

# AGREEMENT PROTOCOL

## Annex A-1

### List of Genetic Mutations and the Associated Breeds December 2017 version

*Each box in the first column shall be ticked individually to mean that the Laboratory has expertise and experience to perform the corresponding test. In order to avoid ambiguities, the total number of checked rows shall be summarized below the table.*

<i>Check if available</i>	Disease	Abr.	Concerned Breeds	Gene	Mutation
	GM1 Gangliosidosis	GM1	Korat	GLB1	c.1448G>C
			Havana Brown		
			Siamese/Oriental & LH		
			Thaï		
	GM2 Gangliosidosis	GM2-K	Korat	HEXB	c.39delC
	GM2 Gangliosidosis	GM2-B	Burmese/Bombay	HEXB	c.1244-8_1250del
			Tonkinese		
	Glycogen storage disease type IV	GSD4	Norwegian Forest Cat	GBE1	334bp ins 5' - 6.2kb del
	Hypertrophic Cardiomyopathy	HCM-MC	Maine Coon	MyBPC3	[A31P]
	Hypertrophic Cardiomyopathy	HCM-R	Ragdoll	MyBPC3	[R820W]
	Hypokalaemic Polymyopathy	HK	Burmese/Bombay	WNK4	c.2899C>T
			Tonkinese		
	Spinal Muscular Atrophy	SMA	Maine Coon	LIX1 (& LNPEP)	140kb del, exons 4-6
	Spasticity	CMS	Sphynx	COLQ	c.1190G>A
			Devon Rex		
	Burmese Head Defect	BHD	American Burmese	ALX1	c.496del CTCTCAGGACTG
			Bombay		
	Nudity	NUDE	Birman	FOXN1	c.1030_1033 delCTGT

*Total number of checked boxes in the table of this page: .....*

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Check if available	Disease	Abr.	Concerned Breeds	Gene	Mutation
	Progressive Retinal Atrophy	PRA-rdAc	Abyssinian/Somali	CEP290	IVS50+9T>G
			American Curl		
			Californian & Cornish Rex		
			Munchkin		
			Chausie		
			German Rex		
			Havana Brown		
			Ocicat		
			Savannah		
			Snowshoe		
			Siamese/Oriental & LH		
			Singapura		
			Thai		
			Tonkinese		
	Polycystic Kidney Disease	PKD	Persian/Exotic	PKD1	c.10063C>A
			British		
			Burmilla		
			Scottish/Highland		
			American SH/WH		
			Ragdoll		
			Birman		
			Selkirk		

Total number of checked boxes in the table of this page: .....

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<i>Check if available</i>	Disease	Abr.	Concerned Breeds	Gene	Mutation
	Pyruvate Kinase Deficiency	PKDef	Abyssinian/Somali	<i>PKLR</i>	c.693+304G>A
			Bengal		
			Chausie		
			LaPerm		
			Egyptian Mau		
			Maine Coon		
			Norwegian Forest Cat		
			Savannah		
			Siberian		
			Singapura		
			Toyger		

*Total number of checked boxes in the table of this page: .....*

Signed, on .....

For the Laboratory,

*(stamp and signature of the person authorised to act on each page)*