GENETIC AND CONGENITAL CAUSES OF FELINE SKIN OR EYE MALAISE

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CATHERINE F LE BARS discusses how to spot dermatological and ocular problems that, in some cases, may result in a need for long-term management of pedigree patients – or even euthanasia

FOLLOWING on from the previous discussion of neurological and musculoskeletal defects (VT38.22), this second article of a five-part series on pedigree cats examines conditions found in these breeds that have a suspected or proven genetic basis, with emphasis on ocular and dermatological conditions.

Not all of these conditions are congenital ( ), but many express themselves in young animals. Where a definite mode of inheritance has been confi rmed, it will be described.

Inherited ophthalmic and dermatological conditions in cats are uncommon, although a number of disorders have been recognised, which may be hereditary.

Many of these reports are based on small numbers of animals. However, certain breeds, such as Persians, Siamese and Himalayans, appear to be over-represented, suggesting a potential genetic basis.

Corneal disease

Corneal sequestrum is unique to cats and recognised in Persians, Himalayans, Burmese and Siamese. It presents as a localised necrosis of the epithelium and anterior stroma, which results in

a characteristic black lesion. Sequestra are often located in the central cornea. Clinical signs include lacrimosis, brown ocular discharge and blepharospasm.

Numerous factors have been implicated in the aetiology, including FHV-1 infection and chronic keratitis secondary to other conditions. Treatment options include surgical removal or medical management, with attention to any underlying predisposing factors.

Medical management is aimed at preventing infection and providing lubrication until the sequestrum is sloughed naturally. Surgical removal may shorten recovery time.

Retinal disease

Two forms of progressive retinal atrophy (PRA) have been recognised in different strains of Abyssinian cats. One form has been described in a group of Abyssinian cats in the UK.

Kittens display nystagmus and mydriasis as early as four weeks of age, and ophthalmoscopy reveals marked retinal changes by eight to 12 weeks of age. Affected cats are usually blind by one year of age. This form of PRA has an autosomal dominant mode of inheritance.

Rod-cone degeneration is the more commonly recognised form of the disease. It presents as a slowly progressive degeneration of the rod and cone systems, with a late onset of clinical symptoms.

Retinas are usually ophthalmoscopically normal until one to two years of age, although some studies using electron microscopy have detected lesions in the central retina as young as seven to 12 weeks. Alterations are seen as “greyish” discolourations, most evident in the mid-peripheral and peripheral tapetal fundus. Retinal vascular attenuation and tapetal hyper-reflectivity are usualy pronounced by the time the cat is presented for evaluation.

Vision loss progresses to total blindness over a period of two to four years. This form has an autosomal recessive mode of inheritance.

Miscellaneous ocular diseases

Congenital strabismus is recognised commonly in the Siamese and Himalayan breeds, often in association with pendular nystagmus, and is the result of abnormal routing of the optic nerve fibres at the optic chiasm.

The strabismus is thought to be an attempt at functional correction of the reduced medial visual fields. The abnormality is linked to the genes that determine coat colour and is believed to be a polygenic trait with a threshold character. Upper eyelid agenesis may affect several kittens in a litter and has been recognised in the Burmese. The degree of agenesis is variable and can range

from mild defects to an almost complete absence of the upper eyelid. In severe cases, the conjunctival fornix is also absent. Clinical signs result from an inability to close the eyelids and the resultant corneal irritation. Other ocular abnormalities, such as persistent pupillary membranes and cataracts, may be present in affected cats.

Treatment options depend on the severity of the agenesis and may be medical or surgical. Inherited entropion is uncommon in the cat (in contrast to dogs), but may occasionally be present in brachycephalic breeds, such as the Persian.

The lower lids are most commonly affected and the mode of inheritance is not yet known. Cysts similar to the apocrine hidrocystomas seen in human beings are occasionally seen in the eyelids of Persian and Himalayan cats.

The number and size of cysts may vary and ocular discomfort is rarely evident. Cysts may be surgically removed, but recurrence is common.

Hereditary cataracts are rare, but have been reported in Himalayans and Persians.

Defects of dermal structural integrity

Dystrophic and/or junctional epidermolysis bullosa s yndromes resul t from defects in the attachment of dermis to the epidermis, and are classified into three groups according to the location of the defect.

In the simplex form, the defect occurs at the level of the epidermal basal cell structure; the junctional form involves the basement membrane; and the dystrophic form involves the subepidermal anchoring fibrils.

All three forms have been recognised in cats, including Siamese and Persians. In affected cats, mild trauma causes the appearance of flaccid bullae, which rupture and form flat ulcers. Lesions may be present at birth or develop within a few weeks, and include paronychia, ulceration on the metacarpal, metatarsal and digital pads, as well as oropharyngeal ulcers.

All forms of the disease except simplex are fatal. The mode of inheritance is unknown, but it is believed to be a single gene defect. Ehlers-Danlos syndrome, also known as feline cutaneous asthenia, is a rare disorder of collagen production that results in hyperextensible and fragile skin.

The skin of affected cats has one-tenth the tensile strength of normal skin and kittens may p r e s e n t wi t h severe lacerations subsequent to mild trauma, such as playing with littermates. As in humans, these cats may also exhibit joint laxity and cardiac disease. Diagnosis is based on histopathological examination of collagen structure, and a skin extensibility index.

Cats with Ehlers-Danlos syndrome may be managed conservatively. However, the longterm outlook is poor, with many clients choosing euthanasia when chronic wounds are present.

Studies have suggested two modes of inheritance: autosomal dominant in domestic shorthairs (DSH) and, less commonly, an autosomal recessive form seen in Himalayans.

Hereditary alopecia and hypotrichosis

Of these two conditions, the more common is hypotrichosis, in which affected animals have less hair than normal.

The sparseness may be generalised, spare the extremities, or correlate with hair colour. The hair bulbs are often poorly formed and the hairs are easily dislodged. Affected cats often have epidermal anomalies and are predisposed to the development of comedones, hair follicle infections and hair foreign body granulomas. Kittens suffering from hereditary alopecia often have a small amount of hair at birth that is lost by two weeks of age, but may regrow to a variable degree. Sphynx cats are selectively bred for the condition. An autosomal recessive mode of inheritance has been identified in some cases.

Follicular dysplasia occurs in rex cats. The hair of affected kittens is woolly, becoming curly and soft as the animal matures. Guard hairs and vibrissae may be abnormal or absent. This defect is inherited as an autosomal recessive trait in rex cats and as an autosomal dominant trait in related long-haired breeds.

Dermatological conditions

• Familial benign mastocytosis has been described in young Siamese cats.

• Seborrhoea has a diverse group of syndromes, which may affect localised parts of the epithelium or be generalised, such as the primary seborrhoea seen in young Persian kittens.

Diagnosis is based on the histopathological demonstration of orthokeratotic hyperkeratosis, and the condition has an autosomal recessive mode of inheritance.

• Dermatophytosis (usually *Microsporum canis*) occurs most often in long-haired cats, with Himalayan and Persian cats appearing to be particularly susceptible. A genetic basis or infl uence is suspected.

• Atopy is a hypersensitivity to environmental allergens. Clinical signs include miliary dermatitis, symmetrical alopecia, head and neck pruritus and eosinophilic granuloma complex. Diagnosis relies on the exclusion of other causes of pruritus. Intradermal testing can be performed to identify the allergen. However, results are less reliable in cats than in dogs.

One study has suggested a genetic component and the condition appears to be more common in pedigree cats.

• Basal cell tumours are one of the most common cutaneous tumours in cats and present typically as small benign masses. Complete surgical excision is the treatment of choice.

A breed predilection has not been clearly established, although some studies suggest that oriental and Siamese breeds may be predisposed. Mammary tumours appear also to be more common in Siamese cats.

Summary

Few DNA tests are available in cats and the heritability of many of the conditions described above is still poorly understood.

However, as mapping of the feline genome progresses, our ability to identify the presence of genetic disease in young animals should improve, allowing the reduction of disease incidence in specific breeds by the employment of selective breeding.

Main photo: ISTOCKPHOTO/ANNA UTEKHINA

Main picture: Sphynx cats are selectively bred for hereditary alopecia. Inset left: Persians suffer from a variety of ocular and dermatological conditions known, or believed to have, a genetic basis.

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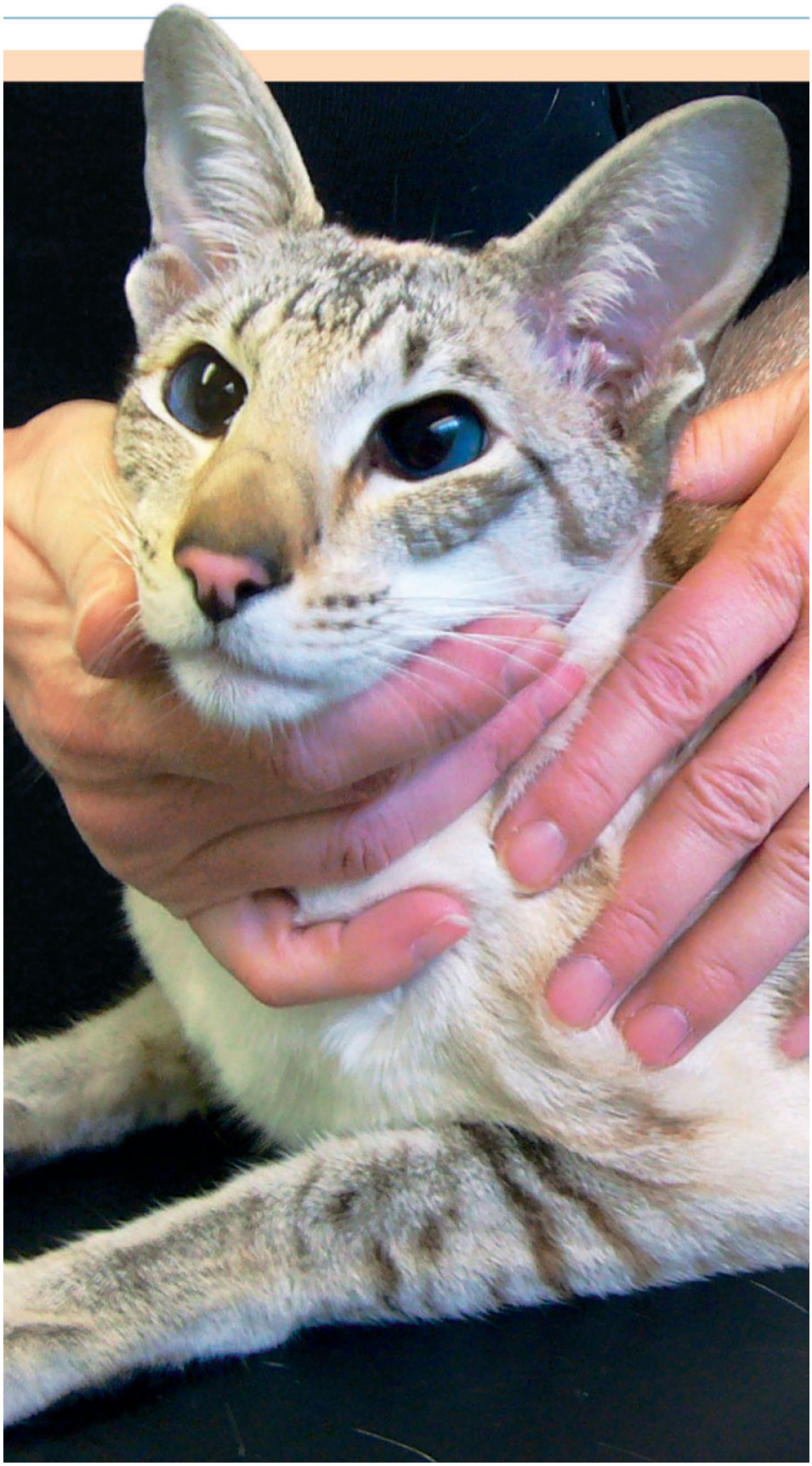

Breed	Disorder
Abyssinian or Somali	Progressive retinal atrophy
Bengal	None recognised
Birman	Congenital hypotrichosis; corneal dermoid; congenital cataract
British shorthair	None recognised
Burmese or Asians	Lipaemia of the aqueous humour; corneal dermoid
Cornish rex	Congenital hypotrichosis
Devon rex	Congenital hypotrichosis; *Malassezia* dermatitis; urticaria pigmentosa
Himalayan	Ehlers-Danlos syndrome (dermatosparaxis or cutaneous asthenia)
Korat	None recognised
Maine coon	None recognised
Manx	Corneal dystrophy
Munchkin	None recognised
Norwegian forest cat	None recognised
Persian or chinchilla	Primary seborrhoea; dermatophytosis; idiopathic facial dermatitis; otitis externa; progressive retinal atrophy; epiphora; corneal sequestration; coloboma; entropion; and cataracts
Ragdoll	Congenital hypotrichosis; junctional epidermolysis bullosa; periocular leukotrichia; mammary tumour; congenital strabismus and retinal degeneration
Scottish fold	None recognised
Siamese or Balinese	Strabismus; dystrophic and/or junctional epidermolysis bullosa
Siberian	None recognised
Snowshoe	None recognised
Sphynx	Alopecia; *Malassezia* dermatitis (and paronychia) and urticaria pigmentosa
Turkish van	None recognised

TABLE 1. Common breeds and their predispositions to dermatological and ocular disorders