“Malattie oculari con base ereditaria o sospetta tale in Schnauzer e Pinscher ”

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Come e quando tutto è nato……..

PASCIA', ch. Italiano 1979, Vicews (RCACIB) 1980

A Scuola con…..

- Massimo Parrino
- Marcello Koch
- Marisa Brivio Chellini
- Milia Pozzi Tarlarini
- Paolo Raimondi
- ……e tanti altri….

Malattie Oculari

1. *Congenite* :(per definizione presenti alla nascita) spesso ***ereditarie*** o con predisposizione di razza;
2. *Non congenite (acquisite)*: ***ereditarie*** (per definizione malattie di sviluppo, compaiono dopo la nascita);
3. *(Acquisite non ereditarie)*.

Malattie Oculari ereditarie

- "*Hereditary Eye Diseases*" (KP-HED);

- Known HED: (K-HED), di provata ereditarietà, ovvero è stata identificata la **mutazione** genetica ed esiste un test genetico disponibile (sono ancora pochi e non per tutte le malattie);

- Presumed HED: (P-HED), di presunta (ma non certa) ereditarietà, perchè non è stata ancora identificata la mutazione genetica e non è quindi disponibile un test genetico affidabile (vale per la maggior parte delle malattie, definite anche *malattie familiari* con predisposizione di razza).

Quando una malattia può essere considerata ereditaria in una determinata razza?

1. **Quando la frequenza in quella razza è più alta che in altre;**
2. **Quando la frequenza aumenta nella medesima razza;**
3. **Quando la frequenza è più grande in soggetti imparentati;**
4. **Quando ha aspetto e localizzazione particolari;**
5. **Quando ha età di insorgenza e progressione particolari;**
6. **Quando è uguale a patologie dimostrate ereditarie in altre razze;**
7. **Quando si suppone una base genetica, ma non è stata individuata la mutazione.**

Perché attuare un programma di prevenzione e controllo della malattie oculari ereditarie?

1. **Per ridurre l'incidenza di malattie oculari gravi e spesso invalidanti;**
2. **Per migliorare il benessere dei nostri animali;**
3. **Per fornire maggior garanzia agli acquirenti;**
4. **Per migliorare il "pool" genetico di razza;**
5. **Per elevare la qualità dei soggetti allevati;**
6. **Per bloccare la diffusione di malattie oculari gravi ed emergenti.**

Come attuare un programma di prevenzione e controllo della malattie oculari ereditarie?

1. **Sottoporre a visita oculistica specialistica <u>tutti</u> i soggetti (adulti) destinati alla riproduzione (esame da ripetere annualmente);**
2. **<u>Associare</u> alla visita la richiesta di esecuzione di test specifici genetici disponibili per ogni razza presso laboratori accreditati (una volta sola nella vita);**
3. **<u>Non riprodurre</u> i soggetti affetti (anche se di pregio!) e, per alcune patologie gravi, anche i figli di tali soggetti;**
4. **Sterilizzare i soggetti malati.**

Modalità di trasmissione delle oculopatie ereditarie

1. **Autosomico recessivo:**

- i cani malati hanno di solito genitori (portatori o carrier) che non manifestano clinicamente la malattia;
- l'incidenza della malattia aumenta con la consanguineità;
- sono colpiti entrambi i sessi;

2. **Autosomico dominante:**

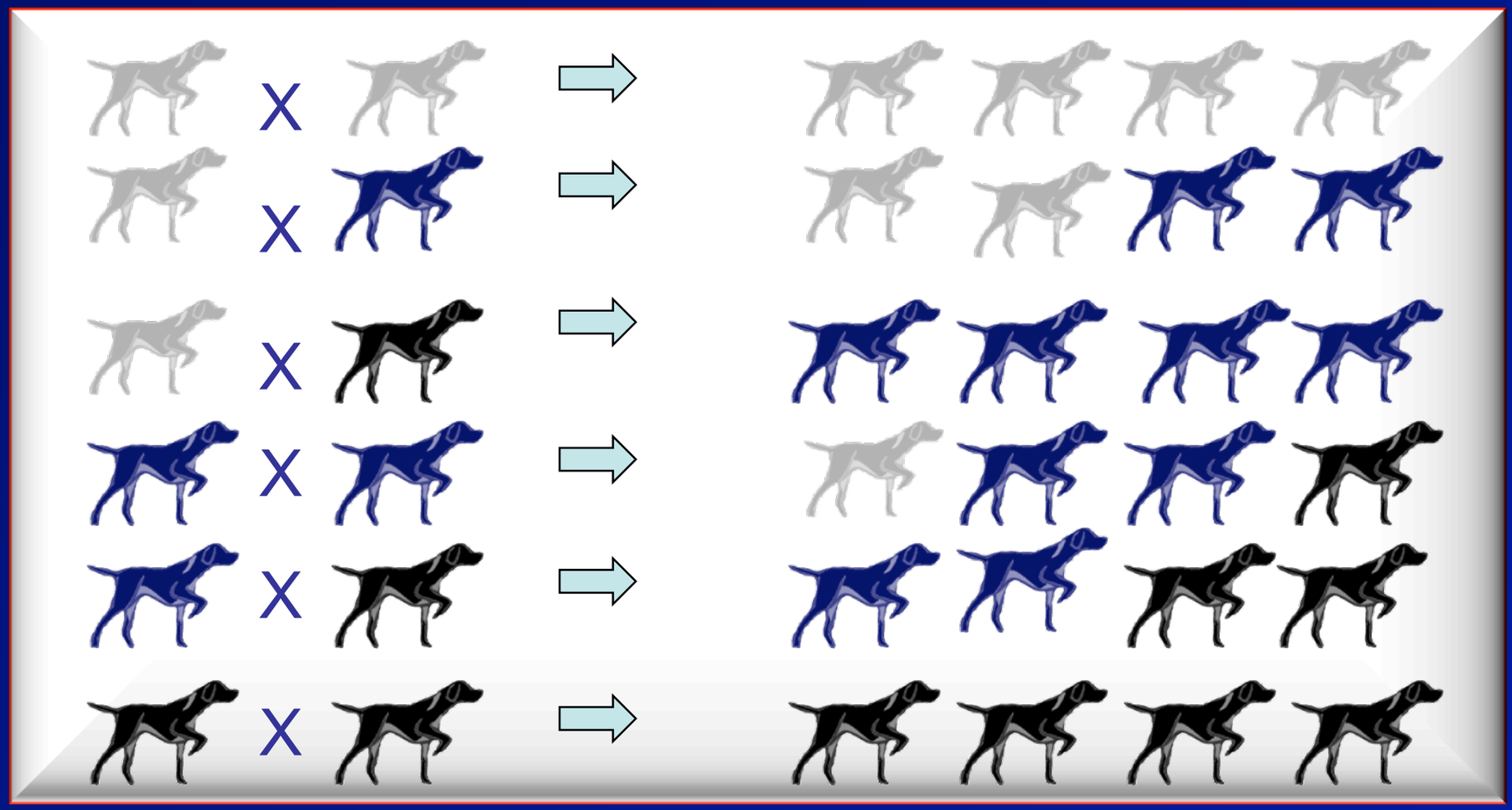
- i cani malati hanno almeno un genitore (affetto) che manifesta clinicamente la malattia;
- il figlio di un malato ha il 50% di possibilità di essere malato;
- sono colpiti entrambi i sessi;

3. **Legata al sesso (X-Linked):**

- i cani malati hanno di solito genitori che non manifestano clinicamente la malattia;
- sono malati prevalentemente i maschi;
- le femmine possono essere malate se si accoppia padre malato con femmina portatrice.

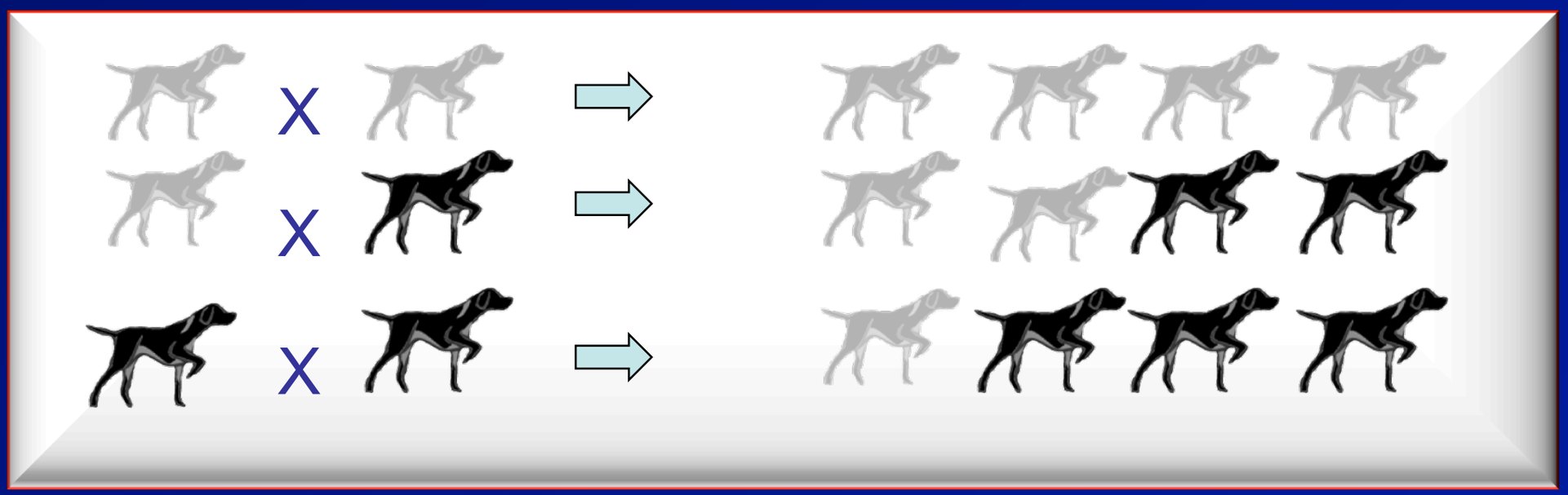
Anomalie di tipo mendeliano a comportamento ***Recessivo***

Possibilità di "gestire" o di "eradicare" la malattia



Anomalie di tipo mendeliano a comportamento ***dominante***

Possibilità di "gestire" o di "eradicare" la malattia



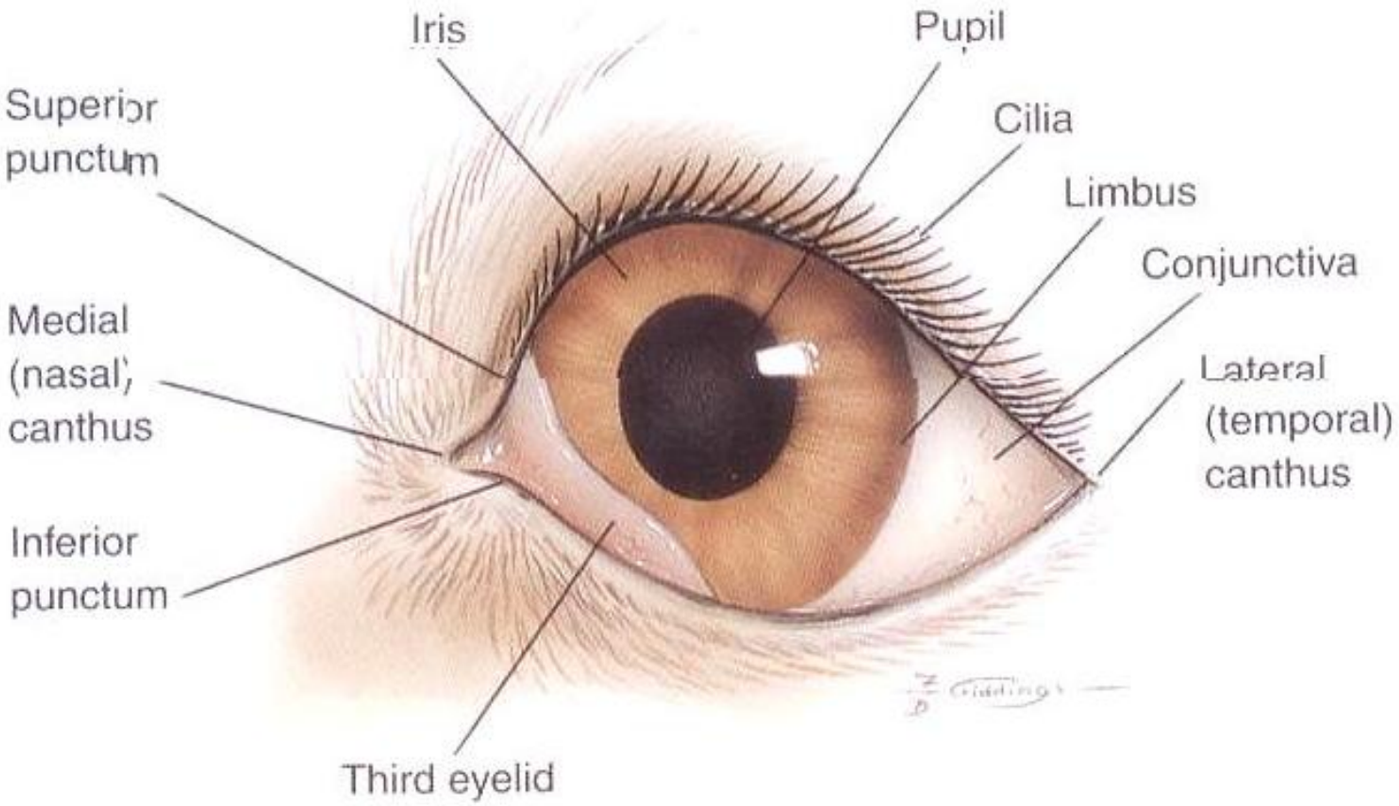
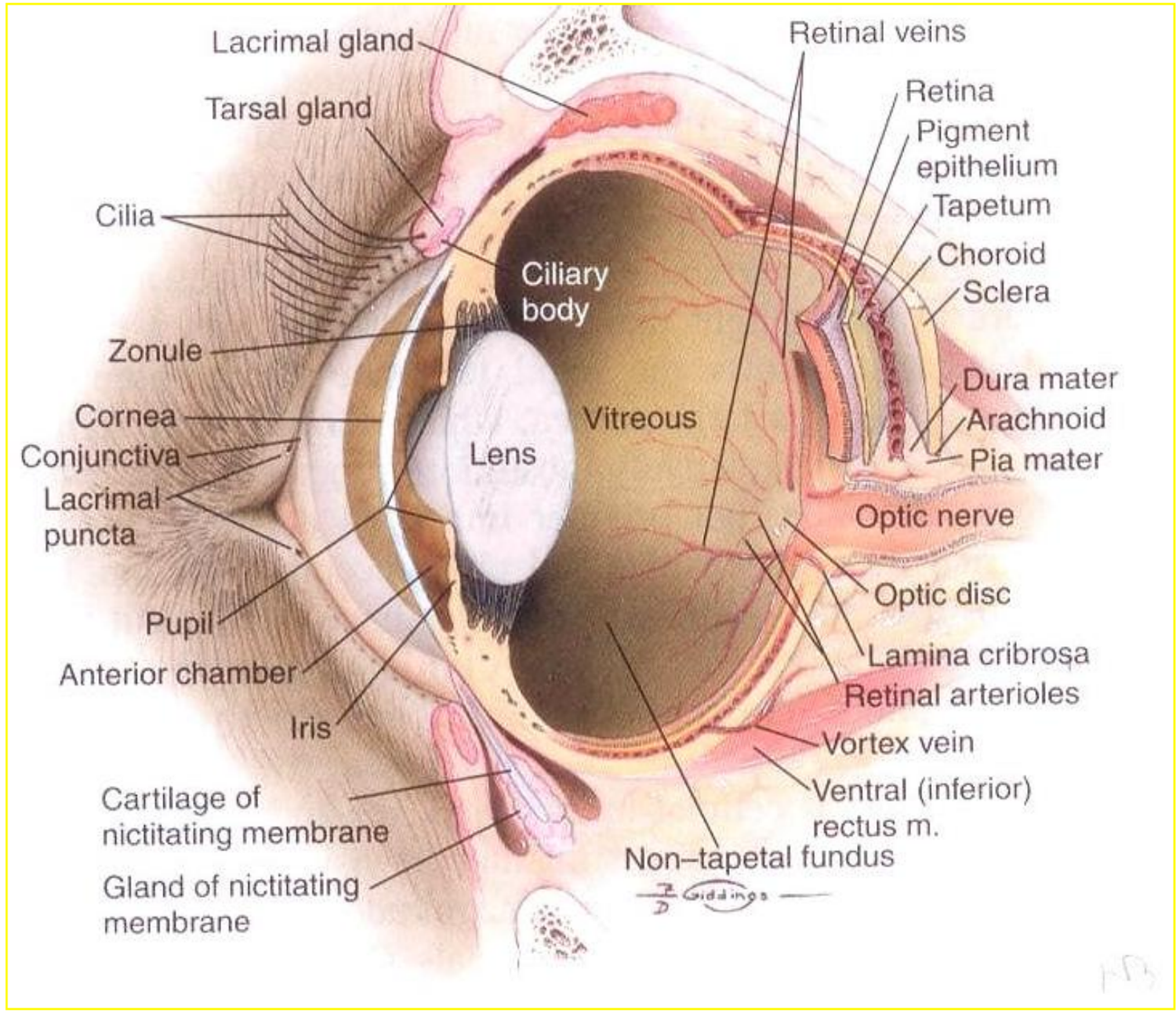
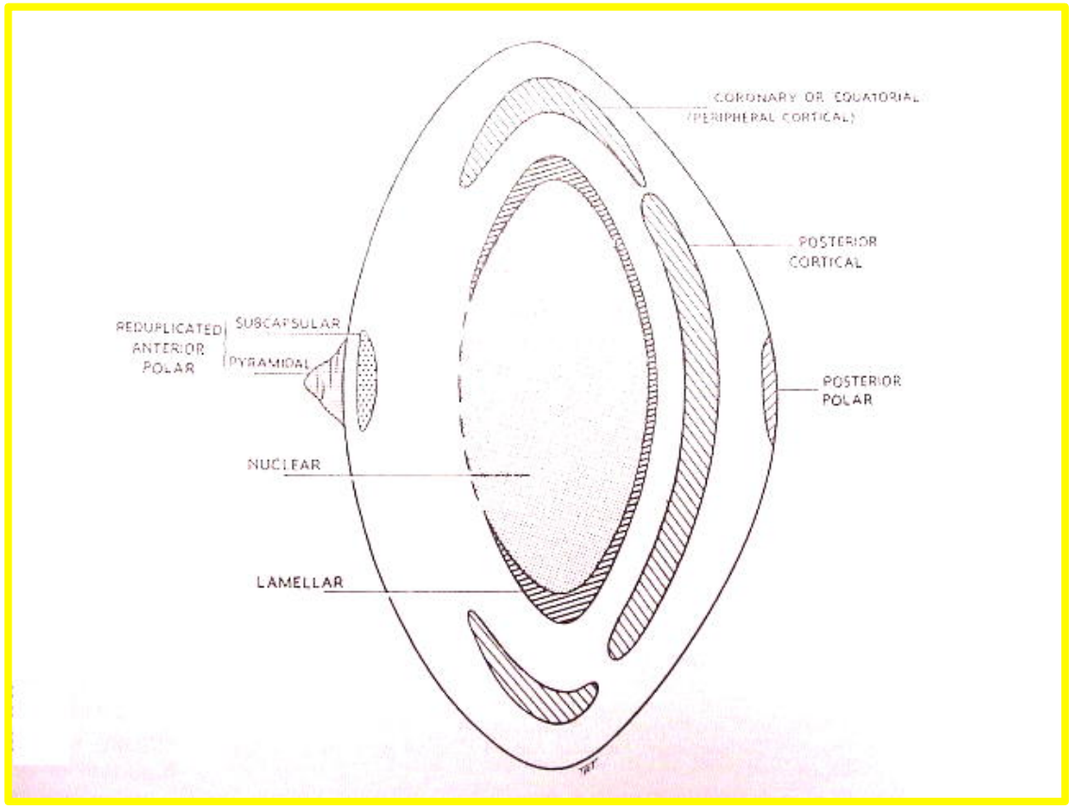
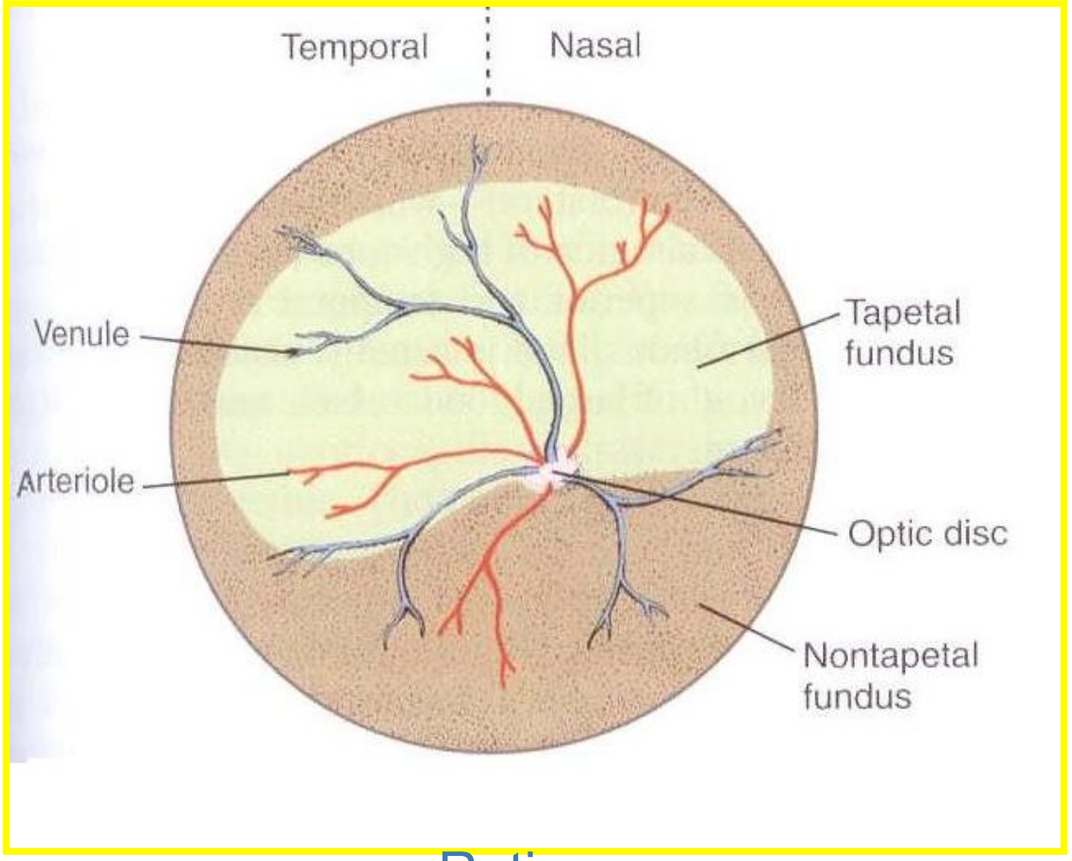


Figure 1-1. Frontal view of the external structures of the canine eye.


Lacrimal gland
Tarsal gland
Cilia
Ciliary body
Zonule
Cornea
Conjunctiva
Lacrimal puncta
Pupil
Anterior chamber
Iris
Cartilage of nictitating membrane
Gland of nictitating membrane
Lens
Vitreous
Retinal veins
Retina
Pigment epithelium
Tapetum
Choroid
Sclera
Dura mater
Arachnoid
Pia mater
Optic nerve
Optic disc
Lamina cribrosa
Retinal arterioles
Vortex vein
Ventral (inferior) rectus m.
Non–tapetal fundus



Cristallino (Lente)



Retina

Giant Schnauzer	

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B	Progressive Retinal Atrophy (PRA)	Initial clinical signs from 3 y.o.	Presumed autosomal recessive	PRCD	1,2,3,4,5
C	Retinal dysplasia -multifocal	Number decreases with growth of the animal	Unknown	NO	1,2
D	Glaucoma		Unknown	NO	1,2

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Other ocular disorders (reported)

	Diagnosis	Source
A	Eversion of the cartilage of the nictitating membrane	ACVO genetics committee
B	Corneal dystrophy -stromal	ACVO genetics committee
C	Primary hyperplastic tunica vasculosa lentis/ Primary hyperplastic primary vitreous (PHTVL/PHPV)	ACVO genetics committee
D	Entropion	French National Panel
E	Persistent pupillary membranes -iris to iris	ACVO genetics committee

References

1. Rubin LF. Inherited eye diseases in purebred dogs. Williams &Wilkins 1989;149-150.

2. Chaudieu G. Chahory S. Affections oculaires héréditaires ou à prédisposition raciale chez le chien.2nd ed. Ed. du Point Vétérinaire 2013;164-165.

3. Acland GM, Ray K, Mellersh CS, Gu W, Langston AA, Rine J, Ostrander EA, Aguirre GD. Linkage analysis and comparative mapping of canine progressive rod-cone degeneration (prcd) establishes potential locus homology with retinitis pigmentosa (RP17) in humans. Proceeding of the Natlional Academy of Sciences of the United States of America (1998): 95, 3048–3053.

4. Acland GM, Ray K, Mellersh CS, Landston AA, Rine J, Ostrander EA, Aguirre GD. A novel retinal degeneration locus identified by linkage and comparative mapping of canine early retinal degeneration. Genomics (1999) 59, 134–142.

5. Zangerl B, Goldstein O, Philp AR, Lindauer SJ, Pearce-Kelling SE, Mullins RF, Graphodatsky AS, Ripoll D, Felix JS, Stone EM, Acland GM, Aguirre GD. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans.Genomics (2006) 88(5):551-63

Cataratta (ereditaria)

- Nel cane spesso cataratte genetiche; molte razze colpite, età variabile; evolutive e non evolutive; sempre **esclusione** dalla riproduzione!;
- 160 razze con sospetta ereditarietà (tratto autosomico recessivo semplice);
- solo un gene identificato (**HSF4**).

- Nell'*uomo* la maggior parte delle cataratte ereditarie sono *congenite*, mentre nel cane compaiono prevalentemente in età giovanile o adulta.

- **Pastore Australiano, Bernese, Boxer, Bassotto, Barboncino, E. Cocker S., Setter Inglese, Siberian H., Yorkshire; Boston T. ;**

Ereditarietà: HSF4

- **Staffordshire Bull Terrier**: autosomico recessivo;
- **Boston Terrier**: autosomico recessivo (f. giovanile);
- **Bouledogue Francese**: autosomico recessivo;
- Test genetico disponibile (HSF4-1);
- **Pastore Australiano**: test genetico disponibile (HSF4-2), ma esistono anche soggetti con mutazioni diverse (sono affetti, ma con test negativo!) (al contrario possibile anche cani con test positivo, ma senza sviluppo della malattia!).
- “””NON ESISTONO TEST GENETICI PER LA CATARATTA PER SCHNAUZER E PINSCHER””
- **Non riprodurre** soggetti malati!

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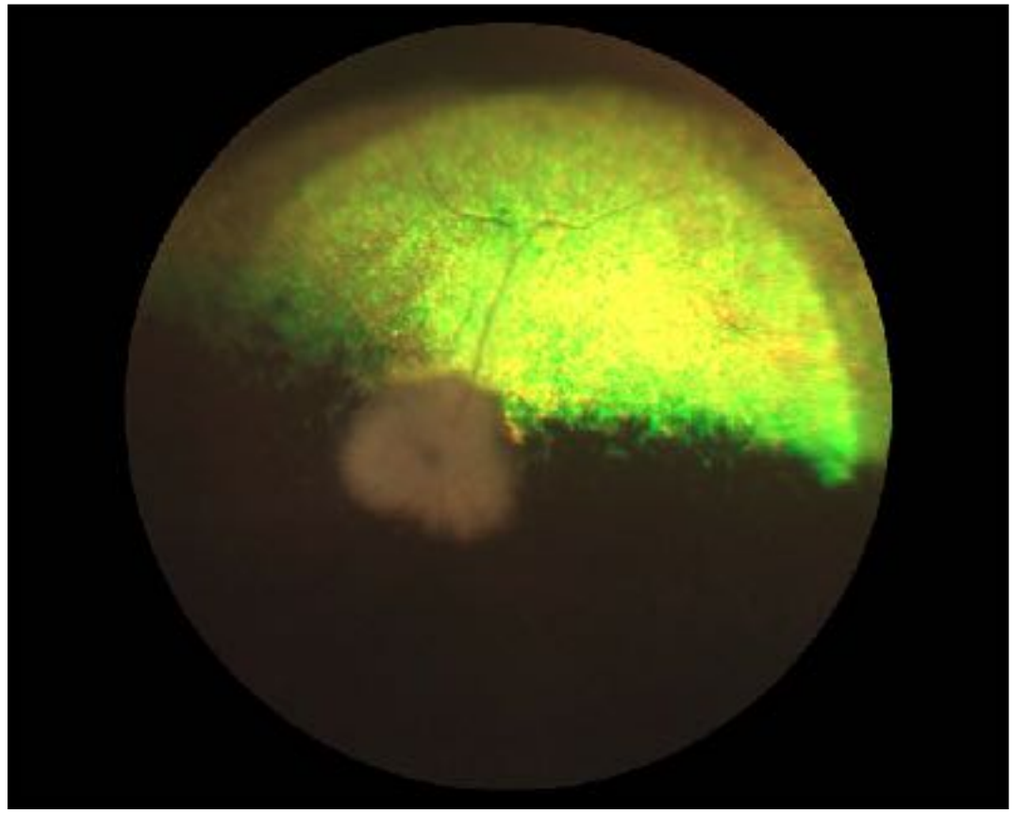
PRA

- Progressive retinal atrophy: termine generico che comprende le **degenerazioni retiniche ereditarie**
- Malattie descritte in più di 100 razze canine (quadri clinici simili, ma diversa trasmissione, insorgenza e patogenesi);
- **PRA**: malattie di sviluppo (displasie) e malattie degenerative dei fotorecettori; quasi sempre cecità totale, **sempre esclusione** dalla riproduzione (soggetti malati, genitori e figli!);
- Per alcune forme in alcune razze sono disponibili i **test genetici**.

Insorgenza precoce:

- Iniziale arresto di sviluppo dei fotorecettori, seguito da rapida degenerazione (displasia dei fotorecettori, es. rcd);

- Trasmissione autosomica recessiva;
- Alterazioni oftalmoscopiche precoci.



PRA Akita, 2aa

Iniziale deficit visivo notturno:

- *Rod-cone Dysplasia Type 1* (rcd1): ***Setter Irlandese*** (PDE6B) e ***sloughi***;

  Cecità notturna a 6-8 sett., completa ad 1 anno;
- *Rod-cone Dysplasia Type 2* (rcd2): ***Collies*** (simile al precedente, ma per mutazione di un gene diverso);

  Cecità notturna a 6 sett., completa entro 1-2 anni;
- *Rod-cone Dysplasia Type 3* (rcd3): **Cardigan Welsh Corgi** (PDE6A);
- Test genetici (Optigen);
- (Possibili forme diverse di malattia in altre razze).

Insorgenza tardiva:

RIESENSCHNAUZER

- *Progressive Rod-cone Degeneration* (prcd): notevole variabilità nell'età di comparsa, nella progressione, nel grado e nella sede delle lesioni retiniche tra e nelle razze colpite;
- Rappresentano le forme più comuni;

- **Degenerazione** (tardiva) prima dei bastoncelli e poi dei coni;
- Di solito alterazioni oftalmoscopiche tardive;

- Maggior parte: trasm. autosomica recessiva.

- ***Barboncino nano e toy***: cecità notturna a 3-5 anni, cecità completa a 5-7 anni (spesso associata a cataratta secondaria);

- ***Cocker inglese***: segni clinici tra 4-8 anni;
- ***Cocker americano***: cecità notturna a 3-5 anni, completa a 6-8;

- ***Labrador R***.: cecità notturna a 4-6 anni, completa a 6-8;

(Presenti forme diverse di malattia in altre razze).

Ereditarietà e genetica

- ***Prcd (PRA)***: tratto autosomico recessivo;

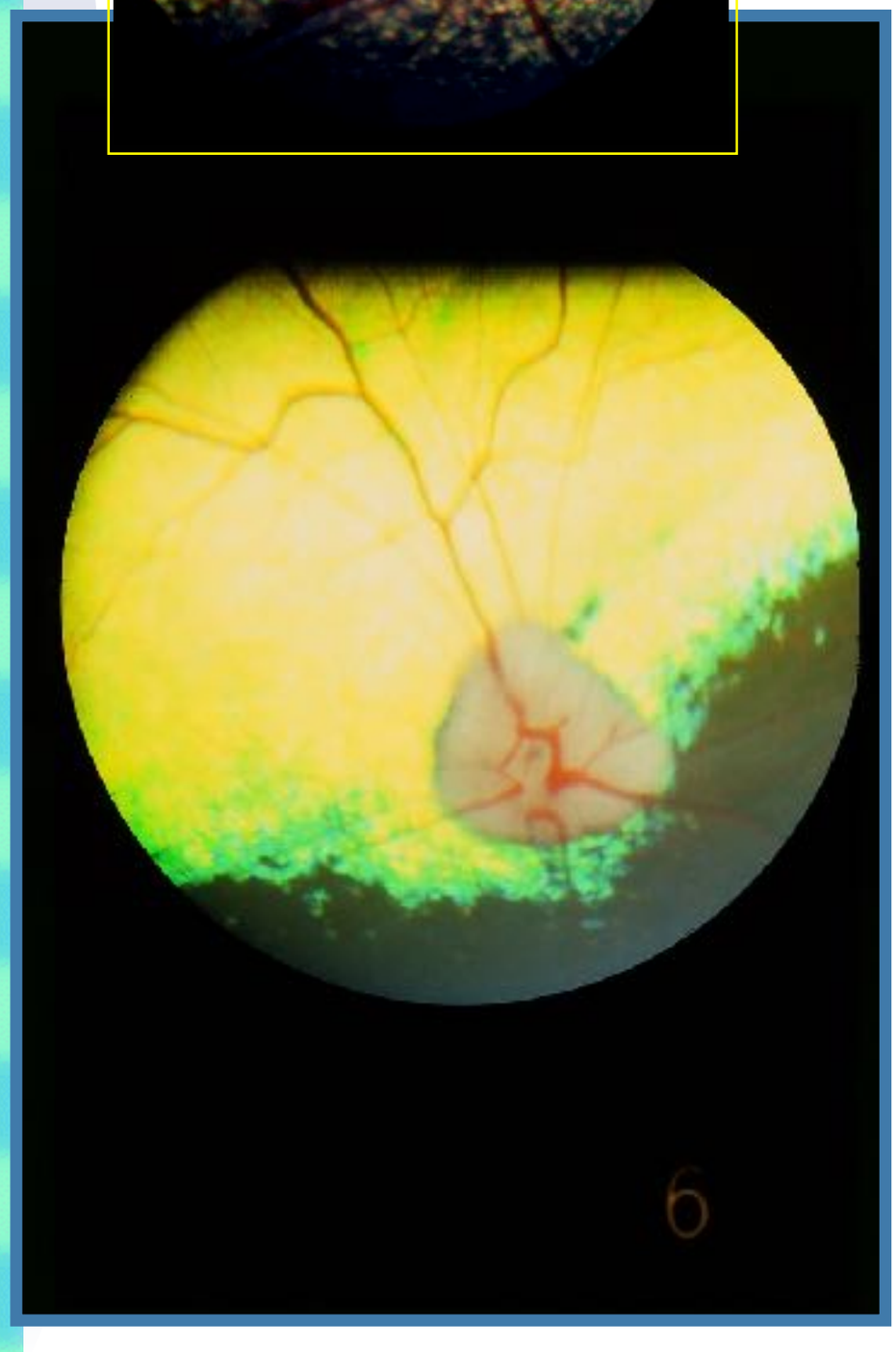
- ***Soggetto "Normale"***: OMOZIGOTE (due geni normali);
- ***Soggetto "Portatore"***: ETEROZIGOTE (un gene normale e uno malato);

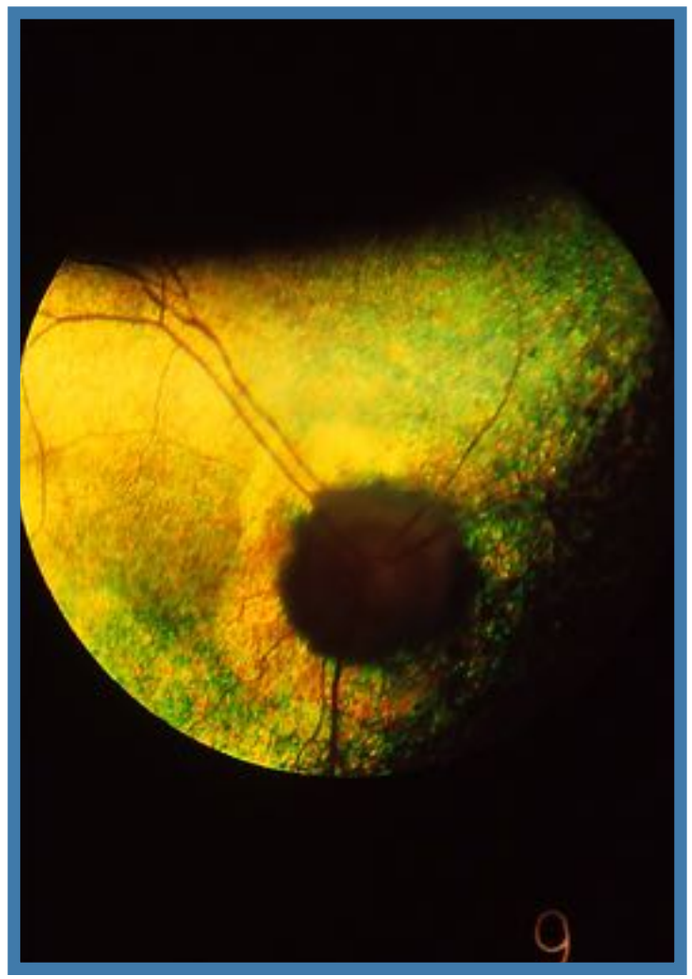
- ***Soggetto "Affetto"***: OMOZIGOTE MUTANTE (due geni malati).
- TEST PER RIESENSCHNAUZER OFFERTO DA OPTIGEN (VETOGENE)

Possible results using the OptiGen *prcd* test			
Genotype	Risk Group	Significance For Breeding	Risk of *prcd* Disease
Homozygous Normal	Normal/Clear	Can be bred to any dog, extremely low risk of producing affecteds	Extremely low
Heterozygous	Carrier	Should be bred only to Normal/ Clear to remove risk of producing affecteds	Extremely low
Homozygous Mutant	Affected	Should be bred only to Normal/ Clear to remove risk of producing affecteds	Very high

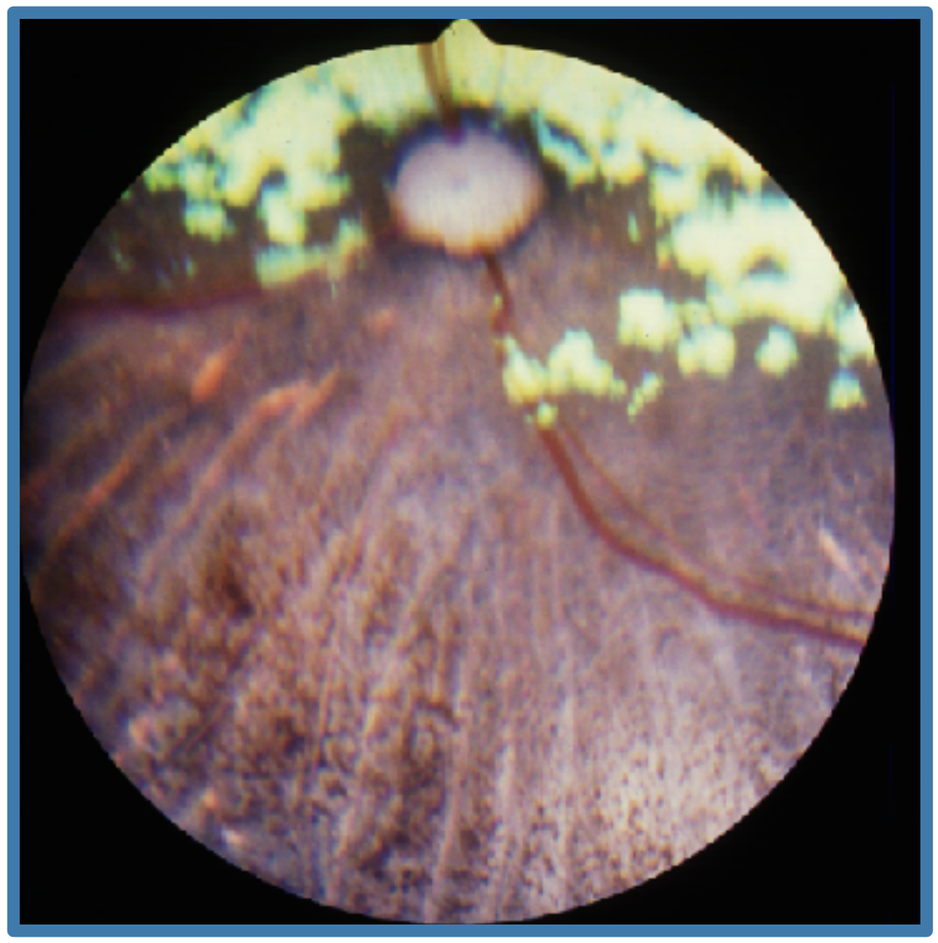
(da campione di sangue)

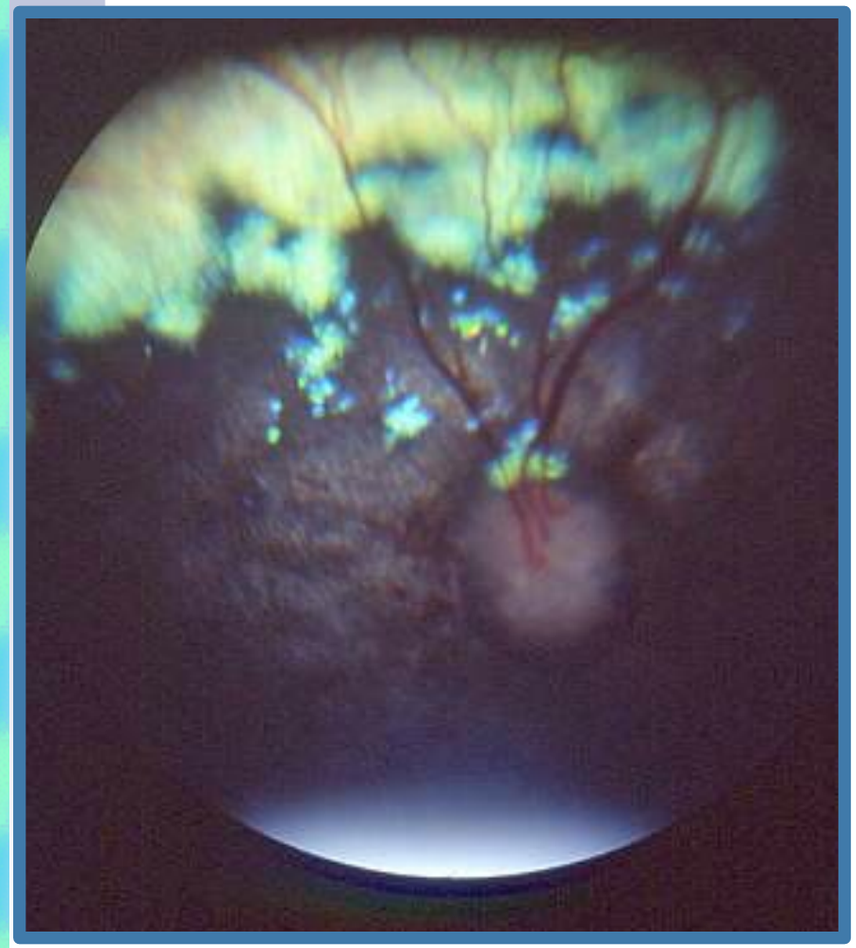
PRA

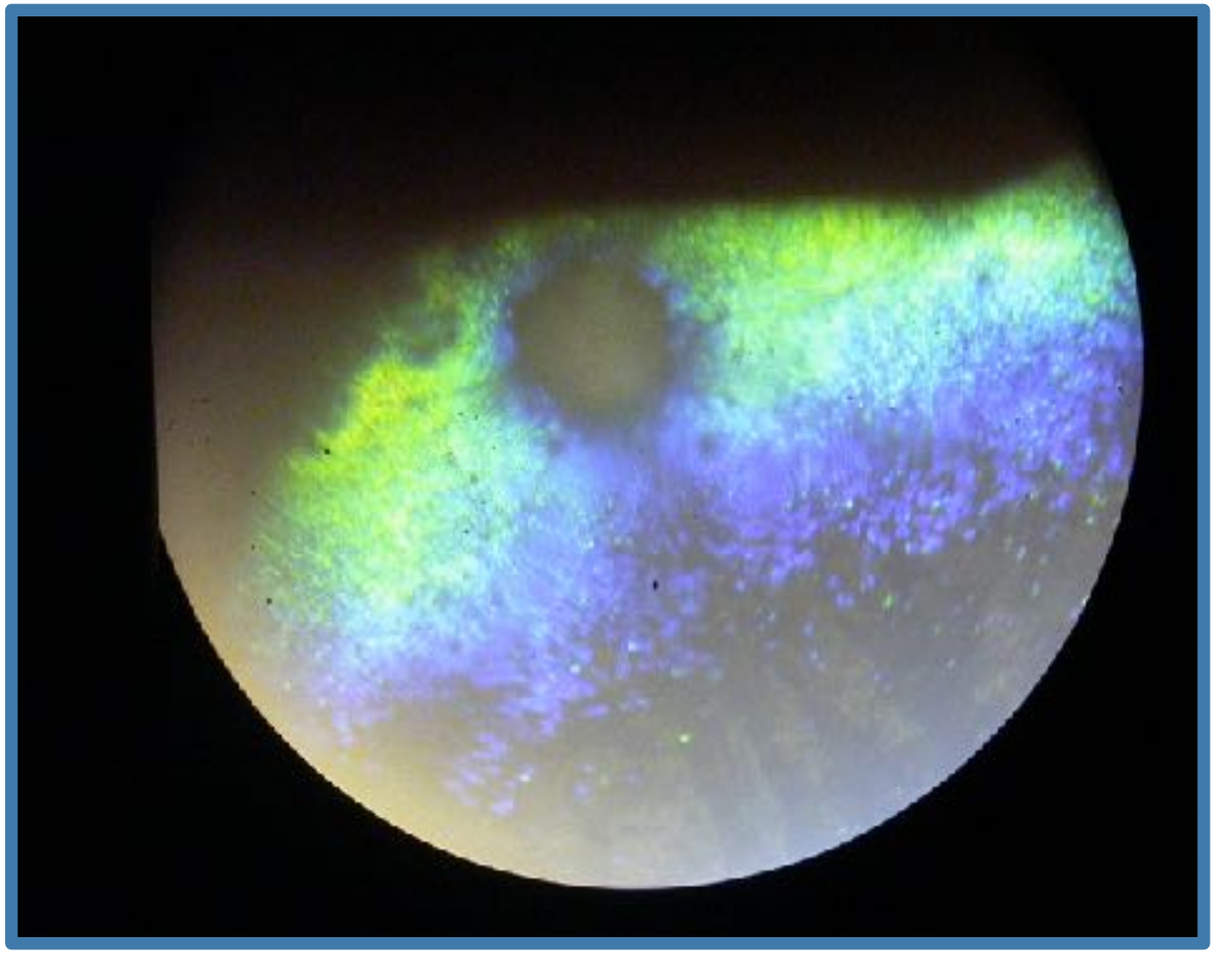




PRA







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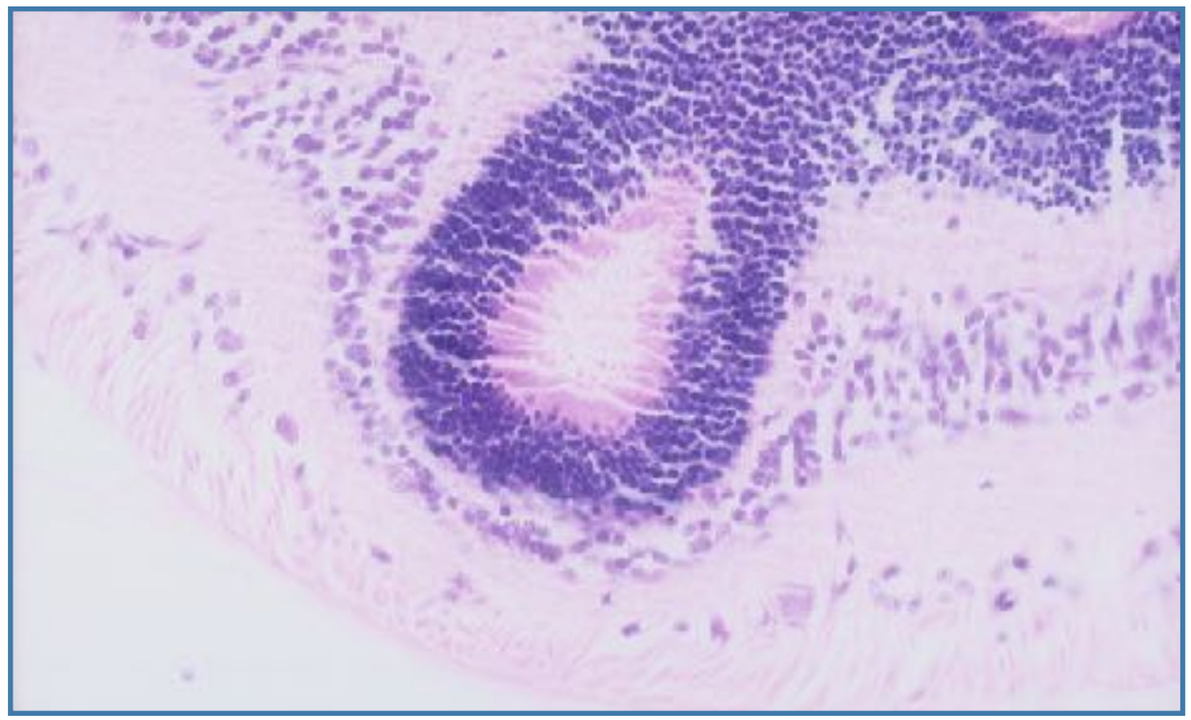
Displasia retinica

- Anomala differenziazione retinica con proliferazione di alcuni suoi elementi; formazione di “rosette” (rosettes) con/senza “pieghe” (folds);
- Tre forme cliniche;
- Ereditarietà in molte razze (modalità?);

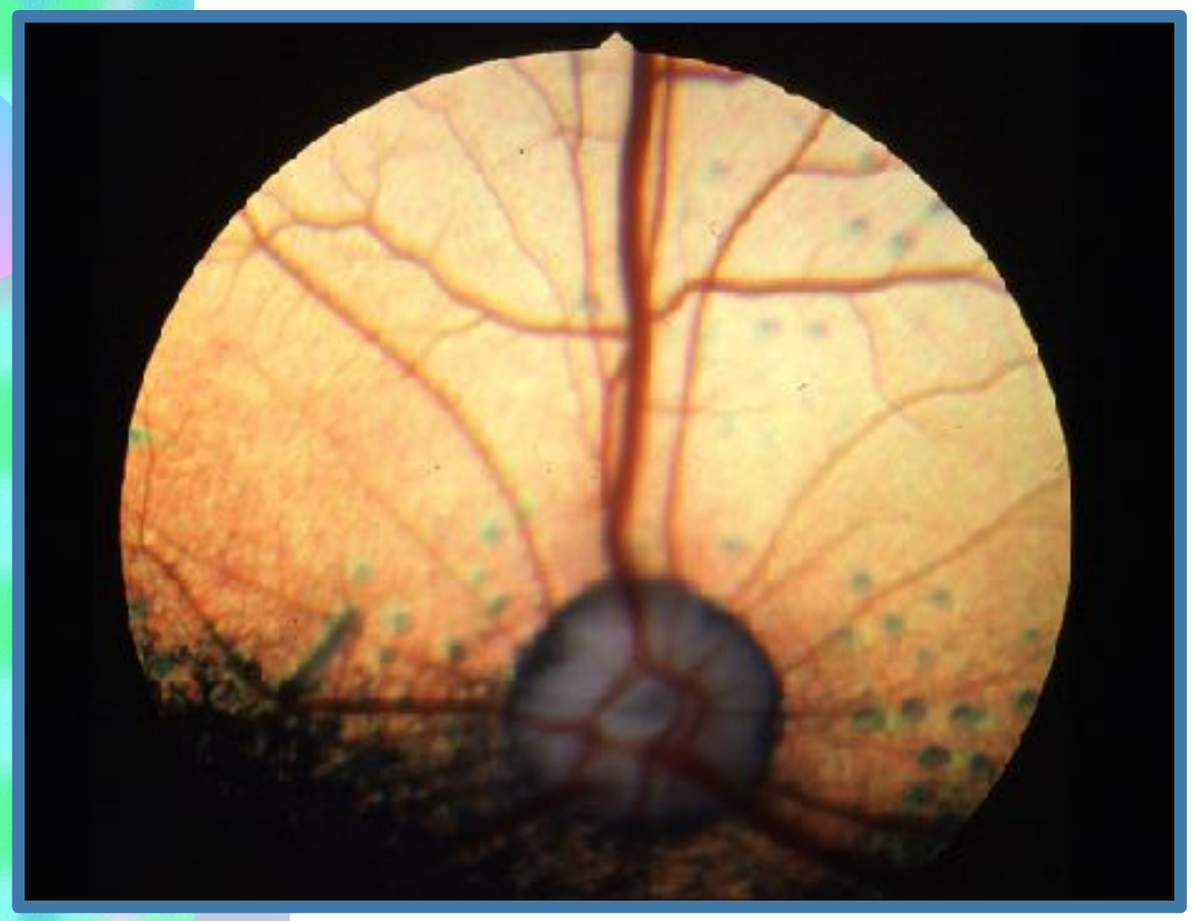
- “Rosette”: vedi descrizione istopatologica.

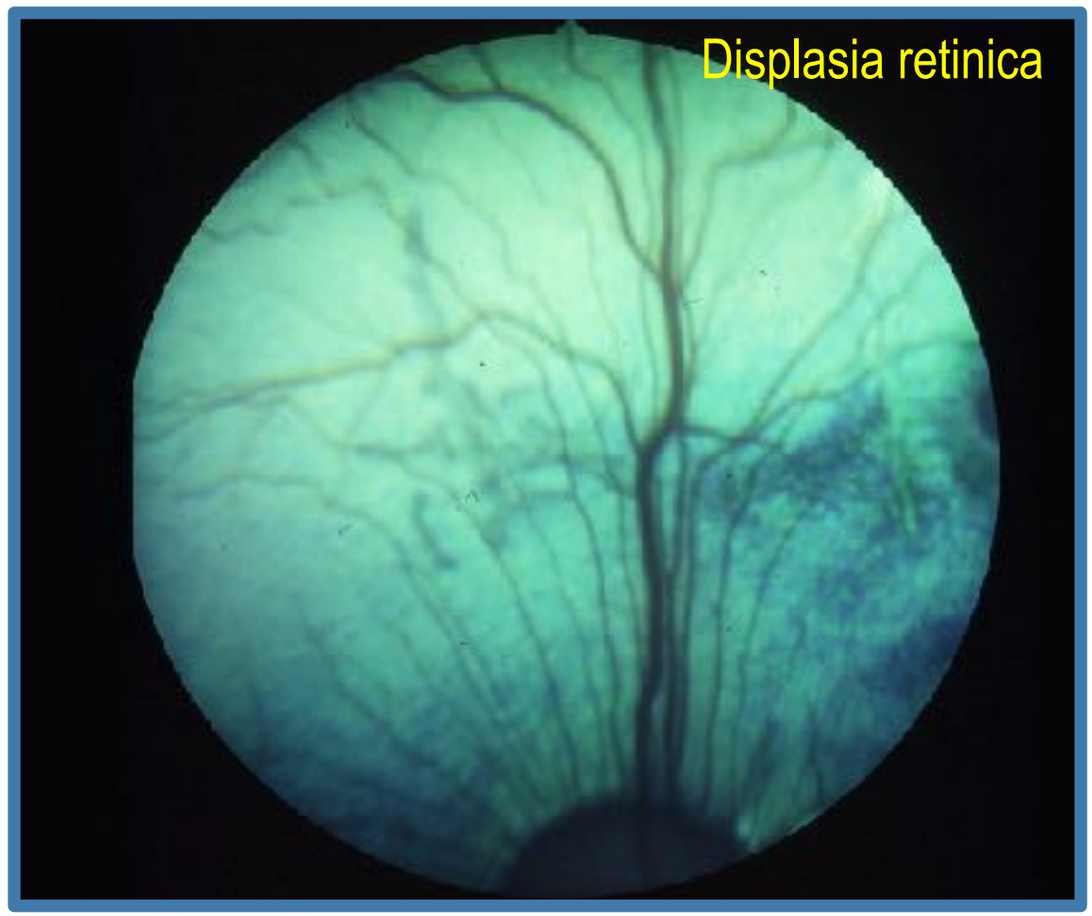


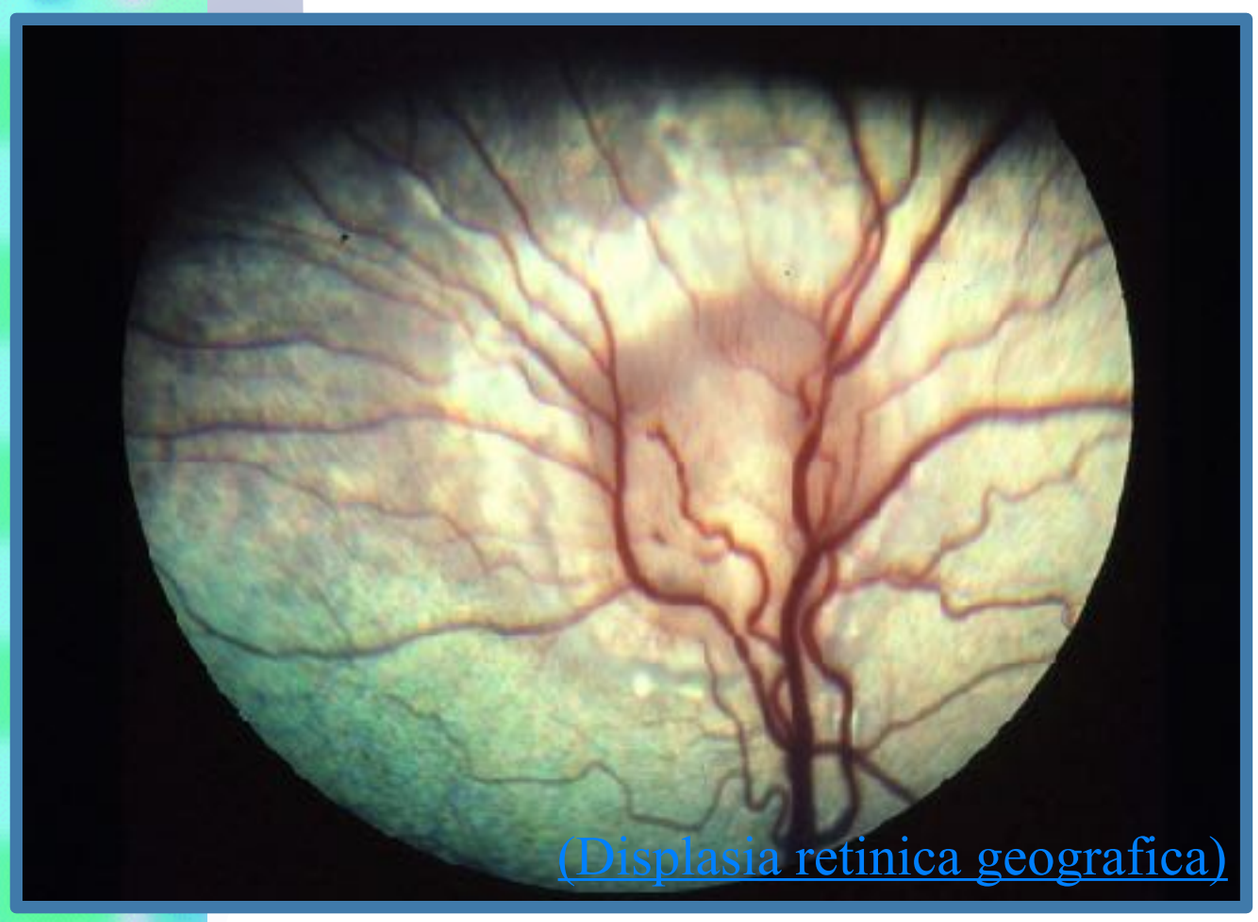
(Displasia retinica)

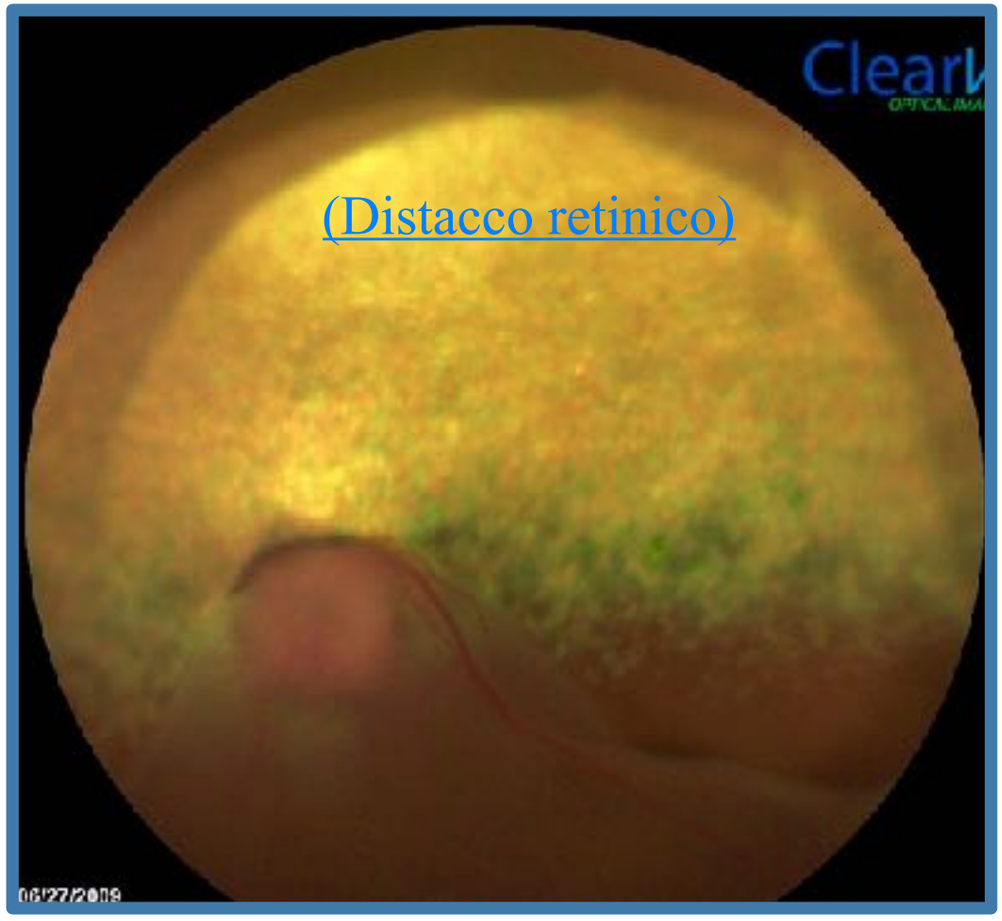
- “Retinal folds”: strie bianche o grigie singole o multiple, rotonde, ovali, triangolari, lineari o curvilinee, a V o Y uni o bilaterali, zona tappetale o non tappetale (displasia focale o multifocale; 30 razze);
- “Forma geografica”: area irregolare con zone retiniche più sottili insieme a zone di retina più spessa (displasia geografica; 11 razze);
- “Forma totale o completa” (displasia retinica completa con distacco o “non attacco”).

Pastore australiano (8 sett)




Displasia retinica


(Displasia retinica geografica)


(Distacco retinico)
06/27/2019

- Displasia focale/multifocale;

Breeder Option! (ma in alcuni casi NO)

Labrador R.

Golden R.

Rottweiler

Beagle

Cavalier KC Spaniel

Springer Spaniel I.

- Displasia geografica;

NO Breeding
+/-

Labrador R.

Golden R.

Cavalier KC Spaniel

Springer Spaniel I.

- Displasia “completa”; NO Breeding!

Akita

Chow Chow

Pastore Australiano

Dobermann

Bedlington T.

Schnauzer nano

- A volte difficile interpretazione delle f. focali (non definita la relazione con forme più gravi);
- ***E. Springer S., Labrador, Samoiedo:*** nelle forme gravi preferibile esclusione dalla riproduzione!

Cane Corso

- Forme primarie: **ereditarie** (più comuni);
- *Test genetico* per alcune forme (RD-OSD); ""QUESTO TEST E' INUTILE PER LE NOSTRE RAZZE; QUINDI NON ESISTONO TEST GENETICI PER LE RAZZE SCHNAUZER E PINSCHER""

- Cause delle rare forme secondarie: infiammazioni (virus), radiazioni, farmaci, traumi fetali, carenza vitamina A, tossicità in gravidanza.

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Glaucoma Primario

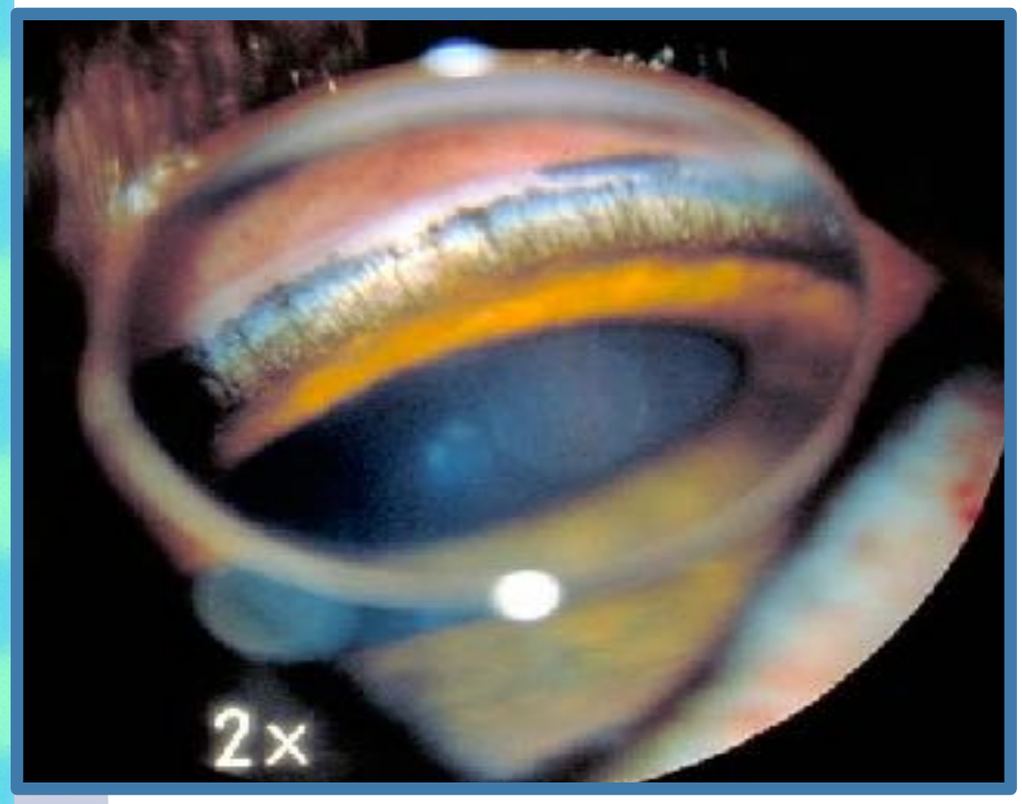
- Definizione: Il glaucoma è una progressiva neuropatia del nervo ottico caratterizzata dalla morte delle cellule gangliari (apoptosi); si accompagna ad un aumento della pressione intraoculare (IOP) con diminuzione del campo visivo e cecità (spesso permanente).

Cocker I.

- Classificazione: Ad angolo aperto (POAG)(raro), stretto o chiuso (PCAG)(frequente);
- Numerose razze predisposte (58);
- Ereditarietà: attualmente informazioni molto limitate;
- Soggetti affetti: sempre **esclusione** dalla riproduzione!

- PCAG nel ***American e English Cocker S., Basset H.; Alano, Barbone, Chow, Golden R., Samoiedo, Siberian H., Sharpei e Welsh Springer S., etc.***

- PCAG nel ***Basset H., Flat Coated R., Shiba Inu, etc.*** *(con anomalie del legamento pettinato)*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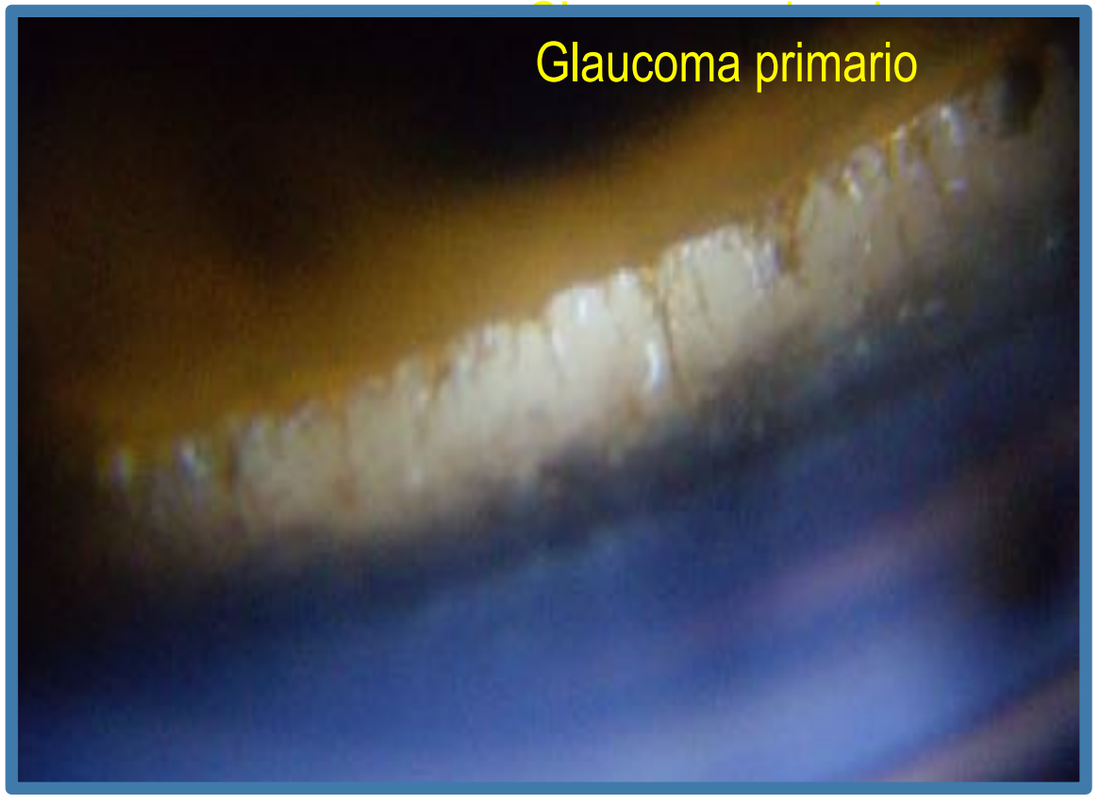
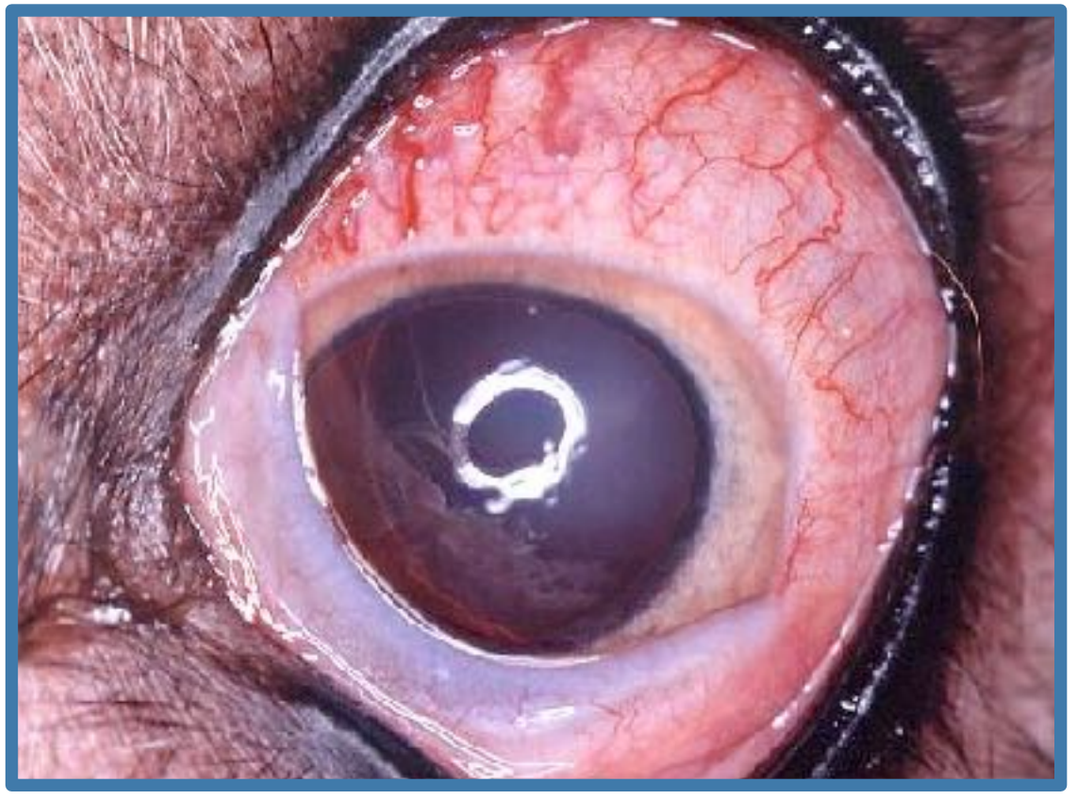

Glaucoma primario



Glaucoma primario

Other ocular disorders (reported)

	Diagnosis	Source
A	Eversion of the cartilage of the nictitating membrane	ACVO genetics committee
B	Corneal dystrophy -stromal	ACVO genetics committee
C	Primary hyperplastic tunica vasculosa lentis/ Primary hyperplastic primary vitreous (PHTVL/PHPV)	ACVO genetics committee
D	Entropion	French National Panel
E	Persistent pupillary membranes -iris to iris	ACVO genetics committee

References

1. Rubin LF. Inherited eye diseases in purebred dogs. Williams &Wilkins 1989;149-150.

2. Chaudieu G. Chahory S. Affections oculaires héréditaires ou à prédisposition raciale chez le chien.2nd ed. Ed. du Point Vétérinaire 2013;164-165.

3. Acland GM, Ray K, Mellersh CS, Gu W, Langston AA, Rine J, Ostrander EA, Aguirre GD. Linkage analysis and comparative mapping of canine progressive rod-cone degeneration (prcd) establishes potential locus homology with retinitis pigmentosa (RP17) in humans. Proceeding of the Natlional Academy of Sciences of the United States of America (1998): 95, 3048–3053.

4. Acland GM, Ray K, Mellersh CS, Landston AA, Rine J, Ostrander EA, Aguirre GD. A novel retinal degeneration locus identified by linkage and comparative mapping of canine early retinal degeneration. Genomics (1999) 59, 134–142.

5. Zangerl B, Goldstein O, Philp AR, Lindauer SJ, Pearce-Kelling SE, Mullins RF, Graphodatsky AS, Ripoll D, Felix JS, Stone EM, Acland GM, Aguirre GD. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans.Genomics (2006) 88(5):551-63

EVERSIONE CARTILAGINE TERZA PALPEBRA

Other ocular disorders (reported)

	Diagnosis	Source
A	Eversion of the cartilage of the nictitating membrane	ACVO genetics committee
B	Corneal dystrophy -stromal	ACVO genetics committee
C	Primary hyperplastic tunica vasculosa lentis/ Primary hyperplastic primary vitreous (PHTVL/PHPV)	ACVO genetics committee
D	Entropion	French National Panel
E	Persistent pupillary membranes -iris to iris	ACVO genetics committee

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3. Acland GM, Ray K, Mellersh CS, Gu W, Langston AA, Rine J, Ostrander EA, Aguirre GD. Linkage analysis and comparative mapping of canine progressive rod-cone degeneration (prod) establishes potential locus homology with retinitis pigmentosa (RP17) in humans. Proceeding of the Natlional Academy of Sciences of the United States of America (1998): 95, 3048–3053.

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Distrofie corneali

- Distrofie **stromali**: difetto metabolico di accumulo di lipidi nello spessore corneale; molte razze colpite, età tra 6 mesi e 6 anni; ereditaria e bilaterale;
- ***Beagle, Siberian H., Shetland sheepdog, Cavalier K.C.S., Airedale T., R. Collie, A.Cocker S***.

- **Riproduzione opzionale!**

- ***Siberian H.***: deposito lipidico nello stroma posteriore che poi si estende (d. Stromale) progressivamente alla parte più anteriore; gene recessivo ad espressione variabile; segni clinici iniziali tra i 5 ed i 27 mesi;

- Nei soggetti affetti: **Riproduzione opzionale** (in Europa!)

Other ocular disorders (reported)

	Diagnosis	Source
A	Eversion of the cartilage of the nictitating membrane	ACVO genetics committee
B	Corneal dystrophy -stromal	ACVO genetics committee
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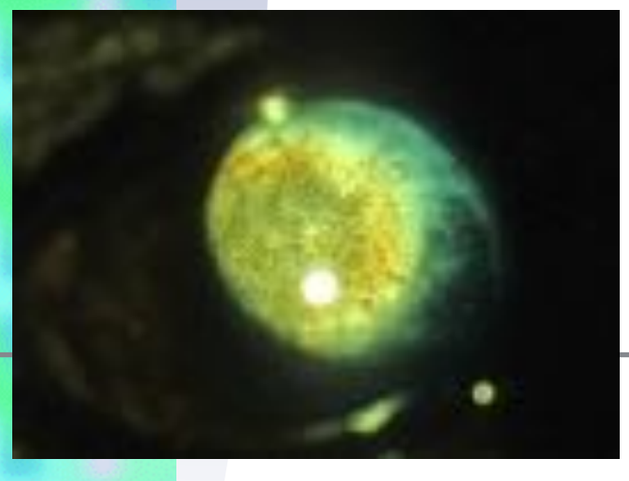
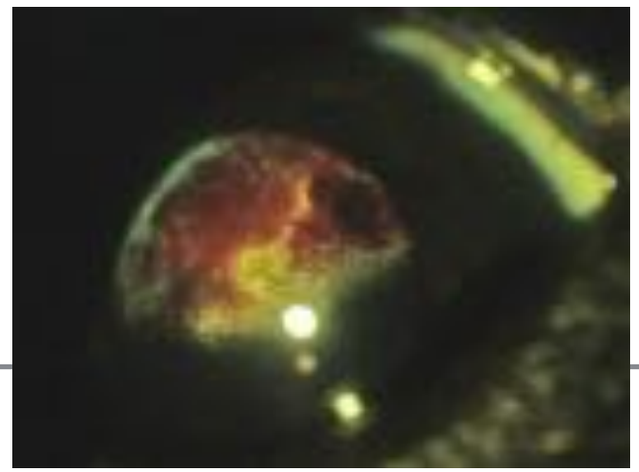
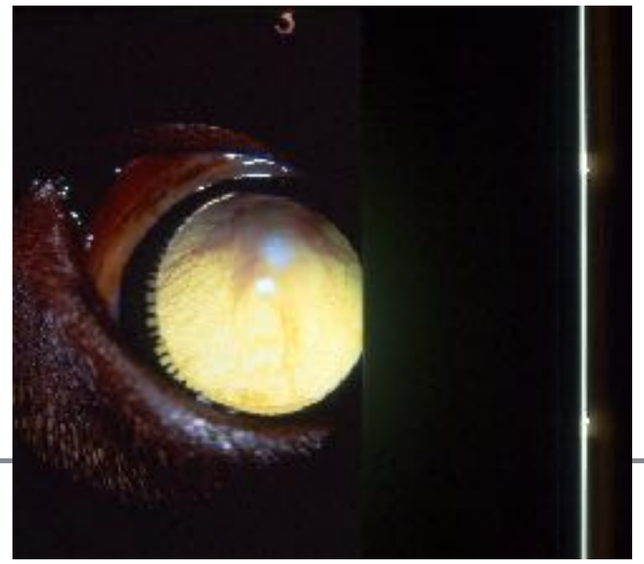
PHPV/PHTVL

Dobermann e Staffordshire Bull Terrier
6 gradi
Ereditarietà presunta dominante a penetranza incompleta
Altre razze (monolaterale)







Other ocular disorders (reported)

	Diagnosis	Source
A	Eversion of the cartilage of the nictitating membrane	ACVO genetics committee
B	Corneal dystrophy -stromal	ACVO genetics committee
C	Primary hyperplastic tunica vasculosa lentis/ Primary hyperplastic primary vitreous (PHTVL/PHPV)	ACVO genetics committee
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1. Rubin LF. Inherited eye diseases in purebred dogs. Williams &Wilkins 1989;149-150.

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5. Zangerl B, Goldstein O, Philp AR, Lindauer SJ, Pearce-Kelling SE, Mullins RF, Graphodatsky AS, Ripoll D, Felix JS, Stone EM, Acland GM, Aguirre GD. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans.Genomics (2006) 88(5):551-63

ENTROPION

Other ocular disorders (reported)

	Diagnosis	Source
A	Eversion of the cartilage of the nictitating membrane	ACVO genetics committee
B	Corneal dystrophy -stromal	ACVO genetics committee
C	Primary hyperplastic tunica vasculosa lentis/ Primary hyperplastic primary vitreous (PHTVL/PHPV)	ACVO genetics committee
D	Entropion	French National Panel
E	Persistent pupillary membranes -iris to iris	ACVO genetics committee

References

1. Rubin LF. Inherited eye diseases in purebred dogs. Williams &Wilkins 1989;149-150.

2. Chaudieu G. Chahory S. Affections oculaires héréditaires ou à prédisposition raciale chez le chien.2nd ed. Ed. du Point Vétérinaire 2013;164-165.

3. Acland GM, Ray K, Mellersh CS, Gu W, Langston AA, Rine J, Ostrander EA, Aguirre GD. Linkage analysis and comparative mapping of canine progressive rod-cone degeneration (prcd) establishes potential locus homology with retinitis pigmentosa (RP17) in humans. Proceeding of the Natlional Academy of Sciences of the United States of America (1998): 95, 3048–3053.

4. Acland GM, Ray K, Mellersh CS, Landston AA, Rine J, Ostrander EA, Aguirre GD. A novel retinal degeneration locus identified by linkage and comparative mapping of canine early retinal degeneration. Genomics (1999) 59, 134–142.

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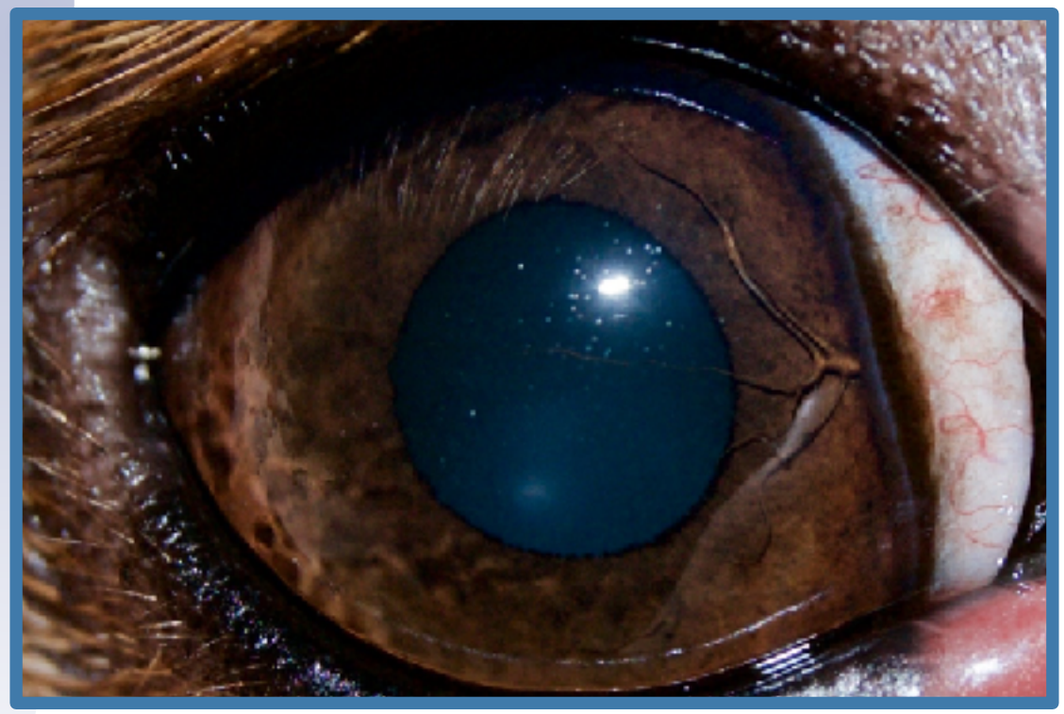
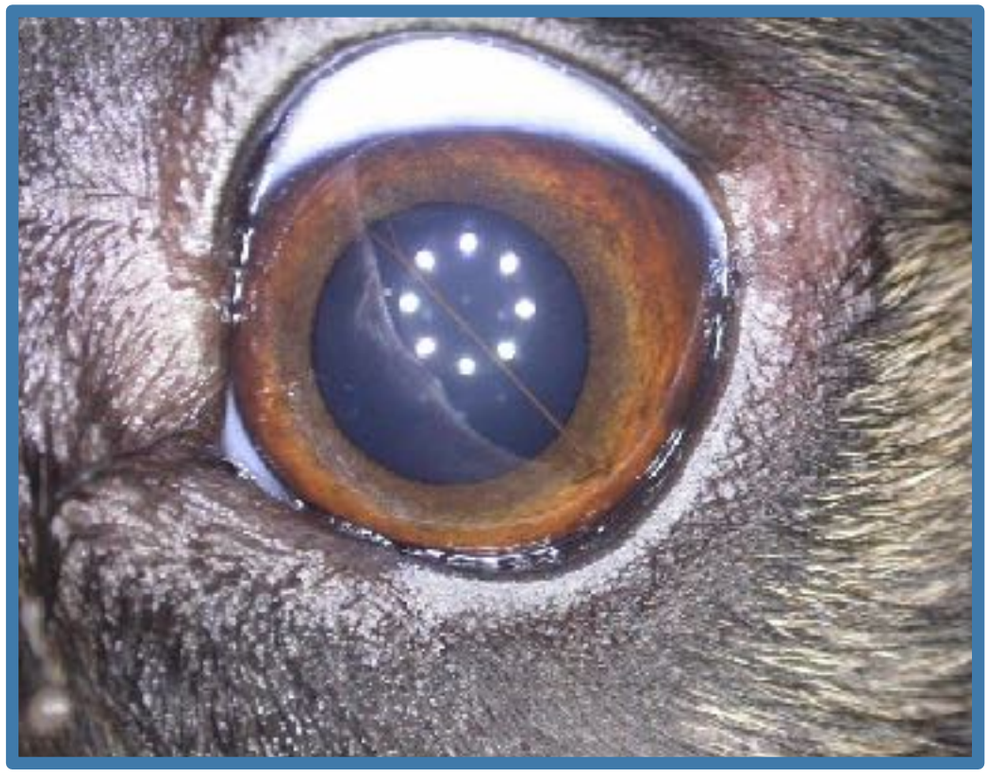
Persistenza Membrana Pupillare (PPM)

- Alta incidenza (>2-6 sett);
- Da segni clinici inosservati fino a gravi deficit visivi!
- **PPM iride-iride**;
- PPM iride-cornea;
- PPM iride-lente.





- PPM: nel ***Basenji*** ereditaria (trasmissione dominante a penetranza incompleta);
- Non riprodurre (f. gravi)!

- ***Petit Basset Griffon Vendéen, Chow, E. Cocker S., Mastiff, Airedale T., Welsh Corgi*** (predisposizione familiare).

Schnauzer (standard)	

Ocular disorders known or presumed to be inherited (published)

	Diagnosis	Description and comments specific to the breed	Inheritance	Gene/ marker test	References
A	Entropion	Inferior lateral, spastic	Unknown	NO	1
B	Eversion of the cartilage of the nictitating membrane	Close to the third eyelid margin	Unknown	NO	1
C	Kerato-conjunctivitis sicca		Unknown	NO	1
D	Cataract	1. Posterior, subcapsular/ cortical ; dogs > 1 y.o.: progressive 2. Posterior subcapsular/ Cortical dogs > 6 y.o.: slowly progressive 3. Congenital Cataract: posterior, nucleo-cortical, mostly non progressive, associated with microcornea	1. Unknown 2. Unknown 3. Autosomal Recessive	NO	1, 2

E	Persistent hyperplastic tunica vasculosa lentis/Persistent hyperplastic primary vitreous (PHTVL/PHPV)		Unknown	NO	1
F	Vitreous degeneration	Dogs > 8 y.o., bilateral asteroid hyalosis	Unknown	NO	1
G	Progressive Retinal Atrophy (PRA)	Blindness up to 4 y. o	Unknown	NO	1

The ECVO's advice relating to hereditary eye disease control

Please see ECVO Manual chapter 8: VET Advice

Recommendations regarding age and frequency for eye examinations

Please see ECVO Manual chapter 7: ECVO Age and Frequency recommendations

Other ocular disorders (reported)

	Diagnosis	Source
A	Corneal dystrophy, epithelial/stromal	ACVO Genetics Committee
B	Retinal dysplasia, focal/multifocal	ACVO Genetics Committee

References

1 C haudieu G, Chahory S. 2013. Affections oculaires héréditaires ou à prédisposition héréditaire chez le chien. Rueil-Malmaison : Ed. du Point Vétérinaire, Wolters Kluwer France.

2 Rubin LF (1987) Inherited eye diseases in purebred dogs. Williams & Wilkins, Baltimore, 283.

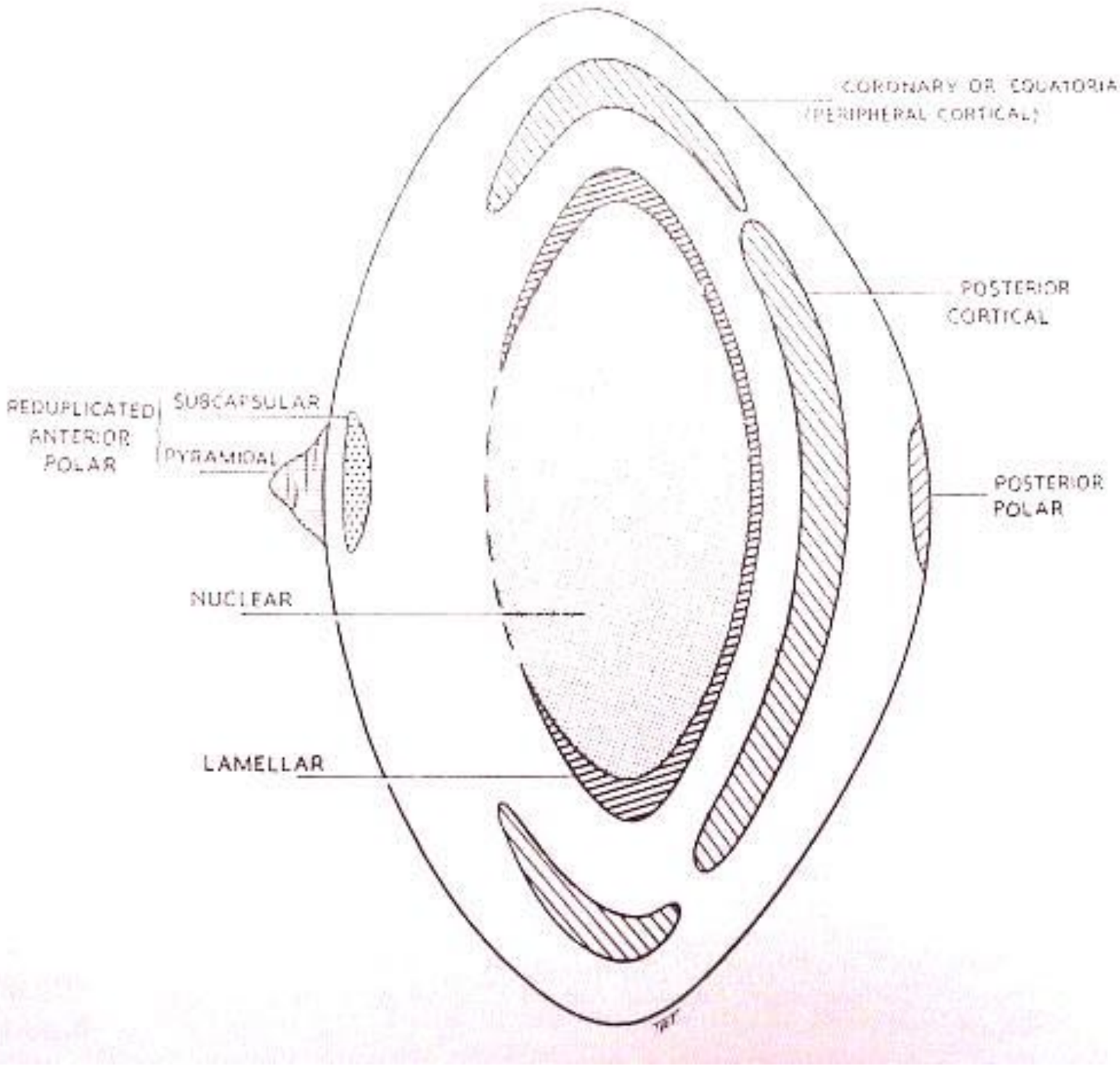
CHERATOCONGIUNTIVITE SECCA

Schnauzer (standard)	

Ocular disorders known or presumed to be inherited (published)

	Diagnosis	Description and comments specific to the breed	Inheritance	Gene/ marker test	References
A	Entropion	Inferior lateral, spastic	Unknown	NO	1
B	Eversion of the cartilage of the nictitating membrane	Close to the third eyelid margin	Unknown	NO	1
C	Kerato-conjunctivitis sicca		Unknown	NO	1
D	Cataract	1. Posterior, subcapsular/ cortical ; dogs > 1 y.o.: progressive 2. Posterior subcapsular/ Cortical dogs > 6 y.o.: slowly progressive 3. Congenital Cataract: posterior, nucleo-cortical, mostly non progressive, associated with microcornea	1. Unknown 2. Unknown 3. Autosomal Recessive	NO	1, 2

Classificazione



E	Persistent hyperplastic tunica vasculosa lentis/Persistent hyperplastic primary vitreous (PHTVL/PHPV)		Unknown	NO	1
F	Vitreous degeneration	Dogs > 8 y.o., bilateral asteroid hyalosis	Unknown	NO	1
G	Progressive Retinal Atrophy (PRA)	Blindness up to 4 y. o	Unknown	NO	1

The ECVO's advice relating to hereditary eye disease control

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Recommendations regarding age and frequency for eye examinations

Please see ECVO Manual chapter 7: ECVO Age and Frequency recommendations

Other ocular disorders (reported)

	Diagnosis	Source
A	Corneal dystrophy, epithelial/stromal	ACVO Genetics Committee
B	Retinal dysplasia, focal/multifocal	ACVO Genetics Committee

References

1 C haudieu G, Chahory S. 2013. Affections oculaires héréditaires ou à prédisposition héréditaire chez le chien. Rueil-Malmaison : Ed. du Point Vétérinaire, Wolters Kluwer France.

2 Rubin LF (1987) Inherited eye diseases in purebred dogs. Williams & Wilkins, Baltimore, 283.

Miniature Schnauzer	

Ocular disorders known or presumed to be inherited (published)

	Diagnosis	Description and comments specific to the breed	Inheritance	Gene/ marker test	References
A	Myopia	Affects up to 40% of dogs; mean refractive error of myopic schnauzers is -1.4D	Unknown	NO	1, 2
B	Keratoconjunctivitis Sicca	Relative risk = 1.8; low meibomian gland production may predispose to KCS	Unknown	NO	3-6
C	Cataract	1. Congenital with posterior lenticonus and microphthalmia: Nucleus and posterior cortex; globe and lens size reduced 10-20%; lenticonus in 20% of cataracts 2. Juvenile posterior cortex: Age of onset 6+ months	1.Presumed autosomal recessive 2. Autosomal recessive	NO	7-12 13,14
D	Ceroid lipofuscinosis	Loss of vision and neurological signs	Presumed autosomal recessive	NO	17-19

E	Progressive Retinal Atrophy (PRA)	1. PRA Type A, (Photo receptor dysplasia), uncommon. ERG and histopathological changes from 8 weeks. Punduscopic changes and visual impairment appear at 2-5 y.o.; 2. Early 2-4 y.o 3. Late-onset form	1. Presumed Autosomal recessive 2.Autosomal recessive 3. Unknown	Type A-PRA	15,16,20,
F	Retinal dysplasia with or without PHPV	Generalized dysplasia, with retinal detachment and/or PHPV in 50% of affected dogs	Autosomal recessive	NO	21,22

The ECVO's advice relating to hereditary eye disease control

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Recommendations regarding age and frequency for eye examinations

Please see ECVO Manual chapter 7: ECVO Age and Frequency recommendations

Other ocular disorders (reported)

	Diagnosis	Source
A	Microphthalmia	ACVO genetics committee
B	Distichiasis	ACVO genetics committee Finnish Kennel Club Database
C	Persistent pupillary membranes	ACVO genetics committee
D	Corneal dystrophy -epithelial/stromal	ACVO genetics committee
E	Vitreous degeneration	ACVO genetics committee

References

1. Murphy CJ, Zadnik K, Mannis MJ. Myopia and refractive error in dogs. Invest Ophthalmol Vis Sci. 1992;33(8):2459-63.

2. Kubai MA, Bentley E, Miller PE, Mutti DO, Murphy CJ. Refractive states of eyes and association between ametropia and breed in dogs. Am J Vet Res. 2008;69(7):946-51.

3. Kaswan RL, Salisbury MA. A new perspective on canine keratoconjunctivitis sicca. Treatment with ophthalmic cyclosporine. Vet Clin No Am: Sm Anim Prac 1990;20:583–613.

4. Helper LC. The tear film in the dog. Causes and treatment of diseases associated with overproduction and underproduction of tears. Anim Eye Res 1996;15, 5–11.

5. Moore CP. Diseases and surgery of the lacrimal secretory system. In: Gelatt, K.N. (Ed.), Veterinary Ophthalmology, 1999 (Third ed). Lippincott Williams & Wilkins, Philadelphia PA, USA, pp. 583–607

6. Ofri R, Orgad K, Dickstein S. Canine meibometry: Establishing baseline values for meibomian gland secretions in dogs. Vet J 2007;174:536-40.

Miniature Schnauzer	

Ocular disorders known or presumed to be inherited (published)

	Diagnosis	Description and comments specific to the breed	Inheritance	Gene/ marker test	References
A	Myopia	Affects up to 40% of dogs; mean refractive error of myopic schnauzers is -1.4D	Unknown	NO	1, 2
B	Keratoconjunctivitis Sicca	Relative risk = 1.8; low meibomian gland production may predispose to KCS	Unknown	NO	3-6
C	Cataract	1. Congenital with posterior lenticonus and microphthalmia: Nucleus and posterior cortex; globe and lens size reduced 10-20%; lenticonus in 20% of cataracts 2. Juvenile posterior cortex: Age of onset 6+ months	1.Presumed autosomal recessive 2. Autosomal recessive	NO	7-12 13,14
D	Ceroid lipofuscinosis	Loss of vision and neurological signs	Presumed autosomal recessive	NO	17-19

Retinoscopia

- Si tratta della determinazione oggettiva dell' errore refrattivo dell' occhio
- Midriasi non necessaria
- La maggioranza dei cani sono entro 1D dall' emmetropia
- Cani di piccola taglia e che vivono in casa hanno tendenza alla miopia, cani di grande taglia che vivono all' esterno hanno tendenza alla ipermetropia

SBISA

Miniature Schnauzer	

Ocular disorders known or presumed to be inherited (published)

	Diagnosis	Description and comments specific to the breed	Inheritance	Gene/ marker test	References
A	Myopia	Affects up to 40% of dogs; mean refractive error of myopic schnauzers is -1.4D	Unknown	NO	1, 2
B	Keratoconjunctivitis Sicca	Relative risk = 1.8; low meibomian gland production may predispose to KCS	Unknown	NO	3-6
C	Cataract	1. Congenital with posterior lenticonus and microphthalmia: Nucleus and posterior cortex; globe and lens size reduced 10-20%; lenticonus in 20% of cataracts 2. Juvenile posterior cortex: Age of onset 6+ months	1.Presumed autosomal recessive 2. Autosomal recessive	NO	7-12 13,14
D	Ceroid lipofuscinosis	Loss of vision and neurological signs	Presumed autosomal recessive	NO	17-19

Cataratta congenita

- *Definizione*: Opacità della lente;
- *Diagnosi*: tra la nascita e le 8 sett. di vita;
- *Eziologia*: per alterazione nello sviluppo delle fibre primarie della lente (possibile associazione ad altre anomalie);
- *Localizzazione*: opacità nucleare fetale (non evolutiva); a volte anche corticale anteriore e posteriore (unilaterale o bilaterale, evolutiva).

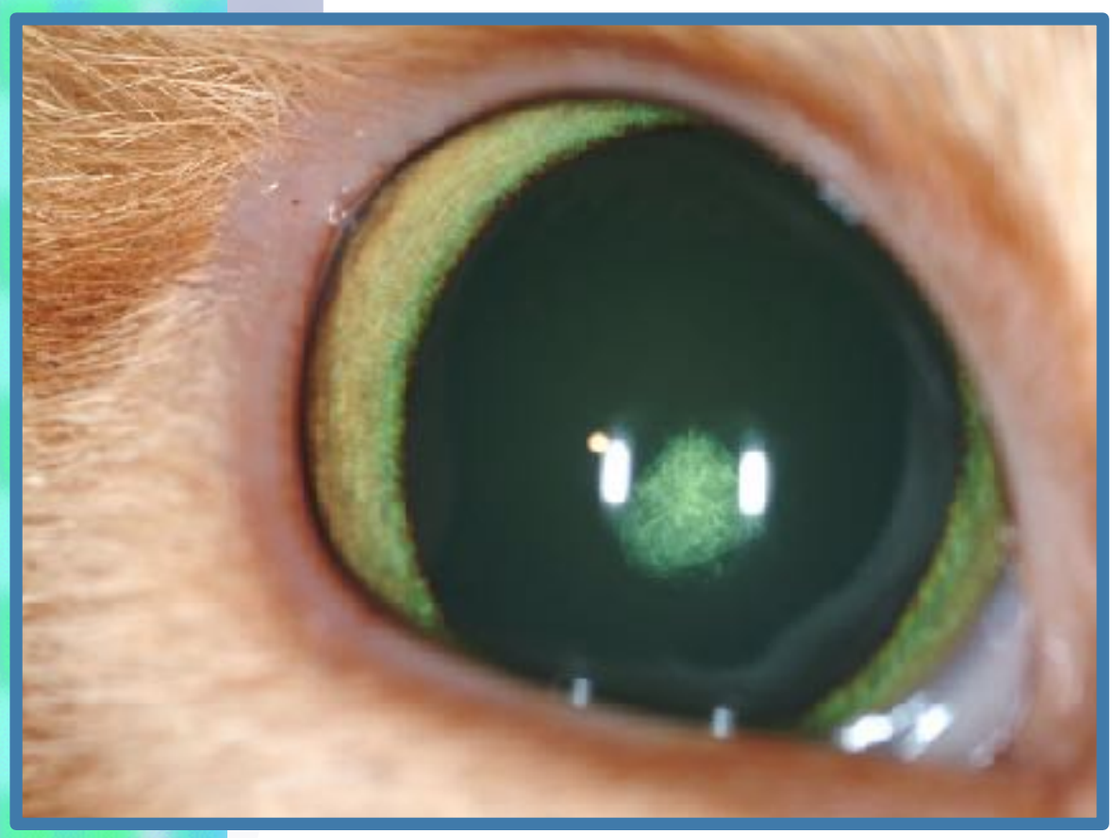
(Cataratta congenita)

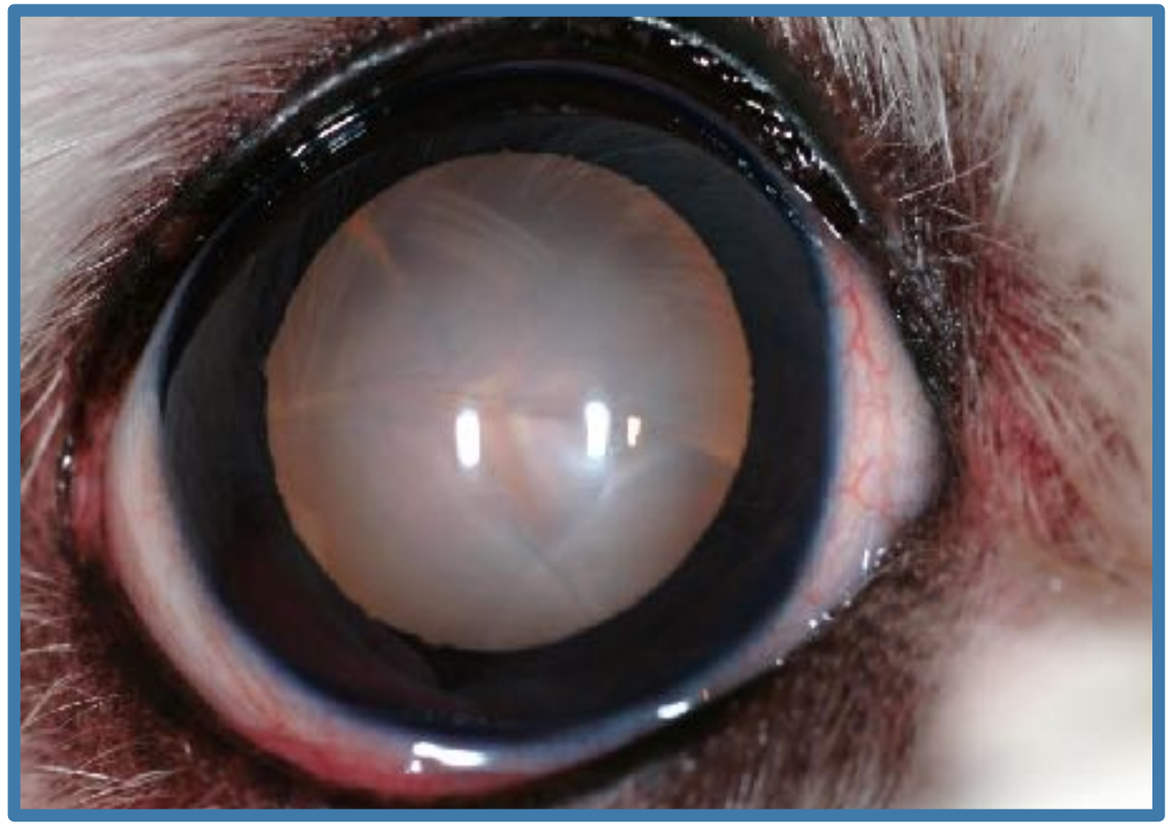
- Ereditarietà: sono considerate ereditarie tutte le cataratte mono o bilaterali, ad eccezione di quelle sicuramente associate a trauma, uveite, malattie metaboliche, tossiche, carenze nutrizionali o senili (> 7aa, 9aa, 11aa).

Schnauzer nano

- ***Schnauzer nano***: ereditarietà recessiva;
- Opacità nucleare o subcapsulare posteriore bilaterale (*rapidamente evolutiva*), spesso associata a microftalmo (lieve).

Cataratta (ereditaria)





E	Progressive Retinal Atrophy (PRA)	1. PRA Type A, (Photo receptor dysplasia), uncommon. ERG and histopathological changes from 8 weeks. Punduscopic changes and visual impairment appear at 2-5 y.o.; 2. Early 2-4 y.o 3. Late-onset form	1. Presumed Autosomal recessive 2.Autosomal recessive 3. Unknown	Type A-PRA	15,16,20,
F	Retinal dysplasia with or without PHPV	Generalized dysplasia, with retinal detachment and/or PHPV in 50% of affected dogs	Autosomal recessive	NO	21,22

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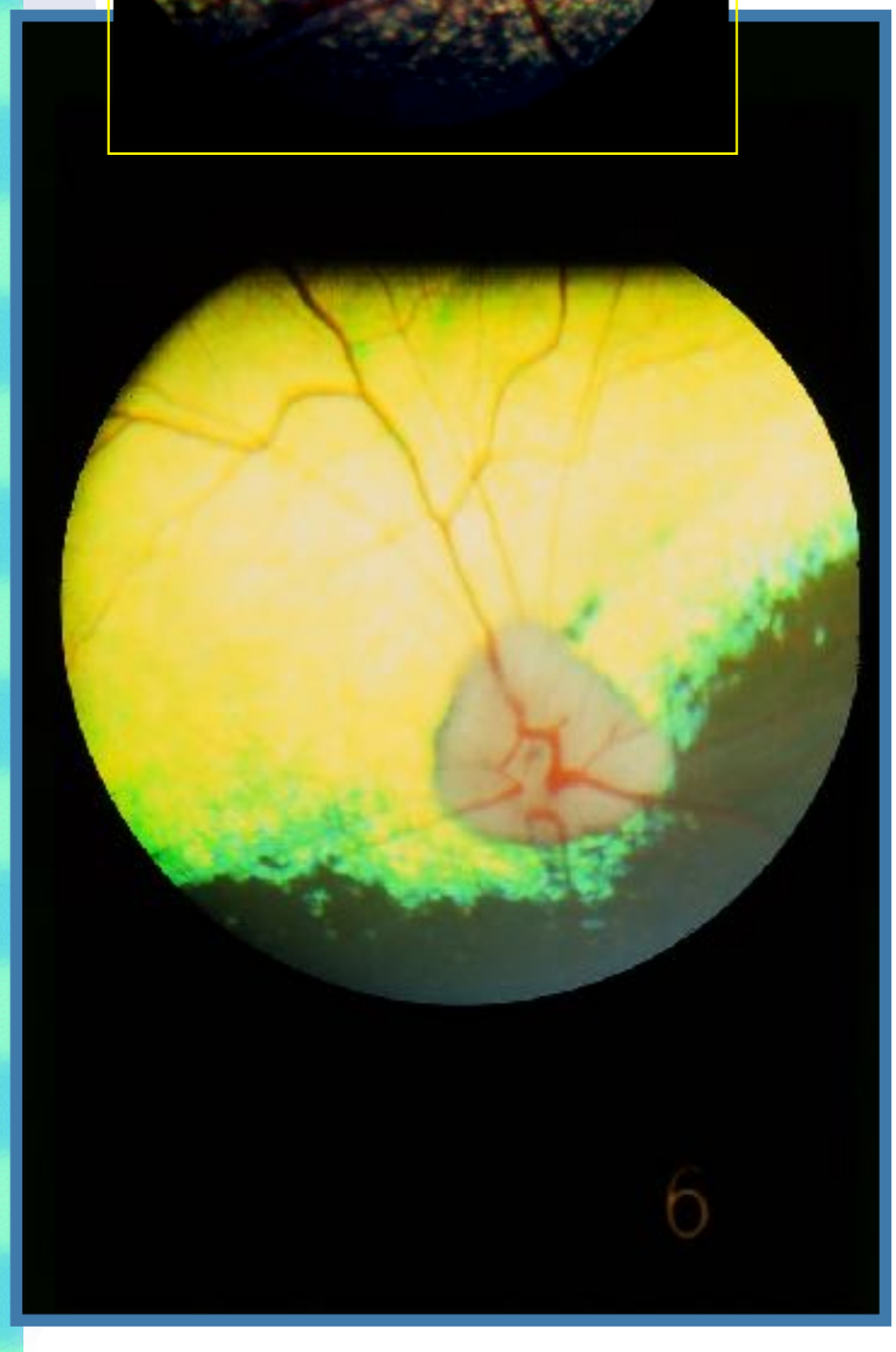
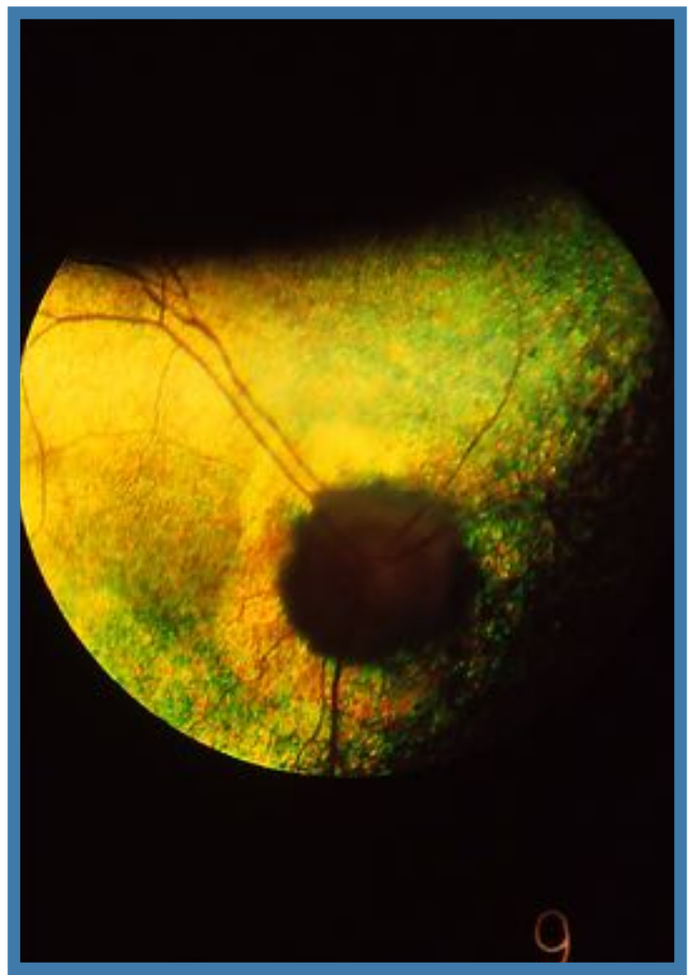
Other ocular disorders (reported)

PRA





PRA SCHNAUZER NANO

- **Type A PRA: nella popolazione di S.N. si è rivelato una mutazione molto rara probabilmente perchè ormai eradicata o presente in bassissima frequenza. NON ha senso ora testare per questo gene**
- **Il gene Fosducina (PDC) non è responsabile per la PRA di tipo A. NON USATE TEST CHE SI AVVALGANO DI QUESTA ASSERZIONE DI PARTENZA**

PRA SCHNAUZER NANO

- **TYPE B PRA: nuova variante che spiega numerosi casi di PRA nello S.N. (offerta da Optigen)**
- **Soggetti omozigoti per questa variante hanno l'86% di probabilità di sviluppare la malattia ; comunque il 14% degli omozigoti per questo gene non sviluppa la malattia entro i 7 anni forse per "penetranza" incompleta del gene o perchè la variante in oggetto è un marker che si trova solo vicino**

PRA SCHNAUZER NANO

  - Di 76 cani con PRA il 55% erano omozigoti per il Tipo B
- Dei rimanenti 45%, l'11% erano eterozigoti e il 34% erano clear per il Tipo B
- Questo significa che esiste ALMENO una ulteriore forma di PRA (oltre A e B) presente nella razza.

Expected results for breeding strategies using the Type B PRA Test			
Parent 1 Genotype	**Parent 2 Genotype**		
	Homozygous Wild Type/**Clear**	Heterozygous/ **Carrier**	Homozygous **Variant**
Homozygous Wild Type/**Clear**	All = Normal/Clear	1/2 = Clear 1/2 = Carrier	All = Carrier
Heterozygous/ **Carrier**	1/2 = Clear 1/2 = Carrier	1/4 = Clear 1/2 = Carrier 1/4 = Variant	1/2 = Carrier 1/2 = Variant
Homozygous **Variant**	All = Carrier	1/2 = Carrier 1/2 = Variant	All = Variant

Solo accoppiamenti in giallo

NON usare accoppiamenti in bianco

E	Progressive Retinal Atrophy (PRA)	1. PRA Type A, (Photo receptor dysplasia), uncommon. ERG and histopathological changes from 8 weeks. Punduscopic changes and visual impairment appear at 2-5 y.o.; 2. Early 2-4 y.o 3. Late-onset form	1. Presumed Autosomal recessive 2.Autosomal recessive 3. Unknown	Type A-PRA	15,16,20,
F	Retinal dysplasia with or without PHPV	Generalized dysplasia, with retinal detachment and/or PHPV in 50% of affected dogs	Autosomal recessive	NO	21,22

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Other ocular disorders (reported)

Veterinary Ophthalmology (2004) 7, 3, 151–158

Inherited retinal dysplasia and persistent hyperplastic primary vitreous in Miniature Schnauzer dogs

Bruce H. Grahn,* Eric S. Storey* and Catherine McMillan†

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Abstract

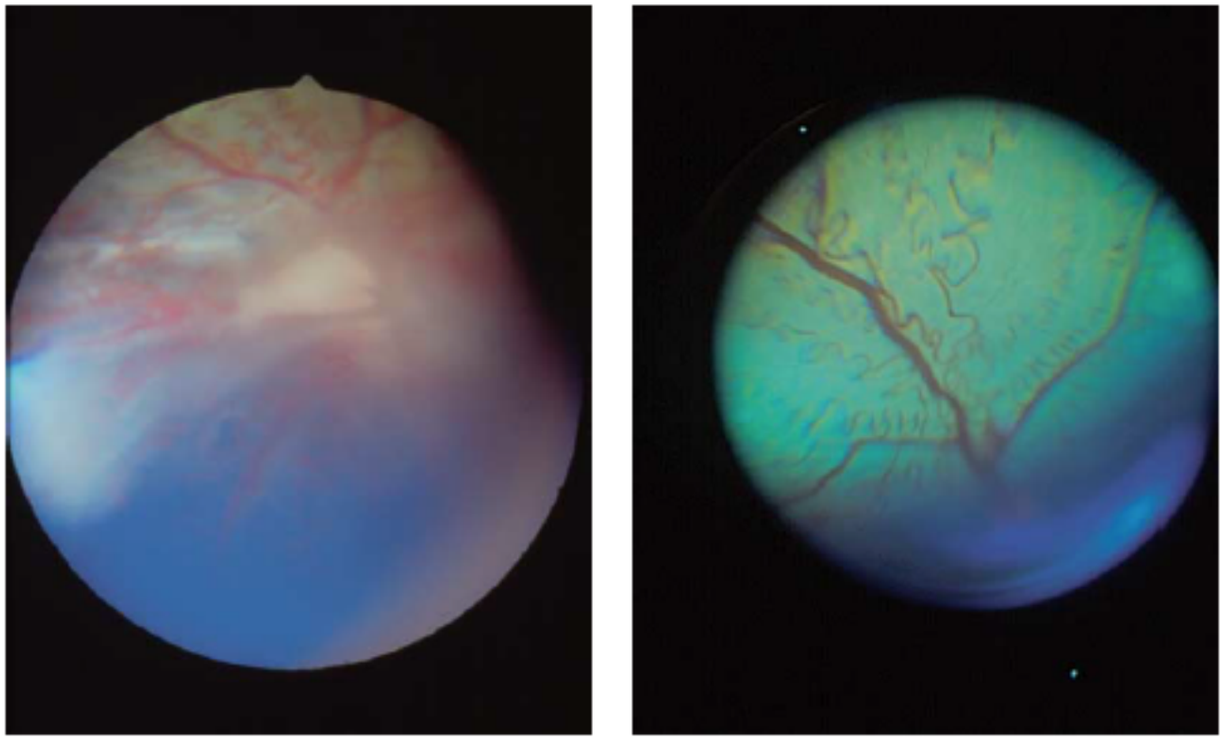
The objectives of this study were to define the clinical syndrome of retinal dysplasia and persistent primary vitreous in Miniature Schnauzer dogs and determine the etiology. We examined 106 Miniature Schnauzers using a biomicroscope and indirect ophthalmoscope. The anterior and posterior segments of affected dogs were photographed. Four enucleated eyes were examined using routine light microscopy and scanning electron microscopy. A pedigree was constructed and related dogs were test bred to define the mode of inheritance of this syndrome. Congenital retinal dysplasia was confirmed in 21 of 106 related Miniature Schnauzer dogs. Physical and postmortem examinations revealed that congenital abnormalities were limited to the eyes. Biomicroscopic, indirect ophthalmoscopic, and neuro-ophthalmic examinations confirmed that some of these dogs were blind secondary to bilateral retinal dysplasia and detachment (nonsynchronous) ($n = 15$), and the remainder had generalized retinal dysplasia ($n = 11$). Fifteen of these dogs were also diagnosed with unilateral ($n = 9$) or bilateral ($n = 6$) persistent hyperplastic primary vitreous. Nutritional, infectious, or toxic etiologies were not evident on physical, postmortem, light microscopic, or transmitting and scanning electron microscopic examination of four affected Miniature Schnauzers. We examined the pedigree and determined that an autosomal recessive mode of inheritance was most likely. Three test-bred litters including those from affected parents, carrier and affected parents, and carrier parents confirmed this mode of inheritance. This study confirms that retinal dysplasia and persistent hyperplastic primary vitreous is a congenital abnormality that is inherited as an autosomal recessive condition in Miniature Schnauzers.

Key Words: inherited retinal dysplasia, Miniature Schnauzer, persistent hyperplastic primary vitreous, retinal detachment

INTRODUCTION

Retinal dysplasia is a common clinical syndrome in dogs. The clinical manifestations include single, multiple, or generalized gray retinal folds in the tapetal or nontapetal fundus.[1] Retinal dysplasia has been morphologically categorized as single folds, geographic, and generalized forms, and the latter two may be associated with retinal detachment.[2] Retinal dysplasia is a congenital lesion, although a developmental form of geographic retinal dysplasia has been recently proposed.[3] The pathogenesis of retinal dysplasia is complex and four developmental types have been proposed:[4] (i) a hyperplastic extension of retina into abnormal sites away from the retinal pigment epithelium (RPE); (ii) within the retina that is detached from the RPE; (iii) within the retina over regions devoid of RPE, i.e. colobomas; and (iv) within the retina underlain by a persistent hyaloid vessel overlying retina or a portion of it with no evidence of detachment from the RPE.[4] Inherited retinal dysplasia in English Springer Spaniels has been the most thoroughly investigated of all the dog breeds. Whitely reported an incomplete retinal differentiation, disruption of the outer limiting membrane, and formation of multiple rosettes within the nuclear and plexiform layers between days 45–50 of gestation in English Springer Spaniels.[5] Multiple etiologies for retinal dysplasia have been identified in animals, including viruses and toxins, although most retinal dysplasia in dogs is inherited.[6–8]

© 2004 American College of Veterinary Ophthalmologists



Affenpinscher	

Ocular disorders known or presumed to be inherited (published)

	Diagnosis	Description and comments specific to the breed	Inheritance	Gene/ marker test	References
A	Cataract	Cataracts have occurred in 2 y.o dogs and have advanced to completion	Unknown	NO	1

The ECVO's advice relating to hereditary eye disease control

Please see ECVO Manual chapter 8: VET Advice

Recommendations regarding age and frequency for eye examinations

Please see ECVO Manual chapter 7: ECVO Age and Frequency recommendations

Other ocular disorders (reported)

	Diagnosis	Source
A	Distichiasis	ACVO genetics committee
B	Persistent pupillary membranes -iris to iris	ACVO genetics committee

DISTICHIASI

German Pinscher (Standard and Miniature)	

Ocular disorders known or presumed to be inherited (published)

	Diagnosis	Description and comments specific to the breed	Inheritance	Gene/ marker test	References
A	Cataract	Posterior cortical; 1,5-3 y.o.	Unknown	NO	1,2,3
B	Persistent hyperplastic tunica vasculosa lentis/ Persistent hyperplastic primary vitreous (PHTVL/ PHPV)		Presumed autosomal recessive	NO	1,3
C	Corneal dystrophy -stromal -endothelial	1. stromal: Clinically evident by 18 months of age 2. endothelial: 7-11 y.o.	Unknown	NO	1,2
D	Glaucoma	3-4 y.o. or 7-8 y.o. PLA	Unknown	NO	1,2
E	Lens luxation	>4 y.o.	Unknown	NO	1
G	Chronic Superficial Keratitis	Usually in dogs >7 y.o.	Unknown	NO	2

H	Vitreous degeneration	Usually in old dogs (>9 y.o.)	Unknown	NO	1
I	Progressive Retinal Atrophy (PRA)	5-7 y.o.	Presumed autosomal recessive	NO	1,2

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Other ocular disorders (reported)

	Diagnosis	Source
A	Persistent pupillary membranes - iris to iris	ACVO genetics committee
B	Optic nerve hypoplasia	ACVO genetics committee
C	Micropapilla	ACVO genetics committee

References

1. Chaudieu G. , Chahory S. Affections oculaires héréditaires ou à prédisposition raciale chez le chien. Ed. du Point Vétérinaire 2013; 162-164.

2. Rubin LF. Inherited eye diseases in purebred dogs. Williams & Wilkins. 1989; 200-201.

3. Leppänen M, Mårtenson J, Miki K: Results of ophthalmologic screening examinations of German Pinschers in Finland – a retrospective study. Vet Ophthalmol 3:165-169, 2001.

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E	Lens luxation	>4 y.o.	Unknown	NO	1
G	Chronic Superficial Keratitis	Usually in dogs >7 y.o.	Unknown	NO	2

DISTROFIA CORNEALE ENDOTELIALE

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E	Lens luxation	>4 y.o.	Unknown	NO	1
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Lussazione della lente

- **La lente si sposta dalla sua sede fisiologica;**

- Classificazione: PRIMARIA (ereditaria), secondaria (Congenita: ***Brittany Spaniel***);
- ***Terriers e Spaniels; Shar Pei*** (presunta ereditarietà, trasmissione recessiva); altre razze;

- Difetto ereditario dell'apparato di sospensione della lente.

Lussazione lente

Lussazione lente

Lussazione lente

- ***Volpino italiano***: presunta ereditarietà; per alterazione delle fibre zonulari;
- ***Tibetan terrier***: autosomico recessivo;
- ***Piccolo levriero italiano;***
- ***PINSCHER***;
- Soggetti affetti: sempre **esclusione** dalla riproduzione.

Possible results using the AHT PLL test			
Genotype	Risk Group	Significance For Breeding	Risk of *PLL* Disease
Homozygous Normal	Normal/Clear	Can be bred to any dog, extremely low risk of producing affecteds (NON trasmettono questa mutazione ai figli!)	Extremely low (esistono altre cause genetiche e acquisite che non possono essere escluse)
Heterozygous	Carrier	Should be bred only to Normal/ Clear to remove risk of producing affecteds (NON vanno eliminati, sono importanti per il pool genetico di razza)	Extremely low (2 %)
Homozygous Mutant	Affected	Should be bred only to Normal/ Clear to remove risk of producing affecteds	Very high

(test su tampone buccale)

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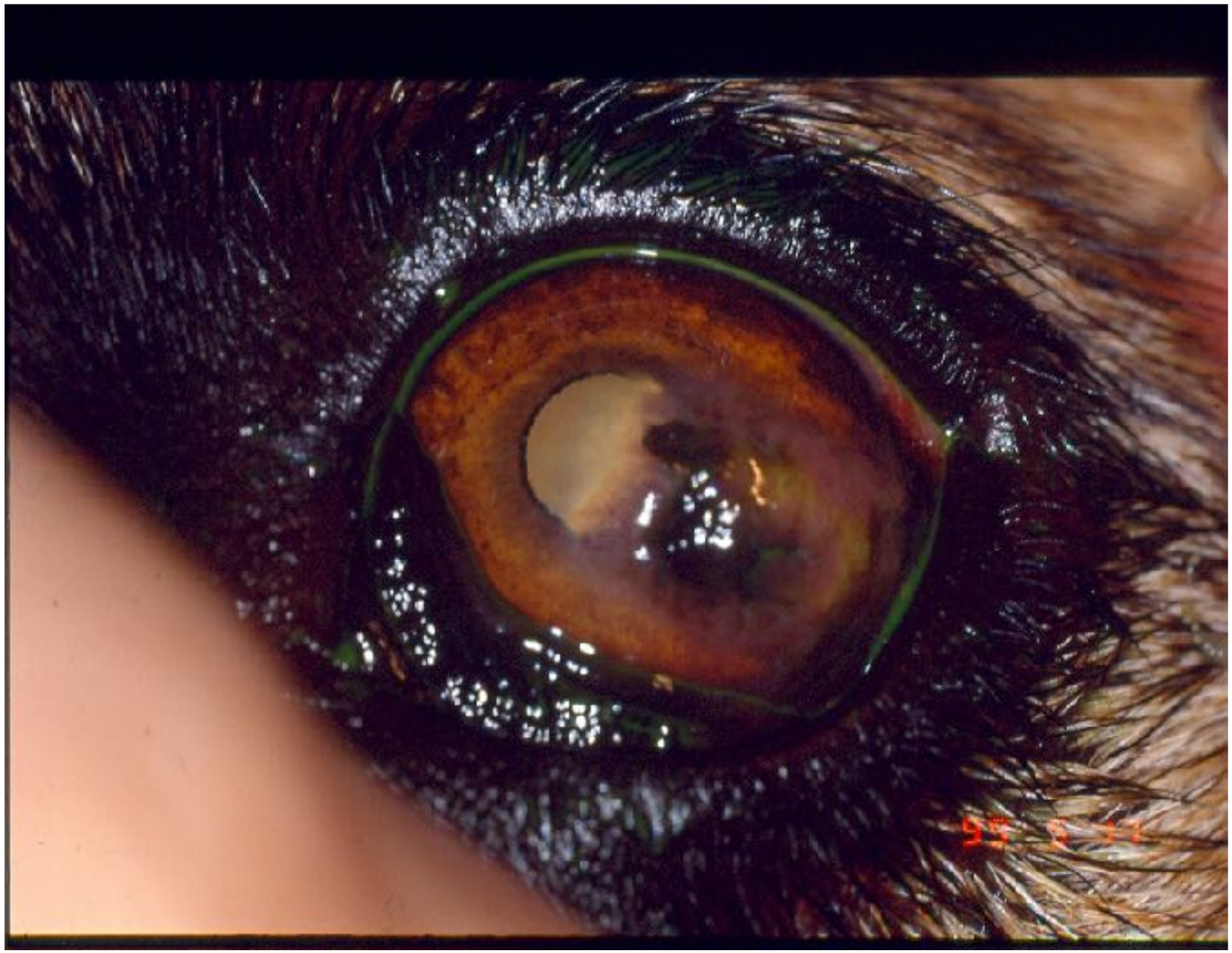
CHERATITE SUPERFICIALE CRONICA



H	Vitreous degeneration	Usually in old dogs (>9 y.o.)	Unknown	NO	1
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IPOPLASIA N.O./ MICROPAPILLA

Malattie oculari per le quali dovrebbe essere "**fortemente sconsigliata**" la riproduzione

Cataratta;

Lussazione o sublussazione della lente;

Glaucoma;

PHPV-PHTVL (2°grado in poi) (es. nel Doberman);

Distacco retinico;

Displasia retinica (f. geografica e con distacco di retina);

Coloboma del N.O. (CEA 2°grado in poi);

Ipoplasia del N.O.;

PRA;

Distrofia corneale endoteliale;

CSK.

Malattie oculari per le quali la riproduzione dovrebbe essere **opzionale**

Distrofia corneale (epiteliale e stromale);

PPM Iride-Iride;

Ipoplasia iridea;

PHPV-PHTVL (1°);

Anomalie palpebrali;

Displasia retinica (f. focali/multifocali);

Ipoplasia della Coroide (CEA 1°).

Malattie autosomiche recessive

Expected Results of Breeding Strategies for Inherited Recessive Diseases			
Parent 1 Genotype	Parent 2 Genotype		
	Normal	Carrier	Affected
Normal	All = Normal	1/2 = Normal 1/2 = Carriers	All = Carriers
Carrier	1/2 = Normal 1/2 = Carriers	1/4 = Normal 1/2 = Carriers 1/4 = Affected	1/2 = Carriers 1/2 = Affected
Affected	All = Carriers	1/2 = Carriers 1/2 = Affected	All = Affected

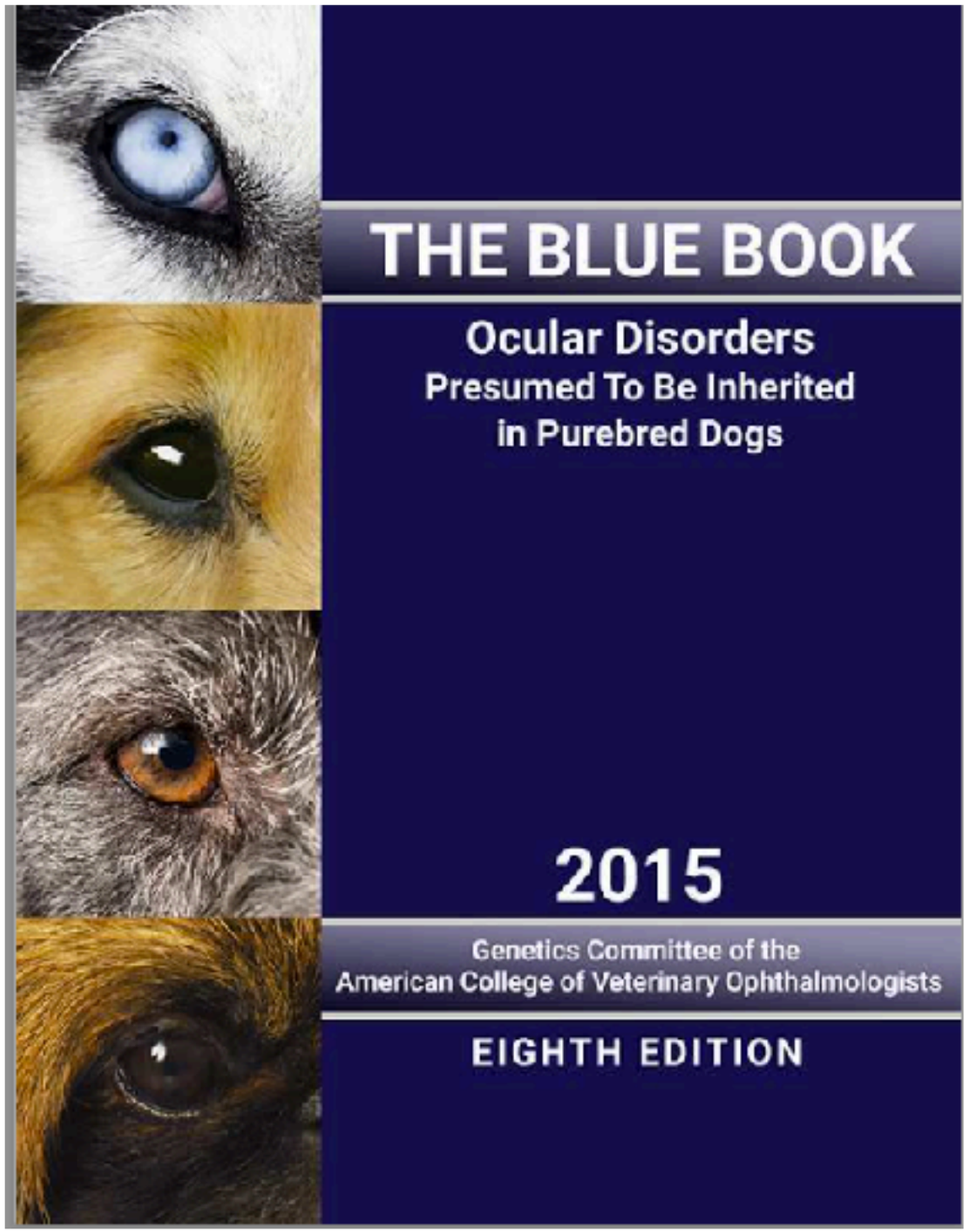
(da www.optigen.com)

Come indagare le Malattie oculari

Lampada a fessura

Oftalmoscopia indiretta

Quali Riferimenti? (ECVO Manual 2017 (www.ecvo.org)/ ACVO Blue Book 2015)



ECVO

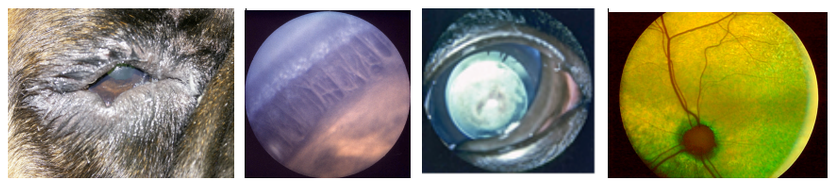
ECVO Manual

for

Presumed Inherited Eye Diseases

in

Dogs and Cats



ECVO HED Committee, 2013

Chi?

- ***Diplomati ECVO***: con certificazione internazionale ECVO (attualmente 5 in Italia);

- ***Accreditati FSA***: con certificazione nazionale FSA (attualmente 30 in Italia);

- ***Abilitati ESE***: con certificazione internazionale ECVO (attualmente nessuno in Italia);

- ***Accreditati ENCI***: con certificazione ENCI (attualmente 8 in Italia).

Quando?

- Nei soggetti ***Riproduttori***: visite effettuate a 1 anno di età e ripetute annualmente fino ai 6-7 anni;

- **Nei soggetti non Riproduttori, tre volte nella vita (1,3,7 anni di età)**;

- Soggetti giudicati ***Esenti***: non presentano segni di malattie ereditarie, possono essere riprodotti, certificato con valenza annuale;

- Soggetti giudicati ***Non definiti/Sospetti***: hanno dei segni di malattia ancora non chiari, si suppone che possano sviluppare una oculopatia ereditaria, vanno riesaminati entro 6-12 mesi ed al momento non riprodotti.

Test DNA **Dove**?

- ***Optigen (US)***
- ***AHT (UK)***
- ***Vetogene (IT)***

- ***Laboklin (D)***
- ***Vetgen***
- ***OFA***
- ***(Antagene)***
- ***(Genefast)***
- ***altri laboratori.***

Selezione

- Soggetti giudicati ***Affetti*** da malattie ereditarie:

1. se malattie “minori”, **Riproduzione opzionale**;
2. se malattie “gravi”, **No riproduzione**.

- ***No Riproduzione***: solo del soggetto malato (es. cataratta);

- ***No Riproduzione***: del soggetto malato, dei suoi genitori e della sua progenie (es. PRA).

ECVO

Certificato di visita oculistica

Certificate of eye examination

European College of Veterinary Ophthalmologists

Registrazione per l'Italia

FIDA - Fondazione Diritti Animali (Animal Health Foundation)
Via Treviso 10
20100 Lissone, Italia
Tel. +39 0312 402671
Fax. +39 0312 403630

ECVO reg.no. Visita reg.no examination

DI-I No. 0006007

Ospedale reg.no Examination reg.no

Animale animal

Nome name

Razza breed

Club di razza owner club

Registrazione n° registration no.

Colore colour

Microchip n° microchip no.

Tatuaggio tattoo

Data di nascita date of birth

Sesso sex: Femmina female / Maschio male

Esame precedente previous examination: No / Sì

Utente ospedale / Soggetto suspected / Non definito undetermined / Affetto affected

Proprietario/agente owner/agent

Da esaminare: data, cert.no. + reg. no. examin.

Nome name

Indirizzo address

Paese Cod. Post. country ZIP code

Città town

Test DNA: Sì / No

Figlio+Data sire + date

Lo/I sottoscritto/i approva la regola del sistema di certificazione razionale e conferma che l'animale portato in visita è quello sopra descritto. La firma indica anche che i risultati sono disponibili per pubblicazione ufficiale o per altri usi approvati dall'ECVO.

The undersigned agree(s) to the rules of the rational scheme and confirm that the animal submitted for examination is the one described above. Signature also means that the results are available for official publication and other ECVO approved use.

Firma proprietario / agente owner / agent signature

Visita oculistica examination

Data date

Giorno day / Mese month / Anno year

Metodo / strumento method / instrument

Midriatico: Oftalmoscopia indiretta e Biomicroscopia binoculare a 10x

Opzionale optional

- Visita pre-dilatazione
- Oftalmoscopia diretta
- Cornioscopia (senza midriatico)
- Altro:

Identificazione identification

Contr. tatuaggio tattoo: Corretto correct / Parzialmente/Non leggibile partly readable / Errato incorrect / Mancante absent

Contr. microchip: Corretto correct / Errato incorrect / Mancante absent

Se è usato un alfa-bebotic, questo stampato ha valore solo se accompagnato da una tecnizoro certificata.

Occhio destro (OD) right eye — Fotografie

ant / post / lat./temp. / med./nas.

Occhio sinistro (OS) left eye — Fotografie

ant / post / med./nas. / lat./temp.

Descrizione description

Malattia oculare so. ocular dis. no. — lieve mild / moderata moderate / grave severe

Nota: affetti/o note: affected by

Nome della malattia in corso di studio non ancora dimostrata ereditaria in questa razza name of disease under investigation not yet proven to be inherited in this breed

Risultati per le malattie ritenute ereditarie: results for the presumed inherited eye diseases

	0 Esente	1/2 Non affetto	1 Affetto	
1. Persistent Pupillary Membranes (PPM)	☐	☐	☐	totale / lieve / severa / grave
2. Persistent Hyperpl. Tunica Vasculosa Lentis/Primary Vitreous (PHTVL/PPV)	☐	☐	☐	grado 1 / grado 2/3
3. Cataratta (congenita)	☐	☐	☐	
4. Displasia della retina (RD)	☐	☐	☐	multifocale / geografica / totale
5. Ipoplasia / Micropapilla	☐	☐	☐	ipoplasia della corioide / vulvulosome / altro
6. Colie Eye Anomaly (CEA)	☐	☐	☐	
7. Altro: other		☐	☐	fibre irlei / lenticolare / coloboma
8. Anomalia L.Pectinis (PLP, only after gonioscopy)	☐			

Risultati validi per 12 mesi results valid for 12 months

	0 Esente	1/2 Sospetto	1 Affetto	
11. Entropion/Trichiasi	☐	☐	☐	
12. Ectropion/Macroblefaro	☐	☐	☐	
13. Distichiasi/Ciglia ectopiche	☐	☐	☐	
14. Distrofia corneale	☐	☐	☐	
15. Cataratta (non congenita)	☐	☐	☐	multiple / post. pol. / l. sut. ant. / punctate / nuclear / other
16. Lussazione lente (primaria)	☐	☐	☐	
17. Degenerazione retinica (PRA)	☐	☐	☐	
18. Altro: other		☐	☐	

Interpretazione interpretation

"Esente" significa che non si evidenziano alterazioni caratteristiche di presunta oculopatia ereditaria, mentre "Affetto" significa che si evidenziano tali alterazioni.
"Unaffected" signifies that there is no clinical evidence of the presumed inherited eye disease(s) specified, whereas "affected" signifies that there is such evidence.

→ L'animale evidenzia alterazioni che potrebbero dipendere dalla presunta oculopatia ereditaria contrassegnata, ma non consentono una diagnosi definitiva.
The animal displays clinical features that could possibly fit the presumed inherited eye disease(s) mentioned, but the changes are inconclusive.

→→ L'animale evidenzia lesioni tipiche delle presunte oculopatia ereditarie contrassegnate. L'esecuzione potrà confermare la diagnosi. Da rieseaminare tra ..... mesi
The animal displays typical lesions of the presumed inherited eye disease(s) mentioned. Further development will confirm the diagnosis. Re-examination in ..... months

Esaminatore examiner

Il sottoscritto ha oggi esaminato l'animale sopra descritto per le indagini sulle oculopatie ereditarie con i risultati sopra riportati.

The undersigned has today examined the above mentioned animal for the hereditary eye diseases with the results as given.

Nome name

Dr Adolfo Guandalini
Spec. in Clin. ne Medica
Socio Op. Vet. S.Giovanni 11-33 00117 RM
Tel 0620993297 Cell 3367659432
C.F. GNDDLF63C24H501J
P.IVA 03397561211
Firma esaminatore autorizzato da ECVO

Colore distribuzione:
Bianco: proprietario/agente
Rosa: esaminatore
Giallo: club di razza nazionale
Bianco: registro nazionale

FSA
Fondazione Salute Animale

FSA- Fondazione Salute Animale (Animal Health Foundation) via Trecchi 20 26100 Cremona, Italia

CERTIFICATO DI VISITA OCULISTICA PER LA DIAGNOSI DELLE MALATTIE OCULARI DI PROVATA O PRESUNTA ORIGINE EREDITARIA NEL CANE

OFFICIAL CERTIFICATE OF EYE EXAMINATION FOR THE DIAGNOSIS OF PROVEN OR PRESUMED INHERITED EYE DISEASES IN DOGS

(Prima parte)

VISITA OCULISTICA DEL: _____**CERTIFICATO N°** _____ **ESAMINATORE Dott.**_____**n° aut FSA**_____

CANE/*DOG*

Nome/*Name*_____**Razza/***breed*_____

Sesso/*sex*_____**Nato il/***date of birth*_____**Colore/***color*_____**Microchip** _____

Tatuaggio/*tattoo* _____ **Test DNA**_____ **no__si/***yes* __ **Data/***date* _____ **Risultato/***result*_____

Esaminato/*checked* __ **Data/***date* _____ **Risultato/***result:* **esente/***unaffected* ___ **affetto/***affected* ___ **non def-sosp/***undet.-susp.* ___

PROPRIETARIO / *OWNER*

Proprietario / owner_____

Indirizzo / *address*_____

Visita, protocollo obbligatorio: Midriatico __ Oftalmoscopia indiretta __ Biomicroscopia binoculare > 10x __ Esame pre-dilatazione x razza __

Altre indagini: Esame pre-dilatazione __ Oftalmoscopia diretta __ Gonioscopia __ Tonometria __ Altro_____

Risultati per le malattie ritenute congenito/ereditarie

	Esente *	Non definito**	Affetto ***	
1. Mem. Pupil. Persistens (PPM)	○	○	○	iride □ lente □ cornea□ lamina□
2. Pers. Hyp.T. Vas.L./Pr. Vit. (PHTVL/PHPV)	○	○	○	grado 1□ gradi da 2 a 6□
3. Cataratta (congenita)	○	○	○	cort□ post pol□ ant pol□ sut□punct□nucl□
4. Retina: displasia (RD)	○	○	○	(multi)focale□ geografica□ totale□
5. Ipoplasia n.o./ Micropapilla	○	○	○	
6.Collie Eye Anomaly (CEA)	○	○	○	ipoplasia coroide□ coloboma□altro□
7. Altro _____	○	○	○	
8. Anomalie L. Pectinatum	○	○	○	fibrae latae□ laminae□ occlusio□

Risultati per le malattie ritenute ereditarie

	Esente*	Sospetto****	Affetto***	
9. Entropion/trichiasi	○	○	○	
10. Ectropion/macroblepharon	○	○	○	
11. Distichiasi/ciglia ectopiche	○	○	○	
12. Distrofia corneale	○	○	○	epiteliale□ stromale□ endoteliale□
13. Cataratta (non congenita)	○	○	○	cort□ post pol□ ant pol□ sut□ punct□nucl□
14. Lussazione primaria lente	○	○	○	
15. Degenerazione retinica (PRA)	○	○	○	
16 Altro _____	○	○	○	

N.B. l'esenzione da oculopatie eretidarie non è permanente ma il cane deve essere rivalutato ogni 12 mesi

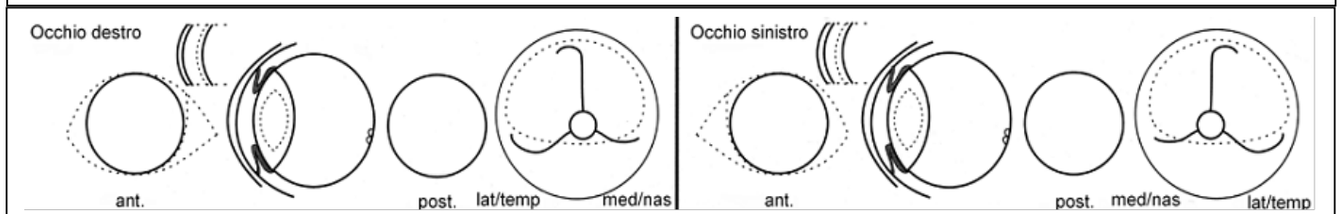
Non affetto, non si evidenziano alterazioni caratteristiche di oculopatie ereditarie * Affetto, si evidenziano tali alterazioni*
***Si osservano alterazioni che potrebbero dipendere da una oculopatia ereditaria ma non sono del tutto patognomoniche. Riesaminare l'animale a distanza di_____mesi*
*****Vi sono alterazioni di lieve entità, si sospetta l'inizio di un'oculopatia ereditaria. Riesaminare l'animale a distanza di _____mesi*

MALATTIA N°_____ **Gonioscopia: anomalia L. Pectinatum lieve __moderata __ grave__**

DESCRIZIONE DEL QUADRO CLINICO_____

_____

Nota: affetto da _____**basi ereditarie non definite in questa razza**



(Seconda parte)

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A cosa servono i certificati ed i test genetici?

1) allevamento “sano”; beneficio per CANI, Allevatori e Proprietari
2) “movimento” internazionale di cani; adeguamento a standard più elevati di lavoro
3) studi epidemiologici

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Epidemiology of ocular disorders presumed to be inherited in three large Italian dog breeds in Italy

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Abstract

Objective To describe the epidemiology and the types of eye disorders that are presumed to be inherited (PIED) in three large Italian dog breeds.

Animals Three large Italian dog breeds: Neapolitan Mastiff (FCI code: 197), Maremma Sheepdog (FCI code: 201), and Italian Corso dog (FCI code: 343).

Procedures All dogs that underwent a complete ophthalmic examination between 1992 and 2012 were included in this prospective observational study. The prevalence of eye disorders with 95% confidence intervals was reported for presumed healthy dogs and for dogs referred to a veterinary center for an ophthalmic consultation. Univariate and multivariate logistic regression techniques were used to generate odds ratios.

Results Of 605 dogs examined during the study period, 351 dogs were affected by at least one PIED (58%; 95% CI: 54–62%). The prevalence of PIED was significantly lower in dogs presented for ophthalmic examination (53.8%) as compared to presumed healthy dogs (62.2%)(OR: 1.4; 95% CI: 1.02–1.9; $P = 0.037$). Also after multivariate adjustment for the period of observation, the odds of Neapolitan Mastiff (92.1%; OR: 21.4; 95% CI: 11.1–41.4) and of Cane Corso (57.7%; OR: 2.5; 95% CI: 1.7–3.6) suffering a PIED were greater than the Maremma Sheepdog (35.4%). The most common PIED in each breed were entropion (24.3% of all the PIED) in the Neapolitan Mastiff, ectropion (36.6%) in the Corso dog, and cataract (27.9%) in the Maremma Sheepdog.

Conclusions Clinicians should be aware that three large Italian dog breeds frequently suffer PIED. Breed standards should be reconsidered, and breeding programs should be directed at limiting such disorders.

Key Words: breed, ectropion, entropion, epidemiology, inherited, predisposition

INTRODUCTION

Inherited eye diseases significantly impair vision in humans and animals.[1–4] As of 2015, 278 genes and loci have been associated with retinal disorders in humans.[5] The identification and characterization of inherited eye disorders are critical for the diagnosis, prevention, and treatment of such disorders.[6,7]

Several inherited and presumed to be inherited eye disorders have been described in pure-bred dogs.[2,8–11] Until the genetic basis of an ocular disorder is properly defined, ocular disorders that are suspected to have a genetic basis are defined as 'presumed to be inherited eye disorders' (PIED). An ocular disorder is defined as 'presumed to be inherited' when it is (i) observed more frequently in one breed compared with other breeds; (ii) observed more frequently within related dogs of a certain breed; (iii) with characteristic appearance and location; (iv) with characteristic age of onset and course of progression; and (v) similar to an entity which has been proven to be inherited in other breeds.[10]

Italy is the homeland of sixteen dog breeds.[12] These include three large-size breeds: the Neapolitan Mastiff [Fédération Cynologique Internationale (FCI) code: 197], the Maremma Sheepdog [FCI code: 201], and the Italian Corso dog [FCI code: 343].[12] Scientific knowledge about

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Conclusioni

- Importanza della **prevenzione** e della diagnosi precoce;

- Importanza delle **visite** oculistiche e dei **tests** genetici (l'uno non esclude l'altro); Possono esserci **mutazioni diverse** per la stessa malattia!;

- Per moltissime patologie **non esiste** ancora il test genetico!;

- Importanza nella **collaborazione** tra veterinari ed allevatori; **Unione** di intenti, fiducia e serietà reciproca!

Domande
???