



Genindexe

La génétique à votre service

Glycogen Storage Disease type IV: Norwegian Forest Cat

GSDIV is an inherited disorder of the Norwegian Forest Cat where an essential enzyme required to produce glucose (energy) is deficient. Normally, excess glucose obtained in the diet or produced by conversion of proteins and fats is stored in many tissues as a very highly branched chain of glucose residues called glycogen. A special biological catalyst called glycogen branching enzyme (GBE for short) is necessary during glycogen synthesis to produce the branching structure. When needed for energy, glucose molecules are removed from glycogen and digested within the tissue or released into the blood circulation for use by other tissues. The ability to add and remove glucose molecules from glycogen efficiently is dependent on its highly branched structure. Affected animals store a very abnormal glycogen, a glucose polymer devoid of branch points, in most tissues and cannot therefore use the stored glucose to produce the required energy.

Clinical Signs:

Affected kittens are usually stillborn but may develop normally until 4-5 months of age before suffering terminal neuromuscular degeneration. By eight months of age, GSD IV results in severe muscular weakness, atrophy and contractures, and inability to use the limbs. The cat may die suddenly from heart failure.

Inheritance:

GSD IV is inherited as a simple autosomal recessive trait. Practically, this means that both parents must be carriers of the trait in order for offspring to be affected.

Because they are clinically-normal, carriers of GSD IV may be active breeders in a cattery, passing their carrier status along to the next generation, and never suspected until an affected kitten is born. Furthermore, because stillbirth or early death is not so uncommon for any of many reasons, a GSD IV affected kitten may be discarded without diagnosis and its carrier parents continue breeding unsuspected.

The parents will pass their carrier status along to 50% of all their offspring, both male and female, when mated to a non-carrier cat. When two carriers are mated, 25% of the offspring will be affected and two-thirds of the clinically-normal littermates will be carriers.

DNA Test Results:

The DNA test detects directly whether the mutation that causes GSD IV is present in a cat's DNA in two copies, as in affected kittens, one copy, as in carriers, or not at all, as in genetically normal cats.

You will be provided with one of the following test results:

Homozygote non carrier +/+	Non carrier	Does not develop GSDIV	Has 2 normal copies of GBE1 gene	Pass a normal copy of GBE1 gene
Hétérozygote carrier +/-	Carrier	Does not develop GSDIV	Has 1 normal copy and 1 mutated copy of GBE1 gene	Can pass a normal or a mutated copy of GBE1 gene (50%)
Homozygote affected -/-	Affected	Neonatal mortality Can stay alive until 15 months	Has 2 mutated copy of GBE1 gene	Pass a mutated copy of GBE1 gene

		Male						
		Non Carrier		Carrier		Affected		
		+	+	+	-	-	-	
Female	Non Carrier	+	+/+	+/+	+/+	+/-	+/-	+/-
		-	+/+	+/+	+/+	+/-	+/-	+/-
	Carrier	+	+/+	+/+	+/+	+/-	+/-	+/-
		-	+/-	+/-	+/-	-/-	-/-	-/-
	Affected	+	+/-	+/-	+/-	-/-	-/-	-/-
		-	+/-	+/-	+/-	-/-	-/-	-/-

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