ICBF), Teagasc, Weatherbys and USDA's Agricultural Research Service.

This custom chip is the very latest design catering for both Beef and Dairy. The chip consists of the illumina LD (7K) base content plus a further 10,000 (10K) SNPs carefully selected to ensure very high imputation accuracy to HD & to convert to Microsatellite data for parentage verification. This extra panel of SNPs provides the very latest dual product for both Beef & Dairy breeds.

Both the core and additional ISAG recommended SNP parentage panels are present on the chip.

The IDB also contains a comprehensive selection of genetic markers to screen for genetic disorders & major genes.

For more details Contact: Weatherbys Ireland DNA Laboratory

+353(0)45875521
jflynn@weatherbys.ie

CHIP CONTENTS FOR DISEASES & TRAITS

- Lethal recessives**
- 1 CVM* Complex Ventrinal malformation
 - 2 DUMPC
 - 3 Brachyspina*
 - 4 BLAD
- Congenital disorders**
- 1 Arthrogyposis (Curly Call)*
 - 2 Fawn Call Syndrome or Contractural Anachondactyly*
 - 3 Hypotichosis: PMelt*
 - 4 Hypotichosis in Belted Galloway, HEPHL1 SNP
 - 5 Hypotichosis: KRT17*
 - 6 Spadeleg- MCC21 gene- Simmental
 - 7 Spadeleg- SCUX gene- Brown Swiss
 - 8 Polledness
 - 9 Mule Foot
 - 10 Tibial Hemimelia (TH)*
 - 11 Black/Red Coat Color/Red Factor
 - 12 Red Recessive coat colour (Different to red factor)
 - 13 Silver Color Dilutor
 - 14 Dun Color
 - 15 RNF-11 (affects growth and stature)
 - 16 Osteopetrosis (Marble Bone Disease)
 - 17 Pink Eye (Infectious Bovine Keratoconjunctivitis)
 - 18 Photophosphorylase Fetaochalataze Gene (Photosensitization)
 - 19 SMA- Spinal muscular atrophy
 - 20 Beta Lactoglobulin
 - 21 Beta Mannosidosis
 - 22 Alpha Mannosidosis
 - 23 Citulinnemia
 - 24 CMDI: Congenital muscular dystonia I
 - 25 CMDII: Congenital muscular dystonia II
 - 26 Crooked Tail Syndrome*
 - 27 Factor XI
 - 28 Heterochromia Irises (White Eye)
 - 29 SDM- Spinal dysmyelination-SFAST Gene
 - 30 Idiopathic Epilepsy*
 - 31 Pulmonary Hypoplasia*
 - 32 Weaver
 - 33 Neuroopathic hydrocephalus* (water head syndrome)
- Major genes**
- 1 DGAT1
 - 2 MSTN (GDF8) Double Muzzling*
 - 3 A1A2 beta casein - *
 - 4 Fertility Haplopoos (beta1, Hbeta, Hbeta, beta1)
 - 5 Kappa Casein I
 - 6 Kappa Casein II
 - 7 ABCC2
 - 8 GH141 and GH2291 (Marbling growth rate)*
 - 9 IGF1-AF017143
 - 10 STAT1*
 - 11 STAT3*
 - 12 STAT5*
 - 13 Calpain (tenderloin) loci



Mulefoot
Duchesne et al., 2006



Double Muscle-Myostatin
ICBF.com

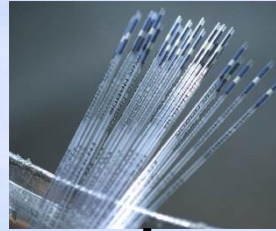
86 Validated Diseases/Traits on the IDBv3

Full Trait Name	Abbreviation	Reason
Infectious Bovine Keratoconjunctivitis	IBK	Beneficial
Polled Celtic	POLL_C	Beneficial
Polled Friesian 1909396D2	POLL_F_1909396D2	Beneficial
Polled Friesian 51D	POLL_F_51D	Beneficial
Polled Friesian C1655463T	POLL_F_C1655463T	Beneficial
Polled Friesian G1654405A	POLL_F_G1654405A	Beneficial
STAT1	STAT1	Beneficial
STAT3 19069	STAT3_19069	Beneficial
STAT3 25402	STAT3_25402	Beneficial
Black gene E, extension	MC1R_ED	Colour
Red factor e	MC1R_e	Colour
Dun Dexter	DUN	Colour
PMEL17 delTTC	PMEL17_delTTC	Colour
Silver Char Dilutor 1	PMEL17_64G_A	Colour
Alpha Mannosidosis 662	AM_662	Lethal
Alpha Mannosidosis 961	AM_961	Lethal
Arachnomelia Syndrome SUOX	SAA_SUOX	Lethal
Beta Mannosidosis	BM	Lethal
Brachyspina	BY	Lethal
Bulldog Dwarfism 1	BD1	Lethal
Cardiomyopathy and woolly haircoat syndrome	CWH	Lethal
Citrullinaemia	CT	Lethal
Complex Vertebral Malformation	CVM	Lethal
Congenital Muscular Dystonia 1	CMD1	Lethal
Congenital Muscular Dystonia 2	CMD2	Lethal
Deficiency of Uridine Monophosphate Synthase	DUMPS	Lethal
Holstein Haplotype 1	HH1	Lethal
Holstein Haplotype 3	HH3	Lethal
Holstein Haplotype 4	HH4	Lethal

Full Trait Name	Abbreviation	Reason
Idiopathic Epilepsy	IE	Lethal
Jersey Haplotype 1	JH1	Lethal
Maple Syrup Urine Disease Shorthorn	MSU_SH	Lethal
Maple Syrup Urine Disease Montbeliarde Haplotype 2	MSU_HER	Lethal
Montbeliarde Haplotype 2	MH2	Lethal
Neuronal ceroid lipofuscinosis	NCL	Lethal
Neuropathic hydrocephalus	NH	Lethal
Osteopetrosis	OS	Lethal
Paunch Calf Syndrome	PCS	Lethal
Tibial Hemimelia Improver	TH_Improver	Lethal
Calpain1 316	CAPN1_316	Meat
Calpain1 4751	CAPN1_4751	Meat
Calpain1 530	CAPN1_530	Meat
Calpastain 282	CAST_282	Meat
Calpastain 2870	CAST_2870	Meat
Calpastain 2959	CAST_2959	Meat
Myostatin C313Y	MYO_C313Y	Meat
Myostatin E226X	MYO_E226X	Meat
Myostatin E291X	MYO_E291X	Meat
Myostatin F94L	MYO_F94L	Meat
Myostatin nt821DEL11	MYO_nt821	Meat
Myostatin Q204X	MYO_Q204X	Meat
Myostatin S105C	MYO_S105C	Meat
ABCG2	ABCG2	Milk
AcylCoA:Diacylglycerol Acyltransferase	DGAT1	Milk
Casein Beta Allele 154G>A	CSN2_154G_A	Milk
Casein Beta Allele 245C>A	CSN2_245C_A	Milk
Casein Beta Allele 259C>G	CSN2_245C_A	Milk
Casein Kappa Allele 342T>C	CSN2_363C_A	Milk

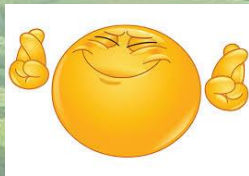
Full Trait Name	Abbreviation	Reason
Growth Hormone Receptor F279Y	GHR_F279Y	Milk
Growth Hormone 2141	GH_2141	Milk
Growth Hormone 2291	GH_2291	Milk
Axonopathy	AX	Unwanted
Bovine Leukocyte Adhesion Deficiency	BLAD	Unwanted
Bovine Progressive Degenerative Myeloencephalopathy	Weaver	Unwanted
Chediak Higashi syndrome	CHS	Unwanted
Congenital Myoclonus	CM	Unwanted
Crooked Tail Syndrome delAG	CTS_delAG	Unwanted
Developmental Duplication	DD	Unwanted
Dystrophic Epidermolysis Bullosa	DEB	Unwanted
Factor XI deficiency Wagyu	FXI_WA	Unwanted
Hypotrichosis KRT71	HY_KRT71	Unwanted
Mulefoot G1199S	MF_G1199S	Unwanted
Mulefoot G907R	MF_G907R	Unwanted
Mulefoot NG1621KC	MF_NG1621KC	Unwanted
Protoporphyrria	PROTO	Unwanted
Pseudomyotonia c491G>A	PMT_164	Unwanted
Pseudomyotonia c632G>T	PMT_211	Unwanted
Pseudomyotonia c857G>T	PMT_284	Unwanted
RNF11 growth retardation	RNF11	Unwanted
Spinal Muscular Atrophy	SMA	Unwanted
STAT5 13244	STAT5_13244	Unwanted
STAT5 13319	STAT5_13319	Unwanted
STAT5 13516	STAT5_13516	Unwanted
Streaked Hairlessness ERCC6L	SH_ERCC6L	Unwanted
Streaked Hairlessness TSR2	SH_TSR2	Unwanted
Thrombopathia	THR	Unwanted

Genetic Disease/Trait Testing Strategy



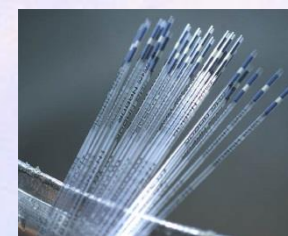
Trickle Down

Everyone



AI Sires

N=730



	National	AI Sires	AA	AU	BA	BB	CH	FR	HE	HO	JE	LM	PI	PT	SA	SH	SI
# Genotyped	499904	730	63	16	20	34	117	30	39	122	12	104	8	10	31	30	59
AM_662	0.00%	0.14%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
BD1	0.00%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
BLAD	0.21%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
BM	0.02%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
BY	0.38%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
CM	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
CMD1	0.21%	0.27%	-	-	-	5.88%	-	-	-	-	-	-	-	-	-	-	-
CMD2	0.09%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
CT	0.07%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
CTS_DELAG	0.63%	0.68%	-	-	-	14.71%	-	-	-	-	-	-	-	-	-	-	-
CVM	0.76%	0.14%	-	-	-	-	-	-	-	0.83%	-	-	-	-	-	-	-
DD	0.11%	0.14%	1.59%	-	-	-	-	-	-	-	-	-	-	-	-	-	-
DEB	0.00%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
DUMPS	0.00%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
HH1	0.46%	0.27%	-	-	-	-	-	-	-	1.63%	-	-	-	-	-	-	-
HH3	0.89%	0.41%	-	-	-	-	-	-	-	1.63%	-	-	-	-	-	-	-
HH4	0.05%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
IE	0.01%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
JH1	0.01%	0.27%	-	-	-	-	-	-	-	-	16.67%	-	-	-	-	-	-
MF_NG1621KC	0.04%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
MH2	0.09%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
MSU_SH	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
NH	0.01%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
OS	0.02%	2.47%	4.76%	6.25%	-	-	2.56%	3.33%	5.13%	2.45%	-	0.96%	-	-	3.23%	-	3.39%
PCS	0.01%	0.41%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
PMT_164	0.00%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
PMT_211	0.00%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
PMT_284	0.01%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
PROTO	1.04%	0.68%	-	-	-	-	-	-	-	-	-	4.81%	-	-	-	-	-
RNF11	0.36%	0.55%	-	-	-	11.76%	-	-	-	-	-	-	-	-	-	-	-
SMA	0.01%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
TH_IMPROVER	0.12%	0.27%	-	-	-	-	-	-	-	-	-	-	-	-	-	3.33%	-
WEAVER	0.00%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-

Purebred Herd

N=93,482



	National	Purebred	AA	AU	BA	BB	CH	FR	HE	HO	JE	LM	PI	PT	SA	SH	SI
# Genotyped	499904	93482	13093	1286	861	1334	25348	301	7446	4733	105	27469	373	707	2151	1581	6460
AM_662	0.00%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
BD1	0.00%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
BLAD	0.21%	0.04%	-	-	-	-	-	-	-	0.70%	-	0.00%	-	-	-	-	-
BM	0.02%	0.01%	-	-	-	-	-	-	-	-	-	-	-	-	0.46%	-	-
BY	0.38%	0.16%	-	-	-	-	0.01%	-	-	3.02%	-	0.00%	-	-	-	-	-
CM	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
CMD1	0.21%	0.05%	0.02%	-	-	2.62%	0.01%	-	-	-	-	0.03%	-	-	-	-	0.02%
CMD2	0.09%	0.02%	-	-	-	1.35%	-	-	-	-	-	-	-	-	-	-	-
CT	0.07%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
CTS_DELAG	0.63%	0.11%	-	-	-	6.90%	0.00%	-	-	0.04%	-	0.02%	-	-	-	-	-
CVM	0.76%	0.19%	0.01%	-	-	-	0.01%	-	-	3.51%	-	0.01%	-	-	-	0.06%	0.02%
DD	0.11%	0.20%	1.45%	-	-	-	0.00%	-	-	-	-	-	-	-	-	-	-
DEB	0.00%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
DUMPS	0.00%	0.00%	-	-	-	-	-	-	-	-	-	0.00%	-	-	-	-	-
HH1	0.46%	0.14%	-	-	-	-	0.00%	-	-	2.64%	-	0.00%	-	-	-	-	-
HH3	0.89%	0.32%	-	-	-	-	-	-	-	6.40%	-	-	-	-	-	-	-
HH4	0.05%	0.08%	-	-	-	-	-	-	-	1.65%	-	-	-	-	-	-	-
IE	0.01%	0.00%	-	-	-	0.07%	-	-	0.03%	-	-	-	-	-	0.05%	-	-
JH1	0.01%	0.01%	-	-	-	-	-	-	-	-	5.71%	-	-	-	-	-	-
MF_NG1621KC	0.04%	0.01%	0.01%	-	-	-	-	-	-	0.06%	-	0.00%	-	-	-	-	-
MH2	0.09%	0.02%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	0.03%
MSU_SH	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
NH	0.01%	0.02%	0.14%	-	-	-	-	-	-	-	-	-	-	-	-	-	-
OS	0.02%	0.04%	0.18%	-	-	0.07%	0.00%	-	-	0.06%	-	0.01%	-	0.14%	0.09%	-	-
PCS	0.01%	0.01%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
PMT_164	0.00%	0.00%	-	0.08%	-	-	-	-	-	-	-	-	-	-	-	-	-
PMT_211	0.00%	0.00%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
PMT_284	0.01%	0.00%	0.01%	-	-	-	0.00%	-	0.01%	-	-	-	-	-	-	-	-
PROTO	1.04%	1.87%	0.02%	0.08%	-	-	0.00%	-	0.01%	0.02%	-	6.35%	-	-	-	-	-
RNF11	0.36%	0.28%	-	-	-	19.57%	0.00%	-	-	-	-	0.01%	-	-	-	-	-
SMA	0.01%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
TH_IMPROVER	0.12%	0.06%	-	-	-	-	-	-	-	-	-	0.00%	-	-	-	3.29%	-
WEAVER	0.00%	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-

Crossbred Herd

N=405,692

Main breed of animal

	National	Commercial	AA	AU	BA	BB	CH	FR	HE	HO	JE	LM	PI	PT	SA	SH	SI
# Genotyped	499904	405692	30744	1426	3139	18234	78763	10647	21041	64493	1671	120861	586	2411	6033	7676	30986
AM_662	0.00%	0.00%	0.01%	-	-	0.01%	-	-	-	0.00%	-	0.00%	-	-	-	0.01%	0.00%
BD1	0.00%	0.00%	0.00%	-	-	-	0.00%	-	-	0.00%	-	0.00%	-	-	-	0.01%	-
BLAD	0.21%	0.25%	0.37%	0.28%	0.16%	0.30%	0.15%	0.23%	0.30%	0.43%	0.78%	0.20%	0.34%	0.12%	0.13%	0.07%	0.18%
BM	0.02%	0.02%	0.01%	-	-	0.03%	0.01%	-	0.00%	0.00%	-	0.01%	-	-	0.43%	0.01%	0.01%
BY	0.38%	0.43%	0.61%	0.49%	0.19%	0.45%	0.18%	0.57%	0.51%	1.10%	0.78%	0.25%	0.17%	0.21%	0.20%	0.09%	0.23%
CM	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
CMD1	0.21%	0.25%	0.13%	0.28%	0.25%	2.46%	0.12%	0.03%	0.07%	0.12%	0.06%	0.20%	-	0.04%	0.10%	0.14%	0.12%
CMD2	0.09%	0.10%	0.06%	0.07%	0.19%	1.10%	0.04%	-	0.02%	0.04%	-	0.08%	-	0.04%	0.05%	0.04%	0.05%
CT	0.07%	0.09%	0.12%	0.14%	0.10%	0.09%	0.06%	0.14%	0.13%	0.12%	0.06%	0.06%	-	0.04%	0.13%	0.03%	0.11%
CTS_DELAG	0.63%	0.75%	0.34%	-	0.80%	9.55%	0.28%	0.05%	0.11%	0.35%	0.12%	0.43%	0.68%	0.33%	0.31%	0.33%	0.28%
CVM	0.76%	0.89%	1.24%	0.35%	0.67%	1.04%	0.40%	1.47%	1.25%	1.94%	2.63%	0.56%	0.68%	0.41%	0.40%	0.39%	0.62%
DD	0.11%	0.09%	0.94%	0.07%	-	0.02%	0.02%	0.01%	0.00%	0.02%	-	0.03%	-	-	-	0.03%	0.00%
DEB	0.00%	0.00%	-	-	-	-	-	-	-	0.00%	-	-	-	-	-	-	-
DUMPS	0.00%	0.00%	-	-	-	0.03%	-	-	-	0.00%	-	0.00%	-	-	-	-	0.00%
HH1	0.46%	0.54%	0.58%	0.21%	0.19%	0.53%	0.16%	1.16%	0.55%	1.70%	1.14%	0.26%	0.34%	0.21%	0.25%	0.08%	0.22%
HH3	0.89%	1.03%	0.54%	-	0.13%	0.15%	0.02%	3.80%	0.49%	5.02%	3.11%	0.08%	-	-	0.05%	0.01%	0.08%
HH4	0.05%	0.05%	0.07%	-	-	0.08%	0.01%	0.11%	0.06%	0.15%	0.12%	0.02%	-	-	-	0.03%	0.02%
IE	0.01%	0.01%	0.01%	-	-	0.05%	0.00%	-	0.01%	0.00%	-	0.00%	-	-	0.02%	-	0.00%
JH1	0.01%	0.01%	0.00%	-	-	0.01%	-	0.03%	0.00%	0.03%	0.36%	0.00%	-	-	-	-	-
MF_NG1621KC	0.04%	0.05%	0.05%	0.07%	0.03%	0.07%	0.01%	0.08%	0.06%	0.18%	0.36%	0.01%	-	-	0.02%	-	0.02%
MH2	0.09%	0.10%	0.10%	0.07%	0.03%	0.17%	0.05%	0.06%	0.07%	0.07%	0.06%	0.09%	-	0.08%	0.10%	0.07%	0.09%
MSU_SH	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
NH	0.01%	0.00%	0.02%	-	-	-	-	0.01%	-	-	-	0.00%	-	-	-	-	-
OS	0.02%	0.02%	0.01%	-	-	0.01%	0.01%	0.07%	0.00%	0.03%	-	0.01%	-	-	0.03%	-	0.02%
PCS	0.01%	0.00%	0.00%	-	-	0.01%	0.00%	-	-	0.00%	-	0.00%	-	-	-	-	-
PMT_164	0.00%	0.00%	-	-	-	-	0.00%	0.01%	-	0.00%	-	0.00%	-	-	-	-	-
PMT_211	0.00%	0.00%	-	-	-	0.01%	-	-	-	0.00%	-	0.00%	-	0.04%	0.02%	-	0.00%
PMT_284	0.01%	0.01%	0.01%	-	-	0.01%	0.00%	0.01%	-	0.01%	-	0.00%	-	0.04%	0.05%	-	0.00%
PROTO	1.04%	0.85%	0.15%	0.21%	0.38%	0.29%	0.32%	0.03%	0.23%	0.20%	-	2.30%	0.17%	0.37%	0.17%	0.09%	0.20%
RNF11	0.36%	0.38%	0.17%	0.28%	0.45%	5.29%	0.13%	0.02%	0.02%	0.15%	-	0.19%	0.17%	0.17%	0.07%	0.13%	0.13%
SMA	0.01%	0.01%	0.01%	-	-	0.01%	0.00%	0.02%	-	0.01%	-	0.00%	-	-	-	-	0.00%
TH_IMPROVER	0.12%	0.13%	0.05%	-	0.13%	0.07%	0.05%	-	0.08%	0.04%	0.12%	0.06%	-	0.12%	0.13%	3.41%	0.06%
WEAVER	0.00%	0.00%	-	-	-	-	0.00%	-	-	-	-	0.00%	-	-	-	-	0.01%

FYI: Ireland has a high percent (~80%) of commercial crossbreds including beef X dairy

0.00% = <0.01%

What Does ICBF Do With a Genotype?



Male? Or Female?



Imputation
SNP + MS



Breed Composition



?



Angus



Limousin



Dexter



Saler

Matchmaker



Carrier Status



Brachyspina
Agerholm et al., 2006

ICBF Genetic Disease Management Plan

- 1) Information Booklet
- 2) Animal Disease Status
- 3) Advised Matings
- 4) Identify New Diseases



Genetic Disease and Trait Definition
and
Understanding Genetics



Disease/Trait Definition Booklets

Minimal Information

Alpha Mannosidosis

Abbreviations: AM 662, AM 967

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Lethal

Breeds found in: Angus (AM_961), Murray Grey (AM_961), Galloway (AM_662)

General: Affected calves are either aborted, born dead, die soon after birth, or die within the first year. Those born alive can show signs of ataxia, head tremor, aggression, and paralysis before death.

Common Ancestor: None identified

Extended Information

Alpha Mannosidosis

Abbreviations: AM 662, AM 967

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Lethal

Breeds found in: Angus (AM_961), Murray Grey (AM_961), Galloway (AM_662)

General: Affected calves are either aborted, born dead, die soon after birth, or die within the first year. Those born alive can show signs of ataxia, head tremor, aggression, and paralysis before death.

Common Ancestor:

Clinical: This lysosomal storage disease is caused by a build-up of mannose-rich compounds caused by deficiency of the alpha-mannosidase enzyme.

Gene: MAN2B1 (Mannosidase Alpha Class 2b Member 1)

Genetic: There are 2 mutations in MAN2B1 that cause this disease:

AM_662

Genetic: g.7:13956640G>A, c.662G>A, p.Arg221His

IDB probe: IDBv20700001524, IDBv20700001525, IDBv20700001526, IDBv20700001527, IDBv207000015248

Flanking Sequence (AM_662):

CGGGTCCCTTATGCATCCTGCCCTCTCTTGTCTCCCATCCCACTCGTCATCCCTCCCATCTCCAGATGGGTTTTGA
CGGCTTCTCTTTGGAC[G/A]CCTGGATTATCAAGACAAGAAGGTGCGGAAAAAGACGCTGCAGATGGAGCAGG
TGTGGCGGGCCAGCACCCAGCCTGAAACCTCCCACTGCCGACC

AM_961

Genetic: g.7:13957949_c.961T>C, p.Phe321Leu

Genetic Disease and Trait Definitions



Available at

http://www.icbf.com/?page_id=2170

Farmer Reports

Genotyped Animal's Carrier Status.

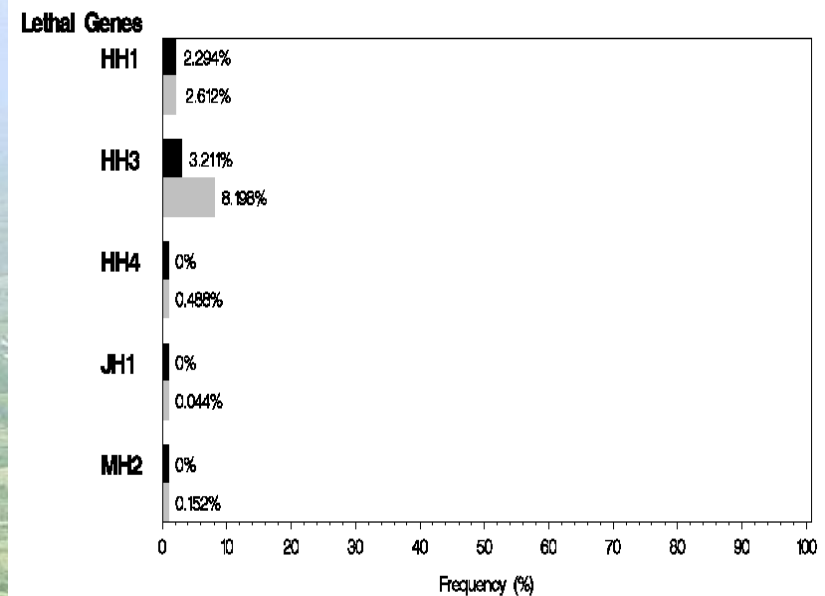


Summary for DAIRY females

Jumbo	Animal tag	Gender	Byear	Lethal	Unwanted
702	IE000123456789	F	2007	HH3/Norm	.
1012	IE000123456790	F	2011	HH3*/Norm	.
1020	IE000123456791	F	2011	HH3*/Norm	.

* = imputed genotype

Allele Frequency in Producer's herd (black bar) versus National dairy/beef herd (grey bar).



Farmer Reporting to ICBF of New Diseases

Congenital defect recording

Congenital defect reporting questionnaire

Thank you for participating in this programme. We hope that through data collection such as this we will be able to identify animals that carry various congenital defects and eradicate the defects that reduce farmer profitability. After the 8 mandatory questions (denoted by a *) in the beginning and 3 at the end, feel free skip any questions that do not apply to the animal you're reporting. If you have any questions about this survey please e-mail Health@ICBF.com Thank you again for your participation!

*** 1. Are you the animal's owner?**

Yes

No

If no, what is your role with the calf (Veterinarian, farm worker, knackery, etc)?

*** 2. What is your herd number?**

*** 3. What is the dam's tag number?**

I'm not o
did you
door ha

Farmer / Vet Reported

Neurological



Video by Catherine Carty



Wry Neck

Ataxia



Video by John Fagan

Thank you

- Weatherbys
 - Paul Flynn
 - Romy Morrin –O'Donnell
 - Rebecca Weld
- ICBF
 - Jennifer McClure
 - Thierry Pabiou
 - John McCarthy
 - Emma Dair
 - Francis Kearny
- Vets/Farmers
 - Catherine Carty
 - John Fagan
 - Joe Garrett
- Teagsac
 - Mike Mullen
 - Sinead Waters
 - Donagh Berry
- Vets/Farmers
 - Catherine Carty
 - John Fagan
 - Joe Garrett
- Multiple labs for controls
 - Cord Drogemuller
 - Tom Lawler
 - OMIA
- Funding
 - DAFM

Closing Thoughts

- Put QC pipeline in place sooner than later
- Recommend using 500+ SNP for parentage
- All animals carry genetic diseases
 - Management better than shotgun
- Farmers / Vets will inform you of novel genetic diseases if you provide a way

Questions?

