

# AGREEMENT PROTOCOL

## Annex A-1

### List of Genetic Mutations and the Associated Breeds November 2020 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		
			Thai		
	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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<i>Check if available</i>	Disease	Abr.	Concerned Breeds	Gene	Mutation
	Progressive Retinal Atrophy	PRA-rdAc	Abyssinian/Somali	<i>CEP290</i>	IVS50+9T>G
	Progressive Retinal Atrophy	PRA-b	Bengal	<i>KIF3B</i>	c.1000G>A
	Polycystic Kidney Disease	PKD	Persian/Exotic	<i>PKD1</i>	c.10063C>A
			Unknown breed		

*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	PKLR	c.693+304G>A
	Folded ears	Fd	Scottish/Highland	TRPV4	c.1024G>T
			American SH		
			British		

*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)*