



## HEREDITARY EYE DISEASES IN GERMAN SHEPHERD DOG

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### ABSTRACT

**Hereditary eye diseases occur to varying degrees in all dog breeds. Individual purebred breeds have specific predispositions to various eye disorders. The German Shepherd is diagnosed mainly with chronic superficial keratitis/pannus, but also with: distichiasis, plasmoma/atypical pannus, corneal dystrophy, persistent pupillary membranes, cataract, cone degeneration, retinal dysplasia, optic nerve hypoplasia/micropapilla, and limbal melanoma. Individual ocular abnormalities are manifested by characteristic clinical manifestations and ophthalmological findings. Some eye diseases can lead to blindness, others affect the comfort of life or work use of the dog to varying degrees. A thorough knowledge of individual ocular pathologies in a particular breed leads not only to the identification of the diagnosis but also to the correct assessment of the dog's breeding usability.**

**Key words: eye diseases; German Shepherd dog; hereditary**

### INTRODUCTION

The German Shepherd is a working dog that was initially developed for guarding and herding sheep [29]. It

is classed under the number 166 in the FCI (Federation Cynologique Internationale) Group 1—Sheepdogs and Cattle Dogs [13]. The German Shepherd Dog is a relatively new breed of purebred dogs that only dates back to 1899. It is the product of Captain Max vom Stephanitz's (1864—1936) vision of creating the perfect working dog that would possess the following essential qualities: intelligence, ability, a weatherproof coat and beauty [29].

Not only nature but also health is important for its usability. The breed is predisposed to several diseases, including eye diseases. Visual disturbances can be caused by a variety of causes. A specific group consists of hereditary eye diseases (HED).

The inherited forms of eye diseases are arguably the best described and best characterized of all inherited diseases in the dog. A principal reason is that the eye is very accessible, and much of it can be examined in detail using non-invasive techniques, making it relatively easy to detect abnormalities, even if they do not impair vision significantly [25]. The HED group involves disorders present at birth (e.g., microphthalmia), but also those appearing in the adult age (e.g., hereditary cataracts, and late-onset progressive retinal atrophy). In addition, these HEDs include sluggish diseases with minimum influence on vision (e.g., a mild form of a persistent pupillary membrane, and multifocal retinal dysplasia); progressive ones seriously impairing vision (e.g., corneal endothelium dystrophy,

and progressive retinal atrophy); polygenic diseases (e. g., entropium, and ectropium); and those transmissible according to simple Mendelian rules (with autosomal recessive/dominant trait) inherited by siblings at high frequency (e. g., prcd (Progressive Rod-Cone Degeneration) form of progressive retinal atrophy) [1, 27, 33, 35, 36, 37].

The Genetics Committee of the American College of Veterinary Ophthalmologists maintains a database of eye diseases in dogs. It provides an up-to-date overview of the prevalence of individual eye abnormalities in specific purebred breeds. According to the database, the most common eye diseases found in German Shepherd Dogs are: distichiasis, plasmoma/atypical pannus, corneal dystrophy, chronic superficial keratitis/pannus, persistent pupillary membranes, cataract, cone degeneration, retinal dysplasia, optic nerve hypoplasia/micropapilla and limbal melanoma [2]. There is also a reference that German Shepherds could be prone to lens luxation when the incidence is found in family lines [21, 26].

### **Distichiasis**

Distichiasis refers to single or multiple hairs arising from the free lid margin. They usually arise singly or with two or more hairs from the Meibomian duct openings. Distichiasis is considered to be inherited, but the exact mode of transmission is unknown [34]. However, the use of the dog in breeding is possible [2].

The clinical signs of cilia disorders are: blepharospasm, epiphora, conjunctival hyperaemia, and corneal ulceration [3]. The treatment for canine distichiasis consists of either temporary removal of the offending distichia by manual epilation or the permanent destruction of the distichia follicle by electroepilation, cryoepilation, or other various surgical procedures [14].

### **Plasmoma/atypical pannus**

Plasma cell infiltration of the nictitating membrane or plasmoma can cause thickening, depigmentation, and follicle formation. German shepherds appear to be predisposed [19]. Patchy depigmentation and nodular thickening of the anterior surface of the third eyelid, usually near the margin, is commonly seen in association with the keratitis or rarely occurs without concurrent corneal lesions. In the latter instance, this syndrome has been called plasmoma, but it is important to realize that this is not a neoplastic condition [24]. German Shepherds appear to be predis-

posed to this condition [19]. The inheritance of plasmoma is not defined, and the use of a dog with plasmoma is not recommended in breeding [2]. Treatment generally consists of topical ophthalmic dexamethasone four times daily, initially, or subconjunctival or systemic corticosteroids. Topical ophthalmic cyclosporine ointment, tacrolimus drops or 1% pimecrolimus administered 2–3 times daily are also effective [19].

### **Corneal dystrophy**

Corneal dystrophy is any primary, bilateral, inherited disorder of the cornea not accompanied by corneal inflammation or systemic disease. Most corneal dystrophies in the dog appear clinically as grey-white or silver, crystalline or metallic opacities in the central or paracentral cornea. The condition is bilateral and often appears as nearly symmetric lesions. Corneal dystrophy may affect the corneal epithelium, stroma or endothelium [4, 5, 6, 7, 38].

In the years 1991–2015, corneal dystrophy capture rate in the German Shepherds was 4.6% and in the years 2015–2020 it was up to 4.5%. The mode of inheritance is not defined; however, the breeding usability of a dog with this diagnosis is possible [2]. In general, corneal dystrophies do not respond to medical treatment. The corneal lesions can be removed by keratectomy if the opacity is obstructing vision significantly. However, it is possible that the opacities will recur after keratectomy. Therefore, surgery is recommended for corneal dystrophy only as a last resort measure in dogs with significant visual deficit [38].

### **Chronic superficial keratitis/pannus**

The most frequent ocular disease in German Shepherds is chronic superficial keratitis (CSK) [17]. It is a progressive, bilateral, inflammatory, and potentially blinding disease of the canine cornea. It is also known as: German Shepard pannus, Überreiter's syndrome, and degenerative pannus. Clinically, CSK is manifested initially at the temporal or inferior temporal limbus as a red, vascularized, conjunctival lesion. Early in the disease, vascularization and pigmentation occur at the temporal cornea and progress centrally (Fig. 1). As the disease progresses, it spreads as a fleshy, well-vascularized lesion that migrates toward the central cornea (Fig. 2) [38].

The German Shepherd Dog has a higher incidence of pannus than any other breed. The MHC (Major Histocompatibility Complex) class II risk haplotype has been



**Fig. 1. Chronic superficial keratitis/pannus in a 5-year-old female German Shepherd (Source: Pavol Zubrický)**



**Fig. 2. Chronic superficial keratitis/pannus in a 3-year-old male German Shepherd (Source: Pavol Zubrický)**

shown. Although there are likely several other genes and environmental factors that contribute to CSK, a recent paper suggested that MHC class II is a major genetic risk factor. It is not recommended to use a dog with pannus in breeding [2].

Pannus cannot be cured. The therapeutic goal should be control and some-times regression of the lesions so that blindness can be avoided. Treatment consists of topical application of a potent and penetrating corticosteroid eyedrop [24]. The prognosis for achieving disease control and preserving vision may be poorer in those animals that show rapid onset of disease in early adulthood [10].

### **Persistent pupillary membranes**

The persistent pupillary membrane (PPM) is a remnant of foetal mesodermal tissue that is responsible for providing nutrition to the forming lens during embryonic development. In dogs, it usually atrophies within six weeks of life. Clinical symptoms can be extremely variable—from insignificant clinical conditions to visual disturbances or the development of cataracts at an older age [40]. In general, small remnants spanning from one portion of the iris to another (iris-to-iris persistent pupillary membranes—PPMs) sometimes cross the pupil, but they have no discernible visual consequences. A visual impairment may occur, however if the strand contacts the cornea (iris-to-cornea PPMs) or lens (iris-to-lens PPMs) it may create an opacity within the visual axis [28]. A therapy is rarely necessary for PPMs [20]. The most commonly diagnosed PPMs in German Shepherds are iris-to-iris PPMs. In this form, heredity is not yet defined. The breeding use of a dog with iris-to-iris PPMs is a breeder option [2].

### **Cataracts**

A cataract is a partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results [2]. Cataracts are divided according to aetiology into primary and secondary. Primary cataracts: all bilateral or unilateral cataracts and especially cortical cataracts are known or presumed hereditary eye diseases (KP-HED). Secondary cataracts: cataracts known to be caused by physical influences (trauma, electric, irradiation), ocular inflammation, metabolic diseases, nutritional deficiencies, age, intoxication or another KP-HED. We recognize cataracts congenital and non-congenital. A congenital cataract is considered a cataract, if it is diagnosed by the age of 8 weeks of age, or if it is diagnosed later in life, but there is a distinct indication whether the cataract is congenital in origin (e.g., in microphthalmos, adjacent to PPM, or persistent hyaloid artery). Non-congenital cataracts are divided according to the location to cortical cataracts, posterior polar cataracts, nuclear cataracts and other lens opacities [32]. The treatment of cataracts is clearly a surgical condition in dogs. There are many different surgical procedures to remove lenses and cataracts in veterinary ophthalmology. Phacoemulsification cataract surgery is the most common type of cataract surgery performed in all species of animals performed by veterinary ophthalmologists world-wide [15].

The most commonly diagnosed cataract in the German Shepherd is a cortical cataract with presumed autosomal recessive inheritance. Affected individuals are not recommended for use in breeding [2].

### **Cone degeneration**

Cone degeneration is an autosomal recessively inherited early degeneration of the cone photoreceptors. Afflicted puppies develop day-blindness and colour blindness, but they remain ophthalmoscopically normal their entire life. Electroretinography is required to definitively diagnose the disorder [2]. Inheritance is autosomal recessive [12], affected dogs are not recommended for breeding [2]. The predominant causes of achromatopsia are mutations of the CNGA3 gene [22].

### **Retinal dysplasia**

The term retinal dysplasia embraces several congenital/neonatal conditions resulting from disorderly proliferation and atypical differentiation of the retina during embryonic life. In addition to genetically determined hereditary retinal dysplasia, a wide variety of extraneous insults (for example, infectious agents such as canine herpes virus and irradiation) to the developing retina may cause acquired, non-inherited, retinal dysplasia [5].

Retinal folds are linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, and detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The breeding use of a dog with retinal folds is possible. The genetic relationship between folds and more severe form of retinal dysplasia is undetermined [2].

### **Optic nerve hypoplasia/Micropapilla**

Optic nerve hypoplasia is a congenital defect of the optic nerve which causes blindness and abnormal pupil response in the affected eye [2], and it is usually diagnosed early in life. There is no therapy [18]. Affected dogs must not be used in breeding [2]. Although most cases happen to be sporadic isolates in families, it is now clear that many cases are caused by mutations in genes involved in eye development. [8]. Micropapilla refers to a small optic disc which is not associated with vision impairment. Inheritance is not defined and the usability of an individual is the breeder's option [2].

### **Limbal melanoma**

Epibulbar or limbal melanocytic neoplasms appear

clinically as darkly pigmented masses arising from the limbus and expanding into the adjacent cornea and sclera or as masses arising posterior to the limbus and expanding into the adjacent sclera [9], [23]. Rare histologically malignant limbal melanomas have been described, and some otherwise benign neoplasms may include areas with cells that are less pigmented or amelanotic and mitotically active [9, 11, 30, 31, 39]. Surgical treatment of limbal and scleral diseases are usually directed toward the removal of neoplastic and inflammatory masses. The German Shepherd breed appears most frequently affected [16]. Dogs with limbal melanoma should not be included in the breeding [2]. The inheritance of limbal melanoma has not been defined [12].

## **CONCLUSIONS**

The German Shepherd is predisposed to several inherited eye diseases affecting different eye structures. As the heredity of most of them have not been yet precisely defined, they can currently be identified on the basis of an ophthalmological finding. Therefore, it is recommended to perform a preventive clinical examination of the eyes before placing the dog in the breed.

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