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Member Education Series

Heritable Defects - Genetics

All Breeder members must comply with the Code of Practice for the Breeding of Animals with Heritable Defects that Cause Disease, a Victorian State Government legislation. Under this code, there are several requirements that all GCCFV Breeder members must be aware of.

Heritable Conditions may be broken down into categories as follows, depending on whether they are caused by simple dominant or recessive genes, or more complicated genetic transfer eg polygenic inheritance

1) Dominant diseases / conditions

A dominant condition only requires one copy of the defective gene to be present for the disease to be caused i.e. both the heterozygous and homozygous states for the defective gene each develop the disease. This also includes dominant conditions that can only show partially or some conditions that only show in a particular sex.

The breeding of cats with the following diseases caused by a heritable defect must be conducted in accordance with the Code. Evidence of testing regime must be maintained and copies of DNA test results held on the cats records. A copy of the test results must be submitted to GCCFV for any breeding cats owned by the breeder or used in matings.

- Polycystic Kidney Disease.
- Folded ears associated with osteochondrodystrophy.
- Aplasia or hypoplasia of long bones

The below table has the following definitions

Carrier (is affected) = heterozygote (i.e. one clear gene and one defective gene), displays degrees of disease.
 Affected = homozygous for genes (i.e. two defective genes) displays severe form of disease.
 Clear = homozygous for clear genes (i.e. two clear genes) and is free of the disease.

Parent combination	Theoretical status of progeny	Heritable disease requirements
Clear x Clear	100% Clear	No restriction
Clear x Carrier	** 50 % Clear	<ol style="list-style-type: none"> 1. Progeny must be tested for the heritable defect**. 2. The severity of the disease in the Carrier progeny must be assessed by a veterinary practitioner and the cat managed in accordance with the instructions of a veterinary practitioner. 3. A diseased (Carrier) cat must not be disposed of to another person without advice of the cats heritable disease status. 4. A diseased (Carrier) cat must be de-sexed unless they are to be used in an approved breeding program, must not be permitted by their owner to suffer from their condition and must be under the supervision and monitoring of a veterinary practitioner.
	50 % Carrier (that may be diseased to some degree)	
Carrier x Carrier	** 25% Clear 50% Carrier (diseased to some degree)	<ol style="list-style-type: none"> 1. Breeding prohibited unless as part of an approved breeding program. 2. All progeny must be tested for the heritable disease. ** 3. The severity of the disease in Affected and Carrier progeny must be assessed by a veterinary practitioner and the animal managed in accordance with the

	25% Affected (usually seriously diseased)	<p>instructions of a veterinary practitioner.</p> <ol style="list-style-type: none"> A diseased (Carrier and Affected) cat must not be disposed of to another person without advice of the cat's heritable disease status. Diseased (Carrier and Affected) cats must be de-sexed unless they are to be used in an approved breeding program, must not be permitted by their owner to suffer from their condition and must be under the supervision and monitoring of a veterinary practitioner.
Affected x Clear	100% Carrier (will have a degree of the disease)	<ol style="list-style-type: none"> Breeding prohibited unless as part of an approved breeding program and only with the purpose of establishing sufficient breeding stock for the breeding program to develop Clear animals. The severity of the disease in progeny must be assessed by a veterinary practitioner and the cat managed in accordance with the instructions of a veterinary practitioner. A diseased (Carrier and Affected) cat must not be disposed of to another person without advice of the cat's heritable disease status. Diseased (Carrier and Affected) cats must be de-sexed unless they are to be used in an approved breeding program, must not be permitted by their owner to suffer from their condition and must be under the supervision and monitoring of a veterinary practitioner.
Affected x Carrier and	50% Carrier, 50% Affected (all will be diseased to some degree or be seriously diseased)	<ol style="list-style-type: none"> Breeding prohibited. Use of these combination is an offence under the Act.
Affected by Affected	100% Affected (usually seriously diseased)	

**Testing is required as in practice the unpredictable nature of the process of gene inheritance in these combinations may cause variation in the actual % outcomes per generation. As carriers may express varying degrees of the heritable disease they must be tested, assessed and monitored by a veterinary practitioner experienced with the disease to determine the impact on the animal.

2) Simple recessive diseases / conditions

A recessive condition or disease requires 2 copies of the affected gene to be present.

Whilst there are currently no diseases or conditions for cats listed under this category, breeders, must be wary of new conditions or diseases that may fall into this category and take appropriate action accordingly.

Parent breeding combinations of cats with a heritable defect of this type that result in a disease that causes suffering and disability in progeny must not be used outside of an approved breeding program and only where the GCCFV believes it to be justifiable in the short term to establish breeding stock that are Clear. Evidence of testing regime must be maintained and copies of DNA test results held on the cats records. A copy must be submitted to GCCFV for any breeding cats.

The below table has the following definitions

Carrier = heterozygote (i.e. one clear gene and one defective gene) and does not exhibit the disease.
 Affected= homozygous for heritable defect genes (i.e. two defective genes) and is affected by the disease.
 Clear = homozygous for clear genes (i.e. two clear genes) and is free of the disease.

Parent combination	Theoretical status of progeny	Heritable disease requirements
Clear x Clear	100% Clear	No restriction.
Carrier x Clear	50 % Clear 50 % Carrier (not diseased)	<ol style="list-style-type: none"> Progeny to be used for breeding purposes must be tested for the heritable defect**. Progeny should all be tested for the heritable defect. Carrier animals must be desexed if not to be used for breeding purposes.
Carrier x Carrier	25% Clear 50% Carrier 25% Affected (diseased)	<ol style="list-style-type: none"> Breeding not recommended. Must only occur as part of an approved breeding program. All progeny must be tested for the heritable defect**. Diseased (Affected) and Carrier progeny must not be disposed of to another person without advice of the animal's heritable defect status.

		<ol style="list-style-type: none"> Diseased (Affected) and Carrier animals must be de-sexed if not to be used for breeding purposes. Diseased animals must not be permitted by their owner to suffer from their condition and must be under the supervision and monitoring of a veterinary practitioner.
Affected x Clear	100% Carrier	<ol style="list-style-type: none"> Breeding not recommended. Must only occur as part of an approved breeding program and only with the purpose of establishing sufficient breeding stock for the breeding program to develop Clear animals. Progeny must not be disposed of to another person without advice of the animal's heritable defect status. Carrier animals must be de-sexed unless to be used in an approved breeding program.
Affected x Carrier	50% Carrier 50% Affected (diseased)	<ol style="list-style-type: none"> Breeding prohibited. Intentional or reckless use of these combinations is an offence under the Act. Affected (diseased)
and		
Affected x Affected	100% Affected (diseased)	

**Testing is required as in practice the unpredictable nature of the process of gene inheritance in these combinations may cause variation in the actual % outcomes per generation.

Recessive conditions / diseases that may take years to develop

There are some simple recessive diseases that may take years to develop signs of the disease in the homozygous state for the defective gene. All progeny will initially appear to be unaffected by the disease. Depending on the severity of the disease and time of onset of the breeding program the cat may therefore have been bred before it is known they carry the heritable defect. Whilst there are currently no diseases or conditions for cats listed under this category, breeders must be wary of new conditions or diseases that may fall into this category and take appropriate action accordingly.

Parent combination	Theoretical status of progeny	Heritable disease requirements
Clear x Clear	100% Clear	No restriction.
Carrier x Clear	** 50 % Clear 50 % Carrier	<ol style="list-style-type: none"> Progeny to be used for breeding purposes should be tested for the heritable defect**. All progeny should be tested for the heritable defect. Carrier animals should be de-sexed if not to be used for breeding purposes.
Carrier x Carrier	** 25% Clear 50% Carrier 25% Affected (may develop disease)	<ol style="list-style-type: none"> Breeding not recommended. Must only occur as part of an approved breeding program. All progeny must be tested for the heritable defect. A diseased (Affected) animal must not be disposed of to another person without advice of the animal's heritable defect status. Affected progeny (or any juvenile offspring confirmed as 'Affected' on test) should be de-sexed unless they are to be used in an approved breeding program, must not be permitted by their owner to suffer from their condition if it develops and should be under the supervision, advice and monitoring of a veterinary practitioner.
Affected x Clear	100% Carrier	<ol style="list-style-type: none"> Breeding not recommended. Should only occur as part of an approved breeding program and only with the purpose of establishing sufficient breeding stock for the breeding program to develop Clear animals. Progeny must not be disposed of to another person without advice of the animal's heritable defect status. Carrier animals should be de-sexed unless to be used in an approved breeding program.
Affected x Carrier	50% Carrier 50% Affected (may develop the disease)	<ol style="list-style-type: none"> Breeding prohibited. Intentional or reckless use of these combinations is an offence under the Act. Under exceptional circumstances the Affected x Carrier combination may occur as part of an approved breeding program but only with the purpose of establishing sufficient breeding stock for the breeding program to develop Clear animals. Progeny must not be
and		
Affected x Affected	100% Affected	

Affected (may develop the disease) disposed of to another person without advice of the animal's heritable defect status.

**Testing is required as in practice the unpredictable nature of the process of gene inheritance in these combinations may cause the actual % outcomes per generation to vary from the theoretical outcomes.

Simple recessive diseases that are sex linked or show weak penetrance and limited expression of the disease

There are some recessive conditions / diseases that may be sex linked or have weak penetrance resulting in only a few affected individuals. Whilst there are currently no diseases or conditions for cats listed under this category, breeders, must be wary of new conditions or diseases that may fall into this category and take appropriate action accordingly.

Advice from a veterinary practitioner and GCCFV should be sought before considering a breeding program where this type of issue is identified.

Simple recessive diseases that are also dependant on over-riding or modifying genetic effects for full expression, before they pose a threat as a debilitating condition. This includes conditions where the vast majority of genetically affected individuals fail to exhibit the full range of clinical signs unless modifying factors are present –All progeny may initially appear to be unaffected by the disease.

Parent combination	Theoretical status of progeny	Heritable disease requirements
Clear x Clear	100% Clear	No restriction.
Carrier x Clear	50 % Clear 50 % Carrier	<ol style="list-style-type: none"> 1. Progeny to be used for breeding purposes should be tested for the heritable defect**. 2. All progeny should be tested for the heritable defect. 3. Carrier animals should be de-sexed if not to be used for breeding purposes.
Carrier x Carrier	** 25% Clear 50% Carrier 25% Affected (may develop disease)	<ol style="list-style-type: none"> 1. Not recommended. Should only occur as part of an approved breeding program. 2. All progeny should be tested for the heritable defect. 3. Affected (may develop disease) animals must not be disposed of to another person without advice of the animal's heritable defect status. 4. Carrier animals should be de-sexed if not to be used for breeding. 5. Affected (may develop disease) should be de-sexed if not to be used for breeding, must not be permitted to suffer from their condition by their owner if it develops and must be under the supervision, advice and monitoring of a veterinary practitioner if it does.
Affected x Clear	100% Carrier	<ol style="list-style-type: none"> 1. Must only occur as part of an approved breeding program. 2. Carrier animals should be de-sexed if not to be used in a breeding program.
Affected x Carrier and Affected x Affected	50% Carrier 50% Affected (may develop the disease) 100% Affected (may develop the disease)	<ol style="list-style-type: none"> 1. Prohibited (see exception). 2. Intentional or reckless use of these combinations outside of an approved breeding program is an offence under the Act. 3. The Affected x Carrier combination may occur but only as part of an approved breeding program and only with the purpose of establishing sufficient breeding stock for the breeding program to develop Clear animals. Affected progeny must not be disposed of to another person without advice of the animal's heritable defect status.

**Testing is required as in practice the unpredictable nature of the process of gene inheritance in these combinations may cause variation in the actual % outcomes per generation.

Polygenic disease / condition

A polygenic disease / condition is one where more than one gene is involved and environmental effects can also add to the severity of the condition. While the following diseases are not listed in the Schedule of the Act they are examples of diseases in this grouping that have widely divergent signs –hip dysplasia and

elbow dysplasia. These are also conditions where simple and/or effective DNA tests are unlikely to be developed.

Recognised inherited diseases

There are some inheritable disease / conditions that produce significant potential health risks in small numbers of affected individuals, but where there is no advance warning mechanism offered through the early onset of signs or the availability of a reliable genetic test, to be able to predict the development of debilitating disease in later life.

Approved breeding programs

Approved breeding programs must be reviewed every 3 years by GCCFV to evaluate progress in reducing the prevalence of the heritable defect and the disease it causes and to ensure that there is compliance by the breeder member with this Code. A breeding program should be developed by the breeder which will reduce the prevalence of the heritable defect.