# **Bovine Genetic Disease Frequencies:**

# A national perspective on commercial and pedigree cattle in Ireland



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# Introduction

Historically, one only discovered if an animal was a carrier for a genetic disease after it had produced an affected offspring. Once identified the livestock producer typically had two choices 1) cull any ancestor or relative of the affected progeny, or 2) risk producing another affected calf. As molecular tests for causative mutations became available carrier animals could be identified, but often only AI bulls or elite pedigree animals were tested. Commercial producers tried to minimize their genetic disease risk by purchasing bulls assumed to be free of genetic diseases. To aid cattle genomics we developed a low cost, custom bovine Illumina single nucleotide polymorphism (SNP) genotype panel (International Dairy & Beef panel; IDB) which contains 39 validated probes for Mendelian diseases. Currently, >140,000 Irish commercial and pedigree beef and dairy animals have been genotyped with the IDBv2. Reports are generated to provide producers with their animal's genetic disease status and allow more informed breeding decisions.

# Disease Carrier Frequencies in Ireland by Animal Type

Tusit	Due e de Deus entre d'In	Deimi	Dune De ef	Cueselaned			
Trait	Breeds Reported In	Dairy		Crossbred			
Alpha Mannosidosis 662	Galloway	0.000%	0.000%	0.003%			
Beta Mannosidosis	Salers	0.000%	0.000%	0.031%			
Bovine Leukocyte Adhesion	Holstein	0.637%	0.003%	0.209%			
Deficiency	Holstein	1.663%	0.006%	0.294%			
Brachyspina Bulldog Dwarfism, Condrysplaia	поізіені	1.005%	0.000%	0.294%			
Buildog Dwarrishi, Condryspiala BD1	Dexter, Dahomey	0.000%	0.000%	0.003%			
Citrullinaemia		0.096%	0.000%	0.104%			
Coat Colour Dilution and	Hereford, Simmental,	0.05070	0.00070	0.10470			
Hypotrichosis	Galloway, Highland	0.019%	3.937%	10.607%			
Complex Vertebral							
Malformation	Holstein	2.482%	0.017%	0.640%			
		0.01.00/	0 4450/	0.000/			
Congenital Muscular Dystonia 1	Belgian Blue	0.010%	0.115%	0.328%			
Congenital Muscular Dystonia 2	Lan Duratania 2 Delaion Dhua			0.133%			
Congenital Muscular Dystonia z	Belgian Blue	0.000%	0.040%	0.15570			
Crooked tail syndrome	Belgian Blue	0.029%	0.230%	0.819%			
Deficiency of Uridine	Holstein	0.005%	0.000%	0.001%			
Monophosphate Synthase							
Dystrophic Epidermolysis	Rotes Hohenvieh	0.000%	0.000%	0.000%			
Bullosa		0.4640/	0.0000/	0.0740/			
HH1	Holstein	2.464%	0.003%	0.271%			
HH3	Holstein	7.376%	0.000%	0.044%			
HH4	Holstein	0.393%	0.000%	0.012%			
Hypotrichosis-KRT71	Hereford	0.010%	0.325%	0.457%			
Idiopathic Epilepsy	Hereford	0.000%	0.000%	0.000%			
JH1 Maple Syrup Uripe Shorthorp	Jersey Shorthorn	0.144%	0.000%	0.001%			
Maple Syrup Urine - Shorthorn MH2	Montbeliarde	0.000%	0.000%	0.000%			
Myoclonus, congenital	Hereford	0.000%	0.000%	0.000%			
Neuropathic Hydrocephalus	Angus	0.000%	0.006%	0.000%			
Osteopetrosis	Red Angus	0.000%	0.020%	0.003%			
Paunch Calf Syndrome	Romagnola	0.000%	0.014%	0.003%			
	Blond de'Aquitaine,						
Protoporphyria	Limousin	0.005%	1.674%	0.564%			
Pseudomyotonia-c.491G>A	Chianina,	0.000%	0.000%	0.000%			
, Pseudomyotonia-c.632G>T	Romagnola	0.000%	0.003%	0.008%			
, Pseudomyotonia-c.857G>T	Romagnola	0.005%	0.003%	0.007%			
Pulmonary Hypoplasis with		0.01.00/	0.000%	0.0040/			
Anascarca 1	Main, Chi, Shorthorn	0.010%	0.000%	0.001%			
RNF11	Belgian Bue	0.000%	0.359%	0.313%			
Spinal Muscular Atrophy	Brown Swiss	0.005%	0.000%	0.009%			
STAT1	Holstein	36.445%	23.287%	27.356%			
STAT3-19069	Holstein	43.947%	34.813%	44.143%			
STAT3-25402	Holstein	41.834%	39.585%	48.026%			
STAT5-13244	Holstein	47.161%	43.475%	47.404%			
STAT5-13319	Holstein	8.594%	13.268% 12.790%				
STAT5-13516	Holstein	47.181%	43.516%	47.421%			
Tibial Hemimelia-Improver	Simmental, Shorthorn,	0.000%	0.213%	0.229%			
· · · · · · · · · · · · · · · · · · ·	Galloway						
Dairy = animal's pedigree is <u>&gt;</u> 92% dairy breed							
Pure Beef = <u>&gt;</u> 92% of a single beef breed							
Crossbred = all others							
Breeds reported in = Bree	eds disease historica	ally repo	orted in				

# **Examples of Farmer Reports**

### Farm report of Genetic Disease/Major Gene Report

Mr/Mrs SMITH Herd IE1513 / BTEnumber

# Number of animals currently present in your herd: 529 # Number of beef animals genotyped for disease/major genes: 4 # Number of dairy animals genotyped for disease/major genes: 361

To increase accuracy an animal must have a genotype call rate of >95% to have its genetic disease and major gene allele status reported

More information about genetic diseases and Major genes reported below can be found at www.icbf.com

Summary for your dairy animals - Number of dairy animals genotyped: 361 The frequencies shown in the graphs are based on the number of genotyped animals

TYPE OF GENOTYPE: LETHAL (see Table 1.3 for details by animals)



National average for dairy

DUMPS	0.28% 0.01%
HH1	0.83%

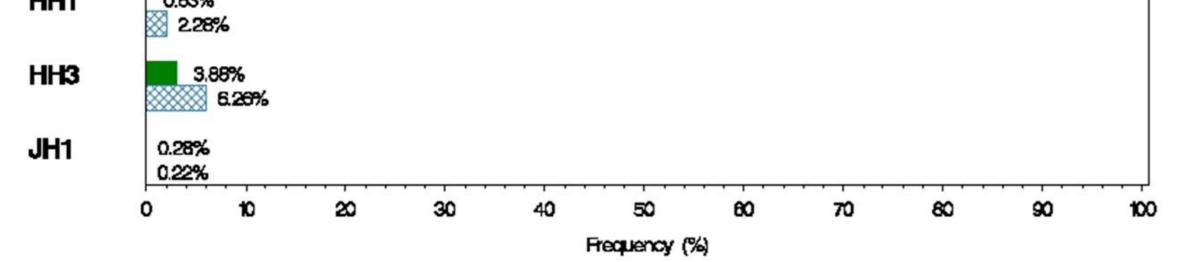


	Table 3. Summary for DAIRY animals									
Jumbo	nbo Animal tag Colour		Disease Major gene		Lethal		Phys. trait	Not genot.		
4388	IE1513 4388	BLACK_E		CAPN1_316 CAPN1_4751 CAPN1_530 CAST_2870 CAST_2959 DGAT1 LGB		KC				
4390	IE1513 4390	BLACK_E		CAPN1_316 CAST_2870 CAST_2959 DGAT1 LGB		KC				
4391	IE1513 4391	BLACK_E RED_E		CAPN1_316 CAST_2870 CAST_2959 DGAT1 LGB		KC				
4393	IE1513 4393	BLACK_E		CAST_2959 DGAT1						
4394	IE1513 4394	BLACK_E		CAPN1_316 CAST_2959 DGAT1 LGB	HH3					
4395	IE1513 4395	BLACK_E		CAST_2870 CAST_2959 DGAT1 LGB		KC				
4397	IE1513 4397	BLACK_E		CAPN1_316 CAST_2870 DGAT1		KC				
4398	IE1513 4398	BLACK_E		CAPN1_4751 CAPN1_530 CAST_2870 DGAT1 LGB		КС				
4400	IE1513 4400	BLACK_E		CAST_2959 DGAT1 LGB		KC				
4401	IE1513 4401	BLACK_E		CAPN1_316 CAST_2870 CAST_2959 DGAT1 LGB	HH3	КС				
4403	IE1513 4403	BLACK_E		CAPN1_316 CAPN1_4751 CAPN1_530 CAST_2870 CAST_2959 DGAT1		KC				
4405	IE1513 4405	BLACK_E		CAPN1_316 CAST_2870 CAST_2959 LGB		кс				
4406	IE1513 4406			CAPN1_4751 CAPN1_530 CAST_2959 LGB		кс				
4408	IE1513 4408	BLACK_E		CAPN1_316 CAPN1_4751 CAPN1_530 CAST_2870 CAST_2959 LGB		KC				

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- Weatherbys Ireland
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# Conclusions

Genetic disease carrier animals were identified for all but 5 of the validated disease probes and 2 of those (Dystrophic Epidermolysis Bullosa and Pseudomyotonia-c.491G>A) are only reported in breeds not present in Ireland (Rotes Honevnieh and Chianina, respectively). Disease carriers for many diseases were found across the breed type (Dairy, Purebred Breed, and Crossbreds) likely reflecting the high level of beef x dairy crossbreeding in Ireland, or that these diseases truly are historically present in multiple breeds.

By providing farmers with reports that identify carrier animals they



### All labs and scientists that donated DNA for probe validation

will be able to design a mating strategy that to reduces their disease

risk while maximizing their genetic gain.