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BRIEF COMMUNICATIONS

A new inherited ocular anomaly in pigmented White Leghorn chickens

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Inherited ocular anomalies in chickens include several types of microphthalmia, retinal dysplasia, retinal degeneration, cataract, buphthalmos and pop-eye, or keratoconus in White Leghorns.[2–5,7,8,12,15–18] In this report, a new ocular anomaly that appeared in pigmented White Leghorns homozygous for a mutation at the dominant white (*I*) locus is described with emphasis on clinical, gross, and histologic findings to aid in the diagnosis of the ocular lesions.

An incompletely dominant mutation, called Smoky Joe (*SJ*), allows the production of feather pigment, which the *I* allele inhibits. The *SJ* mutation originally appeared in ADOL Line 0, a noninbred White Leghorn line maintained at the USDA Avian Disease and Oncology Laboratory (ADOL) in East Lansing, Michigan. Females homozygous for *SJ* have dark grey feathers with barring and homozygous males have much lighter colored plumage. This dimorphism in color is the result of the dilution that occurs with two copies of sex-linked barring in the males. The plumage of heterozygous birds (*I, SJ*) is intermediate in color compared to either parental type.

Phthisis bulbi was noticed in adult females of the second generation of the homozygous *SJ* population. None of the parents of these females had apparent ocular lesions. Approximately 30% of the 85 surviving females had phthisis bulbi, while none of the 100 surviving males were affected. There was no clinical evidence of bacterial or viral infection, and the chickens were housed in pens isolated from chickens with experimental viral infections. Because the ocular anomaly appeared to be recessive and sex-linked, 4 test matings were designed to further study the inheritance.

In matings 1 and 2, sighted *SJ* males, which were suspected of being carriers of the syndrome, were mated to either affected or sighted *SJ* females. These same male birds were then mated to either sighted females of Line 0 (*II*) (mating 3) or sighted females of ADOL Line $15I_5$ (*II*) and Line 0 (*II*) (mating 4). All chicks had ophthalmic examinations at hatch and 2, 4, and 8 weeks of age.

In mating 1,197 chicks were examined. Of the females, 62% were affected, and 10% of the males were affected. Twenty-two progeny from mating 2 were examined, and 40% of the females and 8% of the males were affected. In both of these matings, approximately equal numbers of each sex were produced, and fertility and embryo viability were within a normal range. These data confirmed a much higher incidence of the ocular anomaly in females and therefore indicated a sex-influenced expression.

In matings 3 and 4, when blind or carrier birds were mated to nonpigmented White Leghorns (*II*), only one affected chick was observed in 242 progeny. These data support the hypothesis that the anomaly is autosomal recessive. The affected chick in mating 4 was most likely the result of an error in the pedigree.

Ophthalmic examinations of the newly hatched chicks were performed by using a slit lamp biomicroscope and an indirect ophthalmoscope. Affected chicks had ocular lesions varying in number and severity, but both eyes were always affected. The earliest and most subtle lesion was the suspension of darkly pigmented cells throughout the vitreous body. Other lesions included sluggish or absent pupillary light reflexes, dilated pupils, mild to moderate aqueous flare containing pigmented cells, thickened and grey irides, iridodonesis, temporal and posterior lens subluxation, adherence of pigmented cells to the anterior lens capsule, malformed lenses, and immature cataract localized to the anterior and posterior lenticular cortex. Cataracts were present either at hatch, or developed by 4 weeks posthatch. Buphthalmos was often present at hatch, but intraocular pressures were similar to those of unaffected chicks (Fig. 1). Phthisis bulbi was often present by 8 weeks of age but appeared as early as 4 weeks of age. Other secondary ocular lesions included iridocyclitis, posterior synechia, and iris bombé.

Histologic examination of the globes of 17 affected chicks confirmed the clinical findings. The globes of 9 newly hatched affected chicks had melanin-laden cells in the trabecular meshwork and dispersed throughout a liquefied vitreous body (Fig. 2). The pecten was smaller than those from unaffected chicks and lacked a pleated structure (Fig. 3). The lenticular cortex was liquefied and the ciliary processes did not contact the lens capsule. The peripheral retina had disorganized layers and the retina was detached. In older chicks, phthisis bulbi was characterized by collapse of the anterior chamber, cataract, and metaplastic bone and cartilage filling the vitreous chamber (Fig. 4).

Although cataract and buphthalmos have been previously reported in chickens, the presence of the these lesions concurrent with other ocular lesions is unique.[2,8,12] In addition, dysplasia of the pecten and subluxation of the lens don't appear to be previously reported in chickens. Because the

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Figure 1. Affected pigmented White Leghorn chick with bilateral buphthalmos (**left**); unaffected pigmented White Leghorn chick with normal right eye (**right**).

retina, pecten, and ciliary body are structures containing pigmented cells of neuroectodermal origin, faulty neuroectoderm development may be the cause of the malformed pecten, shortened ciliary processes, and dispersion of pigmented cells.[11] Pigment first appears in the pecten of the chick embryo after 8 days of incubation, and pleats begin to form by the ninth or 10th incubation day.[13] Also, zonular fibers are in their definitive position by incubation day 8 to 9, which allows for fusion of the lens capsule with the limiting membrane of the ciliary body.[13] Because both of these normal conditions are absent in affected chicks, it would appear that the genetic influence is exerted at or before 8 days of gestation.

Not all of the lesions are the result of a primary genetic abnormality. Subluxation of the lens may be secondary to a primary lesion of the ciliary body or secondary to buphthalmos. Lymphocytic iritis is most likely attributable to phacolytic uveitis.[5,6] Cataract may be secondary or may be

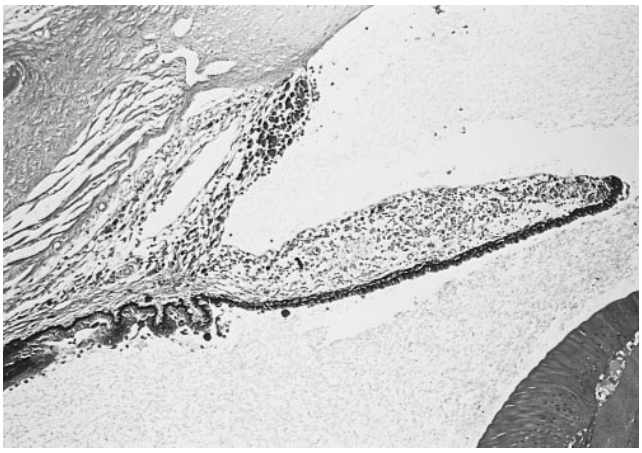

Figure 2. Histologic section of a portion of the eyeball from an affected newly hatched pigmented White Leghorn chick. Notice the melanin-laden cells present in the anterior chamber and aggregated in the trabecular meshwork and the lack of attachment of the small ciliary processes to the lens (**right**). HE.

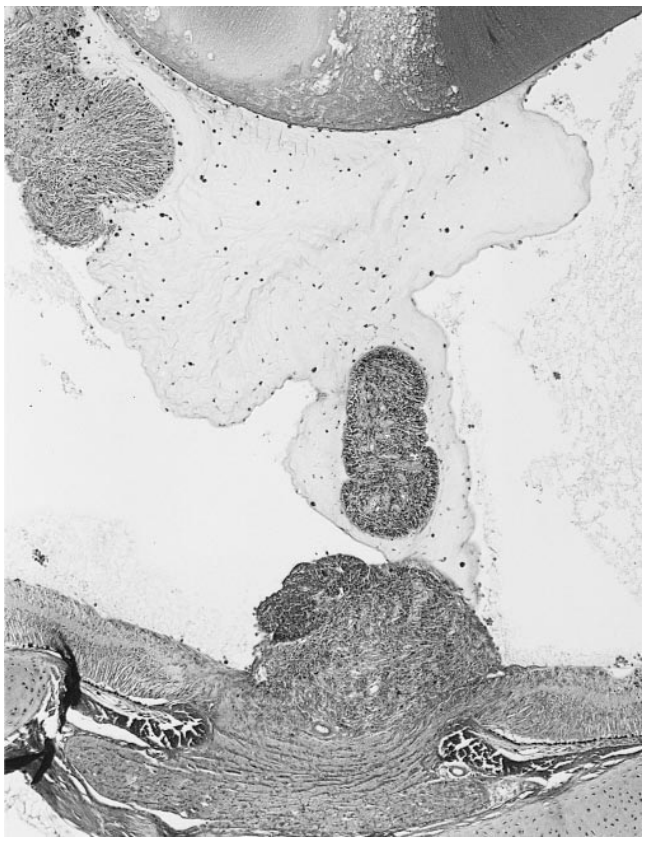

Figure 3. Histologic section of the posterior segment of an eyeball from an affected newly hatched pigmented White Leghorn chick. The cataract is characterized by liquefied cortex, liquefied vitreous body with dispersed melanin-laden cells, distorted pecten (**bottom**), and presence of detached retina immediately posterior to the lens (**left**). HE.

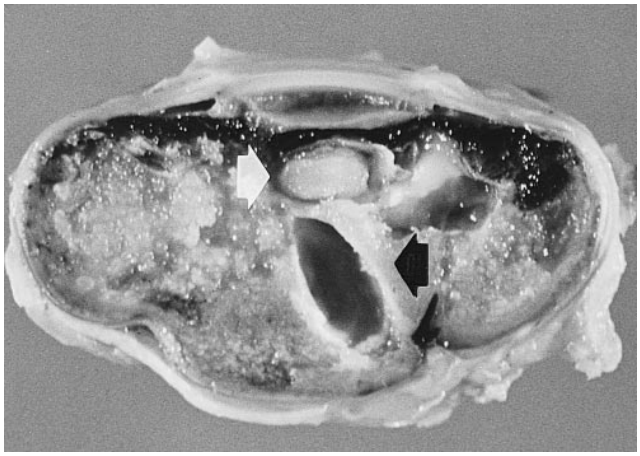

Figure 4. Cut surface of sectioned phthisical globe from a pigmented White Leghorn chick with a flat cornea (positioned up) due to collapse of the anterior chamber, cataract (white arrow) with irregular lens capsule and liquefied cortex, and distorted black pecten (black arrow) surrounded by metaplastic bone filling the vitreous chamber.

a primary lesion due to a defect in development of surface ectoderm.[2,6] Phthisis bulbi with metaplastic bone formation has been reported in chickens secondary to an inherited retinopathy but may occur as a sequela to marked intraocular degeneration.[1,15]

Neural crest cells give rise to ocular connective tissue, including uveal melanocytes,[9,10] and abnormal migration or defective induction by these neural crest cells may cause goniodysgenesis, which causes glaucoma leading to buphthalmos. Therefore, goniodysgenesis may be the result of faulty melanogenesis. Faulty retinal development has also been associated with abnormal melanogenesis. For example, chicks with delayed amelanosis are blind as the result of retinal degeneration.[15] Retinal degeneration is attributable to a reduction and loss of melanin granules in the retinal pigment epithelium. The role that the barring gene plays in the migration and development of embryonic neural crest cells is unknown.[14]

In conclusion, the inherited ocular anomalies appear to be the result of a sex-influenced, autosomal recessive mutation in pigmented White Leghorns.[2,8,12] Further studies of this syndrome will include morphologic examination of globes from affected chick embryos and experimental breeding to determine the possible role of melanin in the cause of this anomaly.

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References

1. Allen CB, Ulshaver RJ, Ellis EA, Woodard JC: 1987, Scanning electron microscope analysis of intraocular ossification in advanced retinal disease. Scan Micro 1:233–239.
2. Bitgood JJ, Swanson JF, Dubielzig RR: 1986, A new mutation resulting in blindness and inherited cataracts in the chicken. Pout Sci 65(Suppl. 1):14.
3. Bitgood JJ, Whitley RD: 1986, Pop-eye: an inherited Z-linked keratoglobus in the chicken. J Hered 77:123–125.
4. Cheng KM, Shoffner RN, Gelatt KN, et al.: 1980, An autosomal recessive blind mutant in the chicken. Poult Sci 59:2179–2182.
5. Chmielewski NT, Render JA, Schwartz LD, et al.: 1993, Cataracts and crooked toes in Brahma chickens. Avian Dis 37: 1151–1157.
6. Cook CS: 1995, Embryogenesis of congenital eye malformations. Vet Comp Opthalmol 5:109–123.
7. DeVolt HM: 1944, Lamellar cataracts in chickens. Poult Sci 23: 346–348.
8. Hutt FB: 1935, Herditary blindness in the fowl. Poult Sci 14: 297.
9. Johnston MC: 1966, A radioautographic study of the migration and fate of cranial neural crest cells in the chick. Anat Rec 156: 143–156.
10. Johnson MC, Noden DM, Hazelton RD, Coulombre JL, Coulombre AJ: 1979, Origins of avian ocular and periocular tissues. Exp Eye Res 29:27–43.
11. O'Rahilly R, Meyer DB: 1961, The development and histochemistry of the pecten oculi. *In:* The structure of the eye, ed. Smelsen GK, pp 207–219. Academic Press, New York, NY.
12. Pollock BJ, Wilson MA, Randall CJ, Clayton RM: 1982, Preliminary observations of a new blind chick mutant (*beg*) *In:* Problems of normal and genetically abnormal retinas, ed. Clayton RM, Reading HW, Haywood J, Wright A, pp. 241–247. Academic Press, London.
13. Romanoff AL: 1960, The avian embryo. Structural and functional development. The MacMillian Co., New York, NY.
14. Schreck RE, Bowers RR: 1989, Effect of the barring gene on eye pigmentation in the fowl. Pigment Cell Res 2:191–201.
15. Smyth Jr. JR, Boissy RE, Fite KV: 1981, The DAM chicken: a model for spontaneous postnatal cutaneous and ocular amelanosis. J Hered 72:150–156.
16. Somes Jr. RG: 1992, Microphthalmia-4: sex-influenced inherited eye condition of the chicken. J Hered 83(2):152–155.
17. Somes Jr. RG, Cheng KM, Bernon DE, Crawford RD: 1990, Mutations and major variants of other body systems in chickens: mutations involving the eye. *In:* Poultry breeding and genetics, ed. Crawford RD, pp. 281–291. Elsevier, Amsterdam.
18. Ulshaver RJ, Allen C, Dawson WW, Wolf ED: 1984, Hereditary retinal degeneration in the Rhode Island Red chicken. I. Histology and E.R.G. Exp Eye Res 39:125–135.
19. Yanoff M, Fine BS: 1975, Ocular pathology: a text and atlas. Harper and Row, Hagerstown, MD.