OCULAR ASSOCIATIONS OF DAPPLING (OR MERLING) IN THE COAT COLOUR OF DOGS

I. CLINICAL AND GENETICAL DATA

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1. CLINICAL OBSERVATIONS ON THE DACHSHUND AND THE COLLIE

The Dachshund

Some breeders of dachshunds specialize in the production of dapples. The self-coloured (or uniformly coloured) dachshund may be black, red, chocolate or cream in colour, but the commonest coat colour is black and tan (which in the context of this study is grouped with self-coloured). The black, and black and tan dapples show streaks of silver in addition to the basic colour, and are known as silver dapples; dappling in the form of light streaks against the normal colour background is also seen in the red and in the chocolate dachshund. The amount of streaking varies considerably—some animals being heavily marked, others only slightly—and tends to fade with age. Breeders do not mate dapple to dapple, for they are aware that this gives troublesome results. In order to maintain their dapple strains they mate dapple to self-coloured animals. Most breeders say that in the litters so obtained there are more self-coloured than dappled animals, and some hold that the dapple is a more fragile animal requiring greater care in rearing—a belief disputed by others.

Breeders also know that some of the dapples have 'wall eye'. Some distinguish between blue eye and 'wall eye'; in blue eye the colour is lacking in a segment or the whole of one iris, and in extreme cases in both irides. In wall eye there is a more extensive anomaly: the limbus is not well defined, and there seems to be some encroachment of the sclera on to the cornea. Wall eye is held to be functionally normal.

Occasionally a dapple to dapple mating occurs. In such litters self-coloured animals and dapples are seen, as well as 'extreme dapples'. The 'extreme dapples' are unusual animals, having a large element of white in their coat. They are known to have abnormal eyes and are frequently blind. They are also reputed to be deaf. One breeder told us that of three 'extreme dapples' in one litter, two had to be destroyed because they could not move about easily, and were held by the veterinary surgeon to have congenital heart disease. Another breeder also had to destroy two out of three 'extreme dapples' obtained in one litter.

These facts are widely known, and through the kind co-operation of several breeders we have been able to examine a number of self-coloured dachshunds and dapples, as well as three 'extreme dapples'.

Observations on the eye

The self-coloured dachshund

In the black, red, and black and tan dachshund, the iris is dark brown; in the chocolate it is hazel in colour. The self-coloured animal shows a brilliantly coloured tapetum above the disk. The colour and its intensity vary with age and coat colour. In the black and tan, the tapetum is a rather brilliant light greenish blue; in the chocolate there is rather more blue merging on to mauve as the tapetum thins out towards the normal reddish background. In the tapetal area no detailed structure can be made out, the striking colour dominating the picture. The disk is not unlike the human disk, except that more branches from the central artery of the retina are seen.

These appearances are apparently constant; we have observed no exception to the uniform coloration of the iris, or to the marked brilliant hue of the tapetum.

The dapple

A total of thirty-five dapples were examined, thirty-one being black and tan silver dapples. Anomalies of the iris, episcleral veins, and the tapetum were observed.

Anomalies of the iris. These are not common. They were observed in only fifteen eyes out of seventy (Table 1), and they ranged from minimal discoloured flecks on either the free margin, or at the base of the iris, to more extensive discoloration involving half or more, or the whole of the iris, and, as observed in two cases, the whole of both irides.

Anomalies of the episcleral veins. In three dapples, two or three markedly engorged episcleral veins were seen when the upper lid was retracted; they were bilateral in one animal.

Anomalies of the tapetum. These were the most striking and constant. In no animal was there a normal tapetum, but the anomalies observed showed some variation.

(*a*) In ten of the thirty-five dapples there was no trace of a tapetum in either eye. The area normally occupied by this structure showed the same background as the rest of the fundus, though in some there was a faintly granular appearance reminiscent of a poorly marked 'pepper and salt' fundus in man.

(*b*) In twenty-one dapples the appearances were interpreted as due to the presence of a rudimentary tapetum. The tapetal area could be distinguished from the rest of the fundus background, but this area was devoid of all colour in all eyes except five. In these five a definite, though patchy, coloration was observed; but there was no suggestion of the intensely brilliant uniform normal tapetum.

(*c*) The remaining four animals showed no tapetum in one eye and a rudimentary tapetum in the other.

Other fundus anomalies. No other marked fundus anomalies were observed except that in one eye there was a sharply defined patch below the disk with exposure of the choroidal vessels, whilst in another eye the tapetal area showed a fundus tigré appearance. A generalized albinotic background was not uncommon.

Correlation of these anomalies. (*a*) *Iris anomalies in relation to tapetal anomalies.* It will be seen from Table 1 that of the ten dapples lacking a tapetum in both eyes, six had normal irides, and three more showed a normal iris on one side and a minimally discoloured iris on the other side. Only one showed total discoloration of both irides.

Table 1 also shows that the twenty-one animals with rudimentary tapeta showed an incidence of the different varieties of discoloration of the iris that does not differ substantially from that observed in the dapples without any tapetum.

(b) Iris and tapetal anomalies in relation to episcleral veins. The relation of this anomaly to the different anomalies of tapetal and iris anomalies are shown in the following summary table:

Animal no.		Engorged episcleral veins	Tapetum		Iris		
			Rudimentary	None	Normal	Totally discoloured	Sectorially discoloured
61	R. eye	+	+	.	+	.	.
	L. eye	+	+	.	.	.	+
56	R. eye	−	.	+	+	.	.
	L. eye	+	+	.	.	+	.
27	R. eye	−	+	.	+	.	.
	L. eye	+	+	.	+	.	.

It will be seen that episcleral veins do not necessarily occur in eyes showing the most severe tapetal or iris anomaly.

(c) Iris and tapetal anomalies in relation to the basic coat colour of the dapple. Of the thirty-five dapples, thirty-one were smooth-haired black and tan silver dapples, three were smooth-haired chocolate dapples, and the remaining one was a 'pepper-and-salt' long-haired dapple.

The incidence of anomalies in the six eyes of the three smooth-haired chocolate dapples was:

Tapetum: Rudimentary in both eyes of two animals. None in both eyes of one animal.

Iris: Normal in five eyes, and sectorially discoloured in one eye of one of the animals with a rudimentary tapetum.

The 'pepper-and-salt' long-haired dapple showed no tapetum in one eye and a rudimentary tapetum in the other, with a normal iris on the side of the rudimentary tapetum and a sectorial discoloration of the iris on the side without a tapetum. These findings do not differ materially from those observed in the silver dapples.

The 'extreme dapple'

The three 'extreme dapples' seen were aged 2, 10 and 18 months. They all showed extensive ocular anomalies. The eyes appeared small and deeply set. The sclerae were thin and bluish in colour. The limbus was ill-defined and there was encroachment of the sclera on to the cornea. Enlarged episcleral veins were present in all eyes. The irides were markedly hypoplastic and almost completely devoid of colour. In one, the pupils were eccentric and the adjacent iris suggested an incomplete atypical coloboma; in another there was a persistent pupillary membrane in one eye. In all six eyes the iris was hypoplastic, fairly translucent, and there were many holes and clefts readily seen with the ophthalmoscope. The lenses were slightly cloudy in two animals, and in the animal with ectopic pupils one lens gave a suggestion of a coloboma at the lower margin. In the third extreme dapple the lenses were subluxated. Vitreous opacities were present in five and possibly all the six eyes. Ophthalmoscopically there was no tapetum in any of the six eyes, and the disks were all markedly atrophic. In addition the fundus showed towards its periphery large scattered areas of white shining background without any overlying choroidal or retinal vessels. Two of these animals moved as if they could see, but the objective findings leave no doubt that vision must be grossly defective.

THE COLLIE

Dappling in the collie is known as merling, and very much the same findings as seen in the dappled dachshund are reputed to occur in the merle collie.

The collie normally is tricoloured (black, brown and white), or sable (brown and white). The blue merle collie is a tricolour animal in which black is replaced in patches by merling. It is valued by breeders in contrast to the sable merle which is not fashionable (and which we have not been able to see). In all nine blue merles (presenting seventeen eyes) were examined. Like the dappled dachshund they are not mated to each other,

but to tricolour; when merle to merle matings occur 'white collies' are reputed to appear in the litter.

Observations on the eye

The tricolour

The iris colour is brown in the tricolour collie and light brown in the sable. The tapetum of the adult is a brilliant yellow-green and a blue-green or blue in the puppy.

The blue merle

As can be seen from Table 1, anomalies of the iris and of the tapetum were observed. No eye showed engorged episcleral veins.

Table 1. *Findings on the iris and tapetum in merled dogs*

	Total no. of dogs seen	Both irides normal in colour	Both irides discoloured			One iris normal and the other discoloured	
			Both totally	One totally and other sectorially	Both sectorially	Totally	Sectorially
Dachshund							
Normal tapeta	—	—	—	—	—	—	—
Rudimentary tapetum in both eyes	21	14	1	—	—	2	4
Rudimentary tapetum in one eye and no tapetum in the other	4	2	—	—	—	1*	1†
No tapetum in both eyes	10	6	1	—	—	—	3
Total	35	22	2	—	—	3	8
Collie							
Normal tapeta	—	—	—	—	—	—	—
Rudimentary tapetum in both eyes	4	2	—	—	—	—	2
Rudimentary tapetum in one eye and no tapetum in the other	1	—	—	1	—	—	—
No tapetum in both eyes	4‡	2½‡	—	—	1	—	—
Total	9	4½	—	1	1	—	2
Miniature collie							
Normal tapeta	—	—	—	—	—	—	—
Rudimentary tapetum in both eyes	1	—	—	—	—	—	1
Rudimentary tapetum in one eye and no tapetum in the other	1	—	—	—	—	1§	—
No tapetum in both eyes	1	—	1	—	—	—	—
Total	3	—	1	—	—	1	1

* On side with the rudimentary tapetum.
† On the side devoid of tapetum.
‡ The right eye of one collie could not be fully examined as the lids were partially adherent on that side.
§ On side lacking tapetum.

Anomalies of the iris. Discoloration of the iris was seen in six out of seventeen eyes. Complete discoloration was only seen in one eye. The remainder had a sector of discoloration, except that one iris was flecked with white radial streaks.

Anomalies of the tapetum. In the seventeen fundi seen there was no tapetum in eight and only a rudimentary tapetum in the remaining nine.

Other fundus anomalies. In one animal there was a segment below the disk in which the choroidal blood vessels were exposed to view—possibly due to depigmentation in the overlying retina.

Other anomalies. In one animal the nictitating membrane was very prominent—a condition reputed to be common in merles, but not tricolours. In another the lids had failed to open fully on one side and the cornea was scarred. This was the eye that could not be further examined.

Correlation of the iris and tapetal anomalies. As in the dappled Dachshund tapetal anomalies were constant, and iris anomalies less frequent—with no correlation between the severity of the tapetal defects and those of the iris (Table 1).

The white collie

None was available. The breeder reported having had one previously which had white irides and was not blind. The nictitating membranes were large and the animal died from the anaesthetic while these were being removed.

THE MINIATURE COLLIE (SHETLAND SHEEP-DOG)

Observations on the eye

The tricolour and sable

Two tricolours and one sable were seen. Their eyes were normal and differed in no way from those of collies except that they were smaller.

Miniature merle collies

Three such animals were seen. Breeders apply the same breeding restrictions on miniature merle collies as on the larger variety.

Anomalies of iris. This was present unilaterally in two, and in both eyes in one animal.

Anomalies of the episcleral veins. None was seen.

Anomalies of the tapetum. No animal had a normal tapetum. The animal with bilateral discoloration of the iris showed no tapetum in either eye.

Miniature white collies

These are reputed to occur from a merle to merle mating. None was seen.

2. BREEDING RESULTS

Table 2 shows breeding results compiled from two sources: breeders' records of litters that were seen by us and from breeders' records of other litters where full details had been kept. Data were available on each of the three breeds of dog.

TYPES OF MATING

Three types of breeding were observed.

(1) *Matings of dapple (or merle) to full coat* (Table 2(*a*))

Dachshund. As can be seen from Table 2(*a*) forty-five dapple to full coat matings resulted in ninety-seven dapples and 102 undappled offspring.

Collies. Five merle to tricolour matings gave rise to nineteen merles and twelve unmerled animals. In addition, details were obtained of one mating between a sable merle and a tricolour, the issue from which were four tricolour and four sables. There was also one mating between a blue merle and a sable resulting in two blue merles and four tricolours.

Miniature collie. Seven matings were recorded between merles and tricolours. The resulting offspring consisted of ten merle and fifteen tricolour. There was, in addition, one mating between sable merle and tricolour which resulted in two sable merles and one tricolour.

(2) *Dapple to dapple (or merle to merle) matings* (Table 2(*b*))

This type of mating does not frequently happen by design; in consequence few records were available.

Table 2. *Breeding data on dappled dogs*

(*a*) Self-coloured to dapple

		Offspring			
		Dapple or merle		Non-dappled	
	Parents	M.	F.	M.	F.
Dachshund	Dappled × self-coloured (45 matings)	45	52	47	55
Collie	Blue merle × tricolour (5 matings)	10	9	5	7
	Sable merle × tricolour (1 mating)	—	—	4*	4†
	Blue merle × sable (1 mating)	1	1	2	2
Miniature collie	Blue merle × tricolour (7 matings)	7	3	11	4
	Sable merle × tricolour (1 mating)	1	1	—	1

(*b*) Dapple to dapple

		Offspring					
		Non-dappled		Dapple or merle		Extreme dapple or merle	
	Parents	M.	F.	M.	F.	M.	F.
Dachshund	Dapple to dapple (6 matings)	4	1	4	7	7‡	3
Collie	Blue merle × blue merle (2 matings)	1	1	2	3	2	1
Miniature collie	Blue merle × blue merle (2 matings)	4	3	1	2	—	—
	Blue merle × sable merle (1 mating)	—	—	2§	2§	—	—

(*c*) Matings involving an extreme dapple

		Offspring					
		Non-dappled		Dapple or merle		Extreme dapple	
	Parents	M.	F.	M.	F.	M.	F.
Dachshund	Extreme dapple × dapple	—	—	1	—	—	1
	Extreme dapple × self-coloured	—	2	—	2	—	—

* Three tricolours and one sable.
† One tricolour and three sables.
‡ One of these came to histological examination when three months old.
§ One blue merle and one sable merle.

Dachshund. Six matings were recorded. They resulted in ten extreme dapples, eleven dapples and five undappled animals.

Collie. The two matings recorded resulted in three extreme merles, five merles and two unmerled animals.

Miniature collie. There were three merle to merle matings resulting in seven merles and seven unmerled offspring; there were no extreme merles.

(3) *Matings involving an extreme dapple* (Table 2(*c*))

Extreme dapples are generally not reared so that matings involving these animals are infrequent:

(i) a mating between a dappled dachshund and an extreme dapple resulted in a dapple and an extreme dapple;

(ii) a mating between an extreme dapple and an undappled dachshund reputedly resulted in two dappled and two undappled animals. (Unfortunately examination of the offspring in this litter was not possible. Minimal dappling of the coat may have been present in the 'non-dappled' animals or perhaps some tapetal anomaly.)

In summary form the results recorded are:

Dachshunds and collies

Mating of self-coloured to dapple or merle

No. of matings	Offspring	
	Dapple	Self-coloured
60	130	142

Mating of dapple to dapple (or merle to merle)

No. of matings	Offspring		
	Full coat	Dapple or merle	Extreme dapple or extreme merle
11	14	23	13

In addition, there were isolated matings of extreme dapple to full coat and extreme dapple to dapple.

SEX IN RELATION TO MERLING

The sex distribution of dappled animals shown in Table 2 (*a*) does not depart significantly from the 1:1 ratio. It was forty-five males and fifty-two females for the dappled dachshunds, eleven males and ten females for the merled collies, and eight and four for the merled miniature collies. There is likewise nothing to suggest that the ratio of dappled to undappled amongst the offspring is influenced by the sex of the dappled parent in matings of dapple to full-coat. This is shown in the following summary table:

Parents ...	♂ Dappled × undappled ♀						♂ Undappled × dappled ♀					
	Offspring											
	Dappled			Undappled			Dappled			Undappled		
	M.	F.	T.	M.	F.	T.	M.	F.	T.	M.	F.	T.
Dachshund	32	42	74	40	45	85	13	10	23	7	10	17
Collies	5	6	11	5	6	11	6	4	10	6	7	13
Miniature collies	1	—	1	1	1	2	7	4	11	10	4	14

3. DISCUSSION

PREVIOUS STUDIES ON THE DOG

Dappling and merling in dogs have attracted attention not only because of the unusual coloration but also because of the striking fact that markedly abnormal offspring may arise from matings of such animals. Studies are available on the dachshund, the dunker-hound, the collie, the foxhound, and the great Dane.

(1) *The dachshund*

Wriedt in 1923 recognized that dappling in the dachshund is due to a dominant gene. He held that it was expressed against black or brown but not against red, and appreciated that 'white tigers' arise only from the mating of 'tiger' to 'tiger'. Two years later Anker

in a study on the relation of the different coat colours gave extensive breeding data extracted from registers, which may be summarized thus:

Mating	Offspring: Undappled	Offspring: Dappled	Offspring: 'White tigers'
Dapple × dapple	15	22	6
Non-dapple × white tiger	2	3	1
Non-dapple × dapple	110	86	—

(2) *The Norwegian dunkerhound*

The occurrence of white dunkerhounds was noted by Wriedt (1919, 1925 and 1930). The white animal is described as white dappled (white with irregular grey spots). It is given as derived from reciprocal crosses of grey dappled with small marking (i.e. merling). Such crosses produce ¼ black (or black and tan), ½ the common grey dapples, and ¼ dappled white. White dunkerhounds are homozygous as shown by cross-breeding, for all offspring of self-colour with white dapple are grey dapples. The white dapple shows blue irides; the pupils are oblique, and microphthalmos with blindness may be present. Glaucoma has been observed in old white dapples. Deafness is frequent and resistance to disease poor.

(3) *The collie*

Mitchell (1935) held that the blue merle was conditioned by a single dominant gene for dilution (**M**) upon the bicolour pattern and that **MM** animals were white. He gives the following distribution among seventy-nine offspring of crosses between blue merles: white collies 20, blue merles 39, and tricolour 20. The white collies are described as having eyes that are pale blue, almost china white, and more or less sightless—anophthalmos being present in extreme cases. When hearing is affected, there is generally total deafness. His own white collie proved fertile.

(4) *The foxhound*

Phillips & Knight (1938) observed two 'calico' hounds—sibs (brother and sister) derived from a cross between a black and tan hound and a cream 'calico' bitch. This pair of calico hounds had dark eyes and good hearing. Two matings are reported:

(1) Mated to each other they produced three calico hounds with a great deal of white, all of which had china eyes and were deaf; one calico hound with one china eye and a suggestion of white on the toes of the forefeet was also deaf; there was in addition, one sound full colour with dark eyes.

(2) The calico dog mated to a full-coloured bitch (black saddle on a tan background) produced normal-hearing black and tan calicos (one of which had one china eye and one dark eye); a tan calico which was deaf and had china eyes; two self tans and one black and tan—all with dark eyes and good hearing.

They also report that other breeders found that matings of calico to calico will sometimes produce a defective white, and that deaf dogs with a large proportion of white and both eyes china are found in every litter of such matings.

(5) *Other dogs*

There is hearsay evidence on the occurrence of merling and anomalies in other breeds of dogs. Thus Mitchell (1935) notes that blue merling similar to that seen in the collie occurs in several breeds which may originally have intermingled with the collie, and lists

amongst these the old English sheepdog, the Shetland sheepdog, some collie-like British herding dogs as well as the great Dane 'and possibly the dappled dachshund'.

We have heard of merles amongst Cardiganshire corgis, the merled strain having apparently died out during the war years. Likewise merling is reputed to occur in the Queensland sheepdog (reputedly an indigenous animal); and blind and deaf white dogs are said to be found amongst the offspring of crosses between such merles.

(6) *The great Dane*

Apart from dappling and merling discussed so far, there are observations on the occurrence of 'white dogs' amongst the offspring of harlequin Danes mated to each other. This was noted by Little & Jones in 1919, whilst Nachtsheim (1940) drew attention to ocular anomalies seen in such animals.

These studies may be taken to establish that:

(1) Merling occurs in the dachshund, the Norwegian dunkerhund, collie and foxhound. The merled strain can be maintained by mating merle to self-colour; the offspring consists of both these types, apparently in equal proportions.

(2) In the offspring of merle to merle in these four breeds 'white dogs' have been observed in addition to self-coloured and merles.

(3) Such white dogs have abnormal eyes and are sometimes deaf.

(4) Except for 'wall eye', which is an inconstant occurrence, no marked anomalies are recorded for merled animals.

(5) The results of breeding merle to self-colour and merle to merle are consistent with the assumption of a dominant factor **M** as responsible for merling. In addition to merling in the heterozygotes the factor sometimes also produces iris discoloration (wall eye). The homozygous animal is markedly abnormal in coat colour, eyes and ears.

In addition, there are observations on the great Dane. The offspring of harlequin mated to harlequin include 'white dogs' with ocular anomalies.

PRESENT STUDY

As can be seen from Table 1 dappled dogs—the dachshund, the collie and the miniature collie—consistently show an anomaly of the tapetum with 'wall eye' as a less constant feature, and engorged episcleral veins as an occasional occurrence. There is no direct relationship between the severity of the tapetal anomaly and anomalies of the iris. In contrast these breeds show completely normal fundi and irides when their coat is self-coloured. The three extreme dappled dachshunds seen all showed grossly abnormal eyes: microphthalmos with marked iris anomalies, vitreous haze and circumscribed defects in the choroid.

Genetics

(a) *Mode of inheritance*

The results recorded in Table 2 and summarized previously have shown that when full-coat is mated to merle the offspring are of the same two types in a distribution consistent with the 1:1 ratio (102:97 in dachshunds; 24:21 in collies; and 16:12 in miniature collies). There were three types in the offspring of merle mated to merle, viz. full-coat, merle, and 'white dogs', the distribution being consistent with 1:2:1 ratio (14:23:13 for the dachshunds and merled collies).

These results therefore suggest that merling is indeed conditioned by a single factor which had been designated **M**. This factor is dominant, or rather intermediate in effect, the heterozygote being a healthy animal, whilst the homozygote is grossly pathological.

There are, however, two difficulties in this reading, as can be seen from Table 2:

(i) There were three merle to merle matings in miniature collies resulting in fourteen offspring, amongst which there were no white dogs, though $3\frac{1}{2}$ were to be expected. This deficiency may, however, have been a chance occurrence (or represent a lethal effect of the **M** gene in the duplex state; it is unlikely to have been a faulty record by the breeder).

(ii) Whilst a mating of extreme dapple to dapple gave one dapple and one extreme dapple—a distribution according to expectation—a mating between a white dachshund and a full-coat animal is reputed to have resulted in two full-coat animals in addition to two dapples. However, it has already been pointed out that these apparent full-coats were not examined. The record is not altogether satisfactory.

(*b*) *The effects of the* **M** *gene*

In the simplex state. The two constant effects of the **M** gene are merling of the hair and abnormality of the tapetum. Both these effects vary from animal to animal, for the degree of merling is variable and so is the extent of the tapetal anomaly. Both effects are, however, marked and obvious. Two effects are inconstant: one of these, heterochromia iridis, is fairly common and has been seen in the three breeds studied; the other—engorged episcleral veins—is infrequent and in this series has been seen only in the dachshund.

It is unlikely that the tapetal anomaly and wall eye in the heterozygotes studied are produced by a different gene from that producing merling, if only because of the constant association of merling with tapetal anomaly. The following observation on a mongrel is also instructive.

A stray mongrel like a heavily built fox terrier had a brown coat except for patches of dappling similar to that seen in the chocolate dappled dachshund. Whilst the irides were brown, the fundi were devoid of tapeta.

In the duplex state. There is a variable pleiotropic effect. The most marked effect produces a white dog, but in the dachshund, at any rate, dirty greyish dark patches are commonly present. The eyes are malformed, and in extreme cases there is marked microphthalmos. The ocular anomaly is obvious when the eyes first open, but apparently progresses with age. We do not know how frequently deafness occurs. There were no other obvious physical defects in the three white dachshunds seen, and the post-mortem examination of one white dachshund two months old revealed nothing of general interest.

The nature of the ocular anomaly presents some difficulty in interpretation. Clinically the eye is small, the cornea shows opacities, the lens may be dislocated, but the most marked changes are in the iris and choroid. The iris is obviously hypoplastic and the choroid shows localized defects of the 'atypical coloboma' type in addition to the lack of tapetum. One would expect these anomalies to correspond histologically to vascular defects and to defects in the pigment layers of the choroid and iris. This is discussed elsewhere (Lucas 1954).

(c) Location of the M gene

(i) Relation to the genes for coat colour

Assuming that black is allelic and dominant to chocolate (Winge, 1951; Burns, 1952) it is clear that dappling is not a member of this series, because the litters observed showed features that were consistent with the interaction of two independent genes and inconsistent with a monofactorial rearrangement. The more significant findings were:

Six matings involving chocolate to silver dapple. Here the results were:

(1) 1 chocolate and 2 chocolate dapples.
(2) 1 chocolate and 1 chocolate dapple.
(3) 3 black and 1 silver dapple.
(4) 3 black and 1 silver dapple.
(5) 3 black and 1 silver dapple.
(6) 3 silver dapples.

In the first two of these matings the silver dapple must have carried black on one chromosome with chocolate as the allelic gene, for chocolate offspring occurred. The gene for dappling cannot therefore be an allele to full colour. In the remaining four matings silver dapples were seen among the offspring, and these could not have arisen if the gene for dappling were allelic to that for full colour: only chocolate dapples would be seen.

Seven matings involving a red animal and a silver dapple. The results were:

(1) Two reds, two silver dapples, and one red dapple.
(2) Four reds, one silver dapple.
(3) Three silver dapples.
(4) Three reds, one black.
(5) Three 'full-coat', and one red dapple.
(6) One black and two red dapples.
(7) Two reds.

On the assumption that red is recessive to black, the silver dapples seen in the first three litters could not occur if the gene for dappling were allelic to that for full colour; nor could the red animals observed in (1), (2), (4) and (7). (The offspring shown in (6) and (7) were from the same parents.)

(ii) Possibility of linkage with the gene for colour

In the thirteen matings involving a silver dapple, assuming once more that red is recessive to black and chocolate to red, the distribution observed in the offspring is such that crossing-over has to be assumed in six instances if the gene for dappling were linked with the gene for colour. Location on another chromosome is therefore more likely.

(iii) Possibility of location on the sex chromosomes

The **M** gene is obviously not located on the X-chromosome, for in addition to three extreme dapples we observed, there are records of six more. Amongst the nine extreme dapples both sexes were represented (six males and three females). This distribution does not, of course, dispose of the possibility that the gene is carried on the homologous portion of the X- and Y-chromosomes. Such an assumption is, however, untenable, for a merled dog would be carrying the **M** gene on either its X-chromosome or Y-chromosome. In the

latter case only its sons would be dappled, whilst if the gene is carried by the X-chromosome only daughters would be dappled. Actually the records of thirty matings show that in twenty-three more than one dapple was present in the litter; of these twenty-three, no less than seventeen matings resulted in dapples of both sexes. The autosomal location of the gene is therefore evident.

Wall eye and tapetal anomaly in other breeds

(*a*) *Breeds with coat pattern suggestive of merling*

In the three breeds studied wall eye is present only in heterozygotes; the homozygotes for full colour have dark eyes. There are several homozygous breeds whose coat pattern suggests merling; the old English sheepdog, the roan spaniel and the blue Kerry terrier. These were examined ophthalmoscopically with the following results.

The old English sheepdog (*bob-tail*). At birth the sheepdog is black and white. As the first coat is shed the black areas are replaced by merle. The top hair in the merled area is banded. Beneath it is a coat of fine soft white hair. Four animals were seen showing wall eye on one or both sides; all wall eyes lacked a tapetum. Three animals with normal irides showed normal tapeta. No animal showed a tapetal or iris anomaly by itself. Wall eye is reputed to be common in this breed.

The roan spaniel. The roan spaniel is either black or white when born, the roan pattern appearing when the puppy coat is shed. The coat of the adult is a mixture of black and white hairs. A blue tapetum was present in two dogs examined. Only two cases of wall eye were known to one of the principal breeders; they were two bitches in a litter of three (the third being a dog with normal irides), deriving from a blue roan sire and a black and white dam.

The blue Kerry terrier. The animals are born black, except that occasionally there is a spot of brown on the leg. In the adult the coat is a mixture of black and white hairs. No case of wall eye was heard of in this breed. The tapetum in three adults examined was blue.

(*b*) *Breeds with coat pattern showing spotting*

It has already been noted that 'white Danes' with ocular anomalies appear when harlequin Danes are mated to each other. There is considerable evidence that the harlequin pattern is the equivalent of, or is associated with, merling. This will be discussed in a separate study. Here it is enough to stress the following observations:

(i) Harlequin Danes show the same tapetal and iris anomalies as dappled dachshunds or merled collies.

(ii) Breeding results also appear to be similar, for the ocular anomaly of the white Dane is probably as severe as that of the white dachshund.

(iii) The ragged patches of the harlequin recall not merling but the spotted pattern of the dalmatian, the English setter, and the springers—breeds that are homozygous (in contrast to the heterozygous harlequin). As with homozygous animals with a coat pattern suggestive of merling, these homozygous spotted animals occasionally show tapetal and iris anomalies.

Analogous conditions in other species and man

Dappling or merling in species other than the dog is known, but not fully studied. It would appear that dappling requires clear differentiating from the roan pattern seen in the roan spaniel and other breeds, as also in the mouse, guinea-pig and in roan cattle. Nothing definite is known of bodily anomalies of such animals, except in the case of the mouse. In *Mus musculus*, Cloudman & Bunker (1945) have described the triad of disturbance in equilibrium, involuntary head movements, and deafness. Early in life the heterozygote shows the roan effect, but later on the darker areas gradually fade, so that the mouse becomes white. In the homozygote all these anomalies are exaggerated. The microphthalmic mouse described by Hertwig (1942) shows in the homozygous state coloboma of the retina with reduction in the size of the eye, virtually complete absence of secondary bone absorption (Grüneberg, 1948), and both fur and eyes are devoid of colour. In the heterozygous condition there is dilution of colour of the eye, whilst the colour of the fur is not appreciably affected, except that under suitable genetic conditions there is much tail spotting, a blaze on the forehead and often a belly spot. The rather complicated genetic background is discussed by Grüneberg (1953).

In the cat, Bamber (1933) has drawn attention to several old observations on the association of white coat with blue eyes and deafness, and has shown that white deaf cats need not necessarily have both eyes blue, and that white, blue-eyed cats are not necessarily deaf. She believed the white colour to be inherited in a dominant manner, and that the syndrome, whether fully or partially expressed, occurs in the heterozygote—a view that agrees with the breeding results recorded by Przibram (1908). There is evidence (Jones, 1922) that in the duplex state the gene is lethal *in utero*. Blue eye is apparently the counterpart of the wall eye of the dog. Histological observations on the inner ears and the eyes of white, blue-eyed, deaf cats have shown bony changes in the labyrinth (Alexander, 1900; Alexander & Tandler, 1905), and lack of pigment in the uveal tract (Lauber, 1906).

We ourselves have seen three white cats, and made the following observations:

	Iris	Tapetum	Hearing
(1) Right eye	Yellow	Normal	Probably normal
Left eye	Yellow	Normal	
(2) Right eye	Total 'wall eye'	Absent	Probably normal
Left eye	Yellow	Normal	
(3) Right eye	Yellow	Normal	Probably normal
Left eye	Yellow	Normal	

The one eye with iris and tapetal anomalies was normal in other respects and apparently fully functioning. Genetic data could not be obtained, nor could breeding be undertaken.

Data on animals with black spotting or marking—as distinct from the white spotting of the mouse and the white coat of the cat—are more plentiful. Black spotting is reminiscent of the harlequin pattern of the great Dane rather than the merling of other breeds of dogs considered.

For the rabbit there are the extensive studies of Nachtsheim (1932, 1933 and 1935). Two types of dark marking are recognized, the flecked recessive type seen in the Dutch rabbit, and the patchy dominant type seen in the English rabbit. 'Wall eye' is common in the Dutch rabbit. English rabbits, even those that are almost white, show only dark eyes, whilst in the Dutch rabbit wall eye is frequent. The iris discoloration in the Dutch

rabbit is proportional to lack of colour in the coat, the rabbit that is almost white always shows blue eyes or iris bicolor. Histologically only the anterior mesodermal layer of the iris is devoid of colour; the posterior ectodermal layers of the choroid retain their pigment. By crossing English with Dutch, the coat pattern is that of the English rabbit, but the wall eye of the Dutch variety now occurs. Blue eye is also seen in the white Vienna rabbit, the anterior layers of the iris being devoid of colour. Crossed with the Himalayan rabbit, it is possible to transfer wall eye to the Himalayan.

In the horse wall eye likewise occurs constantly in the recessive type of piebald. It occurs only occasionally in the dominant type and in self-colour (Klemola, 1931, 1933).

Wall eye is said to occur in some 5–7 % of pigs (Dürr, 1937; Koby, 1923; Rieger, 1951). It does not appear to be related to any particular coat pattern. Merling in pigs is known from a study by Haring & Schaaf (1951), but there are no details on the state of the eyes. Merling is apparently the normal feature of the silver fox (Holceck-Hollschowitz, 1937).

Apart from the occurrence in mammals, there is a somewhat similar occurrence in pigeons (Wriedt, 1930). In these birds there is a factor reducing red colour to yellow, and black to grey. These yellow and grey pigeons when newly hatched are deficient in down on their bodies, and such birds are almost naked if in addition to the colour dilution factor there is also the 'almond' character (irregular spots on the separate feathers). Whilst the pigeons with the dilution factors tend to be weakly, those also having the 'almond' factor show considerable mortality. Ocular anomalies—some said to be similar to those seen in the white dog—also occur.

The closest parallel to merling in dogs appears to be observed in man. The inheritance of white forelock as a dominant trait is an old-established observation. There are also records of the dominant inheritance of heterochromia iridum; likewise there are records of the inheritance of deaf mutism in a dominant manner as an exceptional variant to the very much more common recessive mode of inheritance. The literature contains references to the occurrence of heterochromia iridum and partial albinism as complications of deaf mutism in man (Hammerschlag, 1907; and Urbantschitz, 1910), and a pedigree extending over four generations has been published by Mende (1925); here deaf mutism was frequently combined with white patches on the skin, white forelock and poliosis. Klein (1947, 1950) reported a most striking association of this type together with extensive skeletal and muscular anomalies. The fullest study has come from Waardenburg (1951), who has shown that white forelock, heterochromia iridum, deaf mutism, together with a malformation in the upper part of the face giving a lateral displacement of the medial canthi, blepharophimosis, prominent broad root of the nose, and growing together of the eyebrows at their medial portions, constitute a definite syndrome inherited in a dominant manner. Waardenburg gives the different components as possessing different penetrance. White forelock has a penetrance of only 17 %, deaf mutism 20 %, and heterochromia iridum 25 %. The subsidiary features of the syndrome are considerably more constant: growing together of the eyebrows was present in 45 %, a broad root of the nose in 78 %, and displacement of the puncti and canthi in 99 % of the fourteen families with 164 affected relatives of probands he studied.

The available evidence suggests that the gene for merling, and the anomalies associated with it, is probably present in several species besides the dog and man. If the genes in the dog and in man are indeed corresponding factors, it would seem that the heterozygous state in the dog produces little more than a pleasant physiological variant, whilst in man

it produces one of the milder facial anomalies, but may also give severe pathological disturbances including deaf mutism. The homozygous dog is a grossly pathological animal. The homozygous condition in man is not known—unless the exceptionally severe case reported by Klein represents the homozygous state—a reading for which there is no support in the family history. The partial observations recorded on other species are too meagre to indicate whether the gene is indeed present over a wide range of animals, and whether it is related to the albino series.

SUMMARY

1. 'Wall eye'—partial or total discoloration of the iris—is known to occur frequently but not constantly in dappled dachshunds, merled collies, and other merled breeds of dog. Observations are recorded on the dappled dachshund, the merled collie, and the merled miniature collie, showing that in contrast to the inconstant iris anomaly the tapetum of such animals is always anomalous. It is generally totally lacking and when present it is rudimentary. The anomalies of the tapetum are easily observed with the ophthalmoscope.

2. It is known that the dappled or merled animal is heterozygous, and it has been suggested that there is a gene **M** responsible for merling. Evidence is brought forward to show that the gene is intermediate in effect, independent from the allelic series for coat colour, autosomal in location, and productive of gross ocular anomalies in the homozygote, which also shows marked depigmentation of the coat and frequently deafness as well. In the simplex state the gene is therefore productive of 'physiological' variations, whilst in the duplex state its effects are pathological.

3. Attention is drawn to somewhat similar occurrences in the great Dane, and to the possibility that a gene of this type is widely distributed over different species. The relationship of the appearances seen in the dog to the syndrome described by Waardenburg in man is discussed.

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