

## **Known Genes/Mutations and available Genetic Testing:**

### **CEP290 IVS50+9T>G [1, 2]**

#### ***PRA-rdAc (late onset PRA)***

Abyssinian (Antagene, Genindexe, Genomia, Laboklin, Langford, Progenus, Texas A&M U, UC Davis)

American Curl (Antagene, Genindexe, Genomia, Laboklin, UC Davis)

American Wirehair (Antagene, Laboklin, UC Davis)

Balinese (Antagene, Genomia, Laboklin, Langford, UC Davis)

Bengal (Antagene, Genomia, Laboklin, UC Davis)

California Rex (Genindexe)

Chausie (Genindexe)

Colorpoint (UC Davis, Genomia, Laboklin)

Cornish Rex (Antagene, Genindexe, Genomia, Laboklin, Langford, UC Davis)

German Rex (Genindexe)

Havana Brown (Genindexe)

Javanese (Labokin, Langford, UC Davis)

Mandarin (Genindexe)

Minuet (Antagene)

Munchkin (Antagene, Genindexe, Genomia, Laboklin, UC Davis)

Ocicat (Antagene, Genindexe, Genomia, Laboklin, Langford, UC Davis)

Oriental shorthair (Antagene, Genindexe, Genomia, Laboklin, Langford, UC Davis)

Peterbald (Antagene, Genindexe, Genomia, Laboklin, UC Davis)

Savannah (Genindexe, Langford)

Siamese (Antagene, Genindexe, Genomia, Laboklin, Langford, Texas A&M U, UC Davis)

Singapura (Antagene, Genindexe, Genomia, Laboklin, Langford, UC Davis)

Somali (Antagene, Genindexe, Genomia, Laboklin, Langford, Progenus, Texas A&M U, UC Davis)

Snowshoe (Genindexe)

Thai (Antagene, Genindexe)

Tonkinese (Antagene, Genindexe, Genomia, Laboklin, Langford, UC Davis)

all breeds (Van Haeringen)

### **CRX n.546delC [3]**

#### ***PRA-rdy (early onset PRA, rod-cone dysplasia)***

Abyssinian (Laboklin, Texas A&M U, UC Davis)

Bengal (UC Davis)

Ocicat (Laboklin)

Somali (Laboklin, UC Davis)

all breeds (Van Haeringen)

### **Aipl1: c.577C>T, p.Arg193 [4-6]**

#### ***PRA-PD (Presian derived PRA)***

Angora (Laboklin)

Birman (Laboklin)

British shorthair/longhair (Laboklin)

Chartreux /Laboklin)

Chinchilla (Langford)

Colorpoint (Laboklin)

Exotic shorthair (Laboklin, Langford, UC Davis)

Himalayan (Langford, UC Davis)

Persian (Laboklin, Langford, Uc Davis)  
Ragdoll (Laboklin)  
Russian Blue (Laboklin)  
Scottish Fold (Laboklin)  
Selkirk Rex (Labokin)  
all breeds with potential Persian ancestry (UC Davis)

**unpub mutation [7]**  
***PRA-b (Bengal PRA)***  
Bengal (UC Davis, Langford)

## **STORAGE DISEASES WITH EYE INVOLVEMENT**

### ***Type 1 Gangliosidosis (GM1)***

**GLB1 c.1448G>C [8,9]**

Balinese (Laboklin)  
Javanese (Laboklin)  
Korat (Laboklin, Progenus, UC Davis)  
Oriental shothair (Laboklin)  
Peterbald (Laboklin)  
Seychellois (Laboklin)  
Siamese (Laboklin, Progenus)  
Thai (Laboklin)  
Tonkinese (Laboklin)  
all breeds (Van Haeringen)

### ***Type 2 Gangliosidosis (GM2, Sandhoff)***

**HEXB c.1244-8\_1250del [10]**

Burmese (Genomia, Laboklin, Langford, UC Davis)  
all breeds with potential Burmese ancestry (UC Davis)  
all breeds (Van Haeringen)

**HEXB c.39delC [11]**

Korat (UC Davis, Laboklin, Van Haeringen)  
Siamese (Laboklin)  
Oriental shorthair (Laboklin)

**additional information:** other mutations causing GM2 in cats are described and tests are available for DSH

### ***Alfa Mannosidosis***

**MAN2B1 c.1748delCCAG [12]**

Persian (PennGen, Laboklin, Van Haeringen)

### ***Mucopolysaccharidosis I (Hurler-Scheie syndrome)***

**IDUA c.1107\_1109 del [13]**

DSH (PennGen, Van Haeringen)

### ***Mucopolysaccharidosis VI (Maroteaux-Lamy Syndrome VI, MPSM, MPS6)***

**ARSB c.1427T>C [14]**

Balinese (Laboklin)  
Birman (UC Davis, Laboklin)

Javanese (Laboklin)  
Oriental shorthair (Laboklin)  
Peterbald (Laboklin)  
Seychellois (Laboklin)  
Siamese (Laboklin, Langford, PennGen, Texas A&M U, UC Davis, Van Haeringen)  
Ragdoll (UC Davis)  
Thai (Laboklin)  
Tonkinese (Laboklin)  
all breeds with potential Siamese ancestry (Langford, UC Davis)

**ARSB c.1558G>A [15,16]**

Balinese (Laboklin)  
Birman (Laboklin)  
Javanese (Laboklin)  
Oriental shorthair (Laboklin)  
Peterbald (Laboklin)  
Seychellois (Laboklin)  
Siamese (Laboklin, PennGen, UC Davis)  
Thai (Laboklin)  
Tonkinese (Laboklin)

***Mucopolysaccharidosis VII (Sly Syndrome, Mucopolysaccharidosis, MPS7)***

various mutations in GUSB gene causing MPS7 are described in cats and tests are available for DSH or “all breeds” depending by labs [17,18]

***Mucopolipidosis II (I-cell disease)***

**GNPTA unpub mutation**

DSH (PennGen)

**LABS CONTACTS:**

**Antagene:** Antagene [www.antagene.com](http://www.antagene.com)

**Genindexe:** Genindexe [www.genindexe.com](http://www.genindexe.com)

**Genomia:** Genomia [www.genomia.cz](http://www.genomia.cz)

**Laboklin:** Laboklin [www.laboklin.de](http://www.laboklin.de)

**Langford:** Langford Veterinary Services [www.langfordvets.co.uk/lab\\_pcrnews.htm](http://www.langfordvets.co.uk/lab_pcrnews.htm)

**PennGen:** University of Pennsylvania Veterinary Medicine Section of Medical Genetics  
[www.vet.upenn.edu](http://www.vet.upenn.edu)

**Progenus:** Progenus [www.progenus.be](http://www.progenus.be)

**Texas A&M U:** Texas A&M University [www.catdnatest.org](http://www.catdnatest.org)

**UC Davis:** University of California Davis – Veterinary Genetics Laboratory  
[www.vgl.ucdavis.edu](http://www.vgl.ucdavis.edu)

**Van Haeringen:** Van Haeringen [www.vhlgenetics.com](http://www.vhlgenetics.com)

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