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≪**Review**≫

Mutations of Japanese Quail (*Coturnix japonica*) and Recent Advances of Molecular Genetics for This Species

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Japanese quail (*Coturnix japonica*) is a valuable bird for both research and production. From a mainly biomedical research point of view, this article describes characteristics of mutations of Japanese quail that have been found in the last 20 years, along with the recent (10 or 15 years) advances in molecular genetics of Japanese quail. In the 50 years following the first report of a mutation in Japanese quail in 1940, approximately 50 mutations were found, and during the subsequent 20 years, approximately 25 mutations have been added to the list of mutations in Japanese quail to date. Based on advances in molecular genetics of Japanese quail, two main genetic linkage maps have been constructed, mostly using microsatellite DNA and amplified fragment length polymorphism (AFLP) markers, and several mutations have been solved at the molecular level for their causative genes. It is anticipated that molecular studies will further reveal the nature of Japanese quail in the near future and enhance its value as a laboratory research animal and agricultural (industrial) animal.

Key words: causative gene, gene mapping, genetic linkage map, Japanese quail, mutation

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Introduction

Wild Japanese quail (*Coturnix japonica*), belonging to the order Galliformes and the family Phasianidae, inhabits the eastern part of the Eurasian continent, including Hainan and Taiwan, and the islands of Japan. In each of the continent and the islands of Japan, this species migrates from the northern area to the southern area in winter season and from the southern area to the northern area in summer season (Yamashina, 1961; Taka-Tsukasa, 1967; Kawahara, 1978; Wakasugi, 1984). It is generally thought that Japanese quail was first domesticated in Japan around the 15th century as a pet song bird. Although the true origin and onset of domestication are unclear (Kawahara, 1978; Wakasugi, 1984; Crawford, 1990), it is apparently documented that domesticated Japanese quail were reared in Japan around the 17th century as a pet bird for song and plumage colors (Soseidoh-Syujin, 1649). Thereafter, at the beginning of the 1900s, the improvement and use of Japanese quail for egg production began in Japan (Oda, 1917; Ohmori, 1918). Today, it is widely reared for egg and meat production not only in Japan, but also in several countries of Asia, Europe, and America (Minvielle, 2004).

Besides rearing for eggs/meat production, Japanese quail is also used as an experimental animal. The first report in which Japanese quail was genetically examined was published by Shimakura (1940). Shimakura (1940) reported the first plumage color mutation in Japanese quail, brown-splashed white, and recognized the advantages of using Japanese quail as a laboratory research animal. However, his proposal was not widely recognized throughout the world, probably because the paper was written in Japanese and submitted to a Japanese journal. About 20 years later, Padgett and Ivey (1959) and Wilson *et al.* (1961) again proposed the advantages of Japanese quail as a laboratory research animal. Since then, use of Japanese quail has become wide spread in the biomedical research fields (Homma, 1970; Minvielle, 2004). The merits of this bird are found in its small body size, short generation turnover, and high egg production.

In the genetic research fields employing Japanese quail, a number of morphological, biochemical, and other mutations have been discovered (Somes, 1988; Cheng and Kimura, 1990). The studies on biochemical (*e.g.,* isozymes) and immunological (*e.g.,* blood types) traits were performed from the 1960s to the 1980s with the zenith in the 1970s. Morphological mutants were also actively studied from the 1960s to the 1990s, but the number of studies on classical mutants decreased after 2000. Instead of these classical studies, modern molecular techniques began to be adapted to quail genomes from the 1990s onwards. In chickens, a number of microsatellite and other kinds of DNA markers were developed in the 1990s, and a comprehensive genetic linkage map was constructed

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(Groenen *et al.,* 2000; Schmid *et al.,* 2000). On the other hand, over 100 microsatellite markers and a number of other kinds of DNA markers were also developed in Japanese quail from 2000 to 2005, and later in chickens, genetic linkage maps based on these markers were constructed (Kayang *et al.,* 2006; Sasazaki *et al.,* 2006a).

Biochemical and immunological variants of Japanese quail have been well summarized by Kimura (1982), Somes (1988), and Cheng and Kimura (1990), along with morphological, behavioral (neurological), and other kinds of mutations by Somes (1988) and Cheng and Kimura (1990). It is now 20 years since these reviews were done. During this 20 years however, there are very few biochemical and immunological variants that have been newly discovered. Therefore in this article, the author mostly reviews morphological, behavioral (neurological), and other kinds of mutants that have been discovered in Japanese quail during the past 20 years, along with recent developments in molecular genetics for Japanese quail.

Mutations in Japanese Quail

Table 1 is a list of morphological, behavioral, and some other mutations in Japanese quail that have so far been discovered. Among the loci controlling these mutations, some are multiple-allelic loci, which are also summarized in Table 2. The author herein describes features of the Japanese quail mutations that were found during the last 20 years, with those of some older mutations. Mutations have been categorized as morphological, behavioral or muscular, and other mutations. Morphological mutations have been further classified into plumage color, plumage system, eggshell color, and morphogenetic mutations. Gene symbols presented here are according to those described in each original report, although there is some confusion in nomenclature of mutant genes.

Morphological Mutations

Plumage Color Mutations

Dilute down lethal (*ddl*) (Tsudzuki *et al.,* 1997): This autosomal recessive plumage color mutation was found in 1994. The neonatal mutant chick has the same striped plumage pattern as the wild type. However, the plumage color is slightly lighter than the wild type. The brownish stripes of the wild type are replaced by slightly lighter brownish or yellowish colored stripes. In the wings and thighs of the mutant chick, blurry black markings replace the dense black markings of the wild type. Besides the down color abnormality, 25% of the mutant chicks have bent digits. All the mutant chicks die within three days of hatching.

Dotted white (s^{dw}) (Tsudzuki *et al.,* 1992): This is an autosomal recessive plumage color mutation discovered in 1982. The mutant chick has creamy yellow down plumules with a brown spot on the head and/or back. 12% of the mutant chicks have no colored spot. The creamy yellow and brown plumules of the chick replace white and dark feathers respectively in their adulthood. In the dark feathers, the whole pigmentation is diluted and the wheat-straw colored shaft streak is narrower than the wild type. Furthermore, the transversal bars in individual feathers are completely missing. The dotted white is a bottom recessive allele at the panda (*s*) locus (Mizutani *et al.,* 1974; Tsudzuki *et al.,* 1993). The panda is also a mutant that shows white plumage with dark patches on the head, cheek, back, wing, and tail. In contrast with the dotted white, in the panda mutant the feathers from the dark patches are similar to the wild type in both color and markings (Tsudzuki *et al.,* 1993).

Fawn-2 (Y^{f2}) (Tsudzuki *et al.,* 1996): This is an autosomal incompletely dominant mutation that occurred at the yellow (*Y*) locus (Homma *et al.,* 1967). The neonatal plumage of the homozygote shows a creamy yellow color all over the body with three dark brownish obscure stripes on the back, and no stripes on the head. However, faint dark pigmentation is observed at the tip of some head plumules. In the chicks heterozygous for this mutation, the creamy color is slightly deeper than that of the homozygote. The three stripes on the back are more blackish in color and clearer than those of the homozygote. Sex differences are large in adult plumage of both the homozygote and heterozygote. The homozygous male shows whitish light-brown plumage with a few small black speckles on the back, while the homozygous female shows more creamy plumage than the male and has a relatively large number of black speckles on the back. Irrespective of the yellow mutant, the homozygote of the fawn-2 mutant is fully viable. The face of the homozygous male is rusty with the crown composed of dark and creamy feathers, while the face of the homozygous female is creamy with sparsely dotted black pigmentation. In the heterozygote, the primary plumage pattern is basically the same as that of the homozygote, but the brown color is deeper in the heterozygote than in the homozygote in each sex. The female plumage of the heterozygote has a much larger amount of black pigmentation than the homozygous female, somewhat similar to the yellow mutant plumage. The phenotype and mode of inheritance of the fawn-2 mutant are similar to those of the fawn mutant (Nichols and Cheng, 1988) that is an allele at the yellow locus (Cheng and Kimura, 1990). Furthermore, Minvielle *et al.* (2003) described that the beige mutant is also similar to the fawn mutant both in the phenotype and mode of inheritance. Thus, there is a possibility that these three (fawn, fawn-2, and beige) are identical mutants, or alleles at the yellow locus.

Lavender (formerly bleu) (*LAV*L,* formerly, *bl*) (Minvielle *et al.,* 2002, 2003): This is one of the dilution mutations that was renamed by Minvielle *et al.* (2002, 2003). The previous mutant name was bleu (Cheng and Kimura, 1990). The adult mutant shows bluish gray all over the body with the same primary plumage pattern as the wild type. The secondary plumage pattern (the marking in individual feathers) is also the same as that of the wild type. However, in individual contour feathers, the

Table 1. **Summary of mutations found in Japanese quail (*Coturnix japonica*)[1]**

Category of traits	Mutant names	Mode of inheritance[2]	Gene symbols[3]	References
Morphological mutations				
Plumage color	Beige[4]	AD	*FAW*BE*	Minvielle *et al.* (2003)
	Black[5]	AD	*D*	Chikamune and Kanai (1978), Somes (1984)
	Black at hatch	AD	*Bh*	Minezawa and Wakasugi (1977)
	Brown-splashed white	AR	*p*	Shimakura (1940)
	Buff[6]	AR	*pk*	Sittmann *et al.* (1966)
	Cinnamon[6]	AR	*cin*	Fulton *et al.* (1982a)
	Complete albino	AR	*a*	Homma *et al.* (1968)
	Dilute down lethal	AR	*ddl*	Tsudzuki *et al.* (1997)
	Dotted white[7]	AR	s^{dw}	Tsudzuki *et al.* (1992, 1993)
	Extended brown[5]	AD	*E*	Somes (1979), Truax and Johnson (1979)
	Fawn[4]	AD	Y^F	Cheng and Kimura (1990)
	Fawn-2[4]	AD	Y^{f2}	Tsudzuki *et al.* (1996)
	Imperfect albinism	SR	*al*	Lauber (1964), Homma *et al.* (1968)
	Lavender (Bleu)	AR	*LAV*L*	Minvielle *et al.* (2002, 2003)
	Light down	AD	*L*	Ito and Tsudzuki (1993)
	Light down lethal	AR	*ldl*	Tsudzuki (1995b)
	Marbled plumage	AR	*ma*	Yakovlev *et al.* (1975)
	Orange[6]	AR	*or*	Ito and Tsudzuki (1994)
	Panda	AR	*s*	Mizutani *et al.* (1974)
	Pansy[8]	AR	d^{ps}	Tsudzuki *et al.* (1990)
	Recessive black	AR	*Y*RB*	Fujiwara *et al.* (2005), Hiragaki *et al.* (2008)
	Recessive silver	AR	*rs*	Homma *et al.* (1985)
	Recessive white[7]	AR	*wh*	Roberts *et al.* (1978), Somes (1979)
	Redhead[8]	AR	e^{rh}	Truax *et al.* (1979)
	Roux	SR	*BR*R*	Minvielle *et al.* (2003)
	Rusty	AR	*RU*R*	Minvielle *et al.* (2005a)
	Sex-linked brown	SR	*br*	Wakasugi and Kondo (1973)
	Sex-linked cinnamon[9]	SR	al^c	Wakasugi and Kondo (1973)
	Silver	AD	*B*	Homma *et al.* (1969)
	White[10]	AD	*W*	Wakasugi and Kondo (1973)
	White bib (white crescent)	AR	*bi* (*cr*)	Roberts *et al.* (1978)
	White breasted	AR	*wb*	Roberts *et al.* (1978)
	White primaries	AR	*wp*	Somes (1984)
	White-feathered down	AR	*c*	Sittmann and Abplanalp (1965)
	Yellow	AD	*Y*	Homma *et al.* (1967)
Plumage system	Curly	AR	*CU*C*	Minvielle *et al.* (2005a)
	Defective feathers	AD, AR	*Df, mdf*	Fulton *et al.* (1983)
	Downless	AR	*dl-1, dl-2*	Savage and Collins (1971a)
	Fray	AD, AR	*Fr, mod*	Tsudzuki *et al.* (1998c)
	Partial featherlessness	AD	*Pf*	This article
	Porcupine	AR	*pc*	Fulton *et al.* (1982b)
	Rough-textured	AR	*rt*	Roberts and Fulton (1979)
	Ruffle	AR	*rf*	Somes (1988)
	Short barb	AR	*sb*	Fulton *et al.* (1982c)
Eggshell color	Celadon	AR	*ce*	Ito *et al.* (1993)
	Red eggshell	AD	*R*	Hardiman *et al.* (1975)
	White eggshell	AR	*we*	Poole (1964)

brownish color area is replaced with bluish gray and the wheat-straw color markings with a whitish appearance.

Light down (*L*) (Ito and Tsudzuki, 1993): This plumage color mutation is controlled by an incompletely dominant autosomal gene. The homozygotes, a third of which die before hatching, show creamy yellow down plumules. The eye of the homozygotes is not reddish, but the diameter of the pupils is much larger than that of the heterozygotes. Consequently, the iris area of the homozygotes is extremely small. All of the homozygous chicks that hatched show behavioral abnormalities with variable expressivity. Mildly affected chicks slowly shake their heads right and left, while severely affected chicks rigidly bend the neck inward and subsequently, either draw backward

Table 1. **(Continuation) Summary of mutations found in Japanese quail (*Coturnix japonica*)[1)]**

Category of traits	Mutant names	Mode of inheritance[2)]	Gene symbols[3)]	References
Morphogenesis	Abnormal head	AD	*ab*	Morse and Abplanalp (1970), Somes (1988)
	Arostroperocephaly	AR	*ar*	Dodson and Coleman (1973)
	Chondrodystrophy	AR	*ch*	Collins *et al.* (1968)
	Chondrodystrophy-2	AR	*ch-2*	Hermes *et al.* (1990)
	Crooked neck dwalf	AR	*cn*	Sittmann and Craig (1967)
	Deformed beak	AR	*db*	Cheng and Kimura (1990)
	Ear tuft	AR	*hfd*	Tsudzuki and Wakasugi (1988b)
	Hereditary multiple malformation	AR	*hmm*	Tsudzuki *et al.* (1998a)
	Long beak	AR	*lb*	Cheng and Kimura (1990)
	Micromelia	AR	*m*	Hill *et al.* (1963)
	Short beak	AR	*sbk*	Tsudzuki *et al.* (1998b)
	Short lower beak	AR	*slb-1, slb-2*	Nakane and Tsudzuki (1998c)
	Stumpy limb	AR	*sl*	Tsudzuki (1995a)
	Throat tuft	AR	hfd^{Tt}	Tsudzuki and Wakasugi (1989)
	Zazen	AD, SR	*Zn, moz*	Nakane and Tsudzuki (1998b)
Behavioral or muscular mutations	Back drawer	AR	*bkd-1, bkd-2*	Tsudzuki and Wakasugi (1988a)
	Circular	AR	*cr*	Mizutani (2000)
	Congenital loco	AR	*lo*	Sittmann *et al.* (1965)
	Crooked neck	unknown	—	Dodson and Coleman (1972)
	Dark feather nervous disorder	AR	*dn*	Kawahara (1979, 1980), Ueda *et al.* (1979)
	Hypotrophic axonopathy	AR	*hax*	Mizutani *et al.* (1992)
	Muscular disorder	AD	*Md*	Braga *et al.* (1995a, b)
	Star gazing	AR	*sg*	Savage and Collins (1972)
	Wry neck	AR	*wyn-1, wyn-2*	Savage and Collins (1971b)
Other mutations	Clench	AR	*cl*	Nakane and Tsudzuki (1998a)
	Diabetes insipidus	AR	*di*	Minvielle *et al.* (2007a)
	Glycogenosis type-II	AR	*gly II*	Nunoya *et al.* (1983)
	Twinning	AD	*Tw*	Sittmann *et al.* (1971)

[1)] Biochemical and immunological variants are not included.
[2)] AD=autosomal dominant over the wild-type allele, AR=autosomal recessive to the wild-type allele, SR=sex-linked recessive to the wild-type allele. Almost all AD essentially are incompletely dominant.
[3)] Including some symbols that were not proposed in each original paper but thereafter proposed by Cheng and Kimura (1990) and this article.
[4)] There is a possibility that the three mutations are identical or alleles at the same locus.
[5)] It is very likely that the two mutations are identical.
[6)] There is a possibility that the three mutations are identical or alleles at the same locus.
[7)] There is a possibility that the two mutations are identical or alleles at the same locus.
[8)] There is a possibility that the two mutations are identical or alleles at the same locus.
[9)] There are two synonyms for this mutation, such as red-eyed brown and dark-eyed dilute (Cheng and Kimura, 1990).
[10)] There are three synonyms for this mutation, such as dominant white, autosomal dilute, and dominant dilute (Cheng and Kimura, 1990).

or roll forward. Some severely affected chicks draw the head backward with the beak pointing upward. All homozygous chicks that could hatch die within a week of hatching. The neonatal plumage of the heterozygotes is similar to that of the wild type. However, the black stripes on the head and back are somewhat narrower than those of the wild type, and their color is slightly diluted. Consequently, the brownish (yellowish) stripes are broader than those of the wild type, and the color is also lighter. The whole plumage of the adult heterozygotes is also similar to that of the wild type, but its color is slightly diluted all over the body just like in the chicks (Fig. 1A). In the individual feathers of the adult heterozygotes, the number of transversal bars is increased. For example, in the back feathers, this mutant has four pairs of transversal bars, while the wild type has three pairs.

Light down lethal (*ldl*) (Tsudzuki, 1995b): This plumage color mutant is an autosomal recessive with nearly total lethality. The neonatal plumage pattern of this mutant is the same as that of the wild type. However, black and tan stripes of the wild type are replaced with blurry black and dilute tan stripes, respectively. Almost all of these mutants that could hatch die by three weeks of age, with the majority dying within one week. Moreover, prehatch mortality is high (37%) in this mutant. Among the 103 light down lethal chicks, only two males survived to adulthood and reproduced normally. The adult mutant phenotype is very similar to that of the wild type.

Table 2. **A list of multiple-allelic loci on morphological characters**

Loci[1)]	Alleles[2)]	References
Black	$D > d^{+} > d^{ps}$	Tsudzuki *et al.* (1990)
Extended brown	$E > e^{+} > e^{rh}$	Truax *et al.* (1979)
Hyomandibular furrow closure defect	$Hfd^{+} > hfd^{Tt} > hfd$	Tsudzuki and Wakasugi (1989)
Imperfect albinism	$Al^{+} > al^{c} > al$	Wakasugi and Kondo (1973), Cheng and Kimura (1990)
Panda	$S^{+} > s > s^{dw}$	Tsudzuki *et al.* (1993)
Sex-linked brown	$BR{*}N > BR{*}B > BR{*}R$	Minvielle *et al.* (2000b, 2003)
Yellow	$Y^{f2} > Y > y^{+} > Y{*}RB$	Tsudzuki *et al.* (1996), Hiragaki *et al.* (2008)
	$Y^{F} > Y > y^{+}$	Cheng and Kimura (1990)

[1)] The black locus is very likely to be identical to the extended brown locus.
[2)] Descriptions of the gene symbols are based on those that appeared in each original report.

Orange (*or*) (Ito and Tsudzuki, 1994): This is an autosomal recessive mutation. The plumage pattern of this mutant is the same as that of the wild type, but the pigmentation is obviously lightened in both chicks and adults. The mutant chicks have chocolate brown and light brownish-yellow stripes on the head and back. In the adult orange males, light reddish-brown is predominant on the whole body (Fig. 1B). The color is especially conspicuous on the face and breast. In contrast, in the adult orange females, the light reddish-brown color is less marked. The eye of the orange mutant is reddish when held to the light. Hatchability of the orange mutant is significantly lower than that of the wild type because of the increased prehatching mortality, but fertility in this mutant is normal. Furthermore, posthatch mortality within five weeks of age is greatly increased in the orange mutant over that of the wild type (60% *vs.* 8%). There is a possibility that the orange mutant is identical to the buff (Sittmann *et al.,* 1966) or the cinnamon mutant (Fulton *et al.,* 1982a). Ito and Tsudzuki (1994) did not propose a gene symbol for this mutation in their report. Here, for the sake of convenience, the author proposes the gene symbol *or,* because this mutation was first reported in 1994. The symbol may be *OR*OR,* when adopting the new nomenclature proposed by Crittenden *et al.* (1996).

Pansy (d^{ps}) (Tsudzuki and Wakasugi, 1987; Tsudzuki *et al.,* 1990): The mutant chicks have light yellow plumules with three black stripes on the back. The black stripes are narrower and more obscure than those of the wild type. The head plumules are brownish when compared to the light yellow color seen on the back. The plumage pattern of the adults is totally different from that of the wild type. A shaft stream and transversal bars seen in the wild-type feathers are absent in the pansy mutant. Adult plumage shows a mixture of rust, black, and white (Fig. 1C). In the male plumage, the face, chin, and throat are brown or heavy rust in color. There are large variations in neonatal and adult plumage. The neonatal plumage in some chicks is somewhat similar to that of the fawn-2 (Tsudzuki *et al.,* 1996), and consequently some adult males resemble homozygous fawn-2 males in plumage color. Such a type of the pansy male shows fawn color all over the back with a small number of black spots. The pansy mutant is controlled by a bottom recessive allele (d^{ps}) at the black (D) locus (Chikamune and Kanai, 1978; Somes, 1984). The pansy plumage resembles the redhead plumage which is a bottom recessive allele (e^{rh}) at the extended brown (E) locus (Somes, 1979; Truax *et al.,* 1979). As described in a later section, the black mutant will be identical to the extended brown mutant. Thus, there is a possibility that the pansy and redhead mutants are identical mutants, and that at least these two are alleles.

Recessive black (*Y*RB*) (Fujiwara *et al.,* 2005): This mutant was first reported by Fujiwara *et al.* (2005), and the gene symbol *rb* was assigned. Later, Hiragaki *et al.* (2008) found the allelism of this mutation at the yellow (Y) locus (Homma *et al.,* 1967). This is a bottom recessive allele at the yellow locus. Both sexes of this mutant exhibit blackish (or dark brownish) plumage throughout the body that is very similar to the plumage of the black (Chikamune and Kanai, 1978; Somes, 1984) or the extended brown mutant (Somes, 1979; Truax *et al.,* 1979). In individual feathers on the back of this mutant, the wheat-straw colored shaft stream seen in the wild-type feathers is missing, although transversal bars are present as in the wild type. However, the width of the transversal bars becomes narrower when compared with that of the wild type, which gives it a blackish or dark brownish appearance in this mutant. This is the first black colored mutation occurring at the agouti signaling protein (*ASIP*) locus in poultry (Hiragaki *et al.,* 2008), and is also described in a later section.

Roux (*BR*R*) (Minvielle *et al.,* 2000b, 2003): This is a sex-linked mutation that belongs to the sex-linked brown (*br* or recently *BR*) locus (Wakasugi and Kondo, 1973; Cheng and Kimura, 1990) with a bottom recessive effect. The brown locus is linked to the imperfect albinism locus, and the recombination rate of 35% was first reported by Wakasugi and Kondo (1973) and of 38% by Minvielle *et al.* (2000b) between the two loci. The plumage color of the roux mutant is also brownish as is the brown mutant, but is paler than that of the brown mutant. Judging from the color photo by Minvielle *et al.* (2000b), the plumage color of the roux mutant is grayish light brown, rather

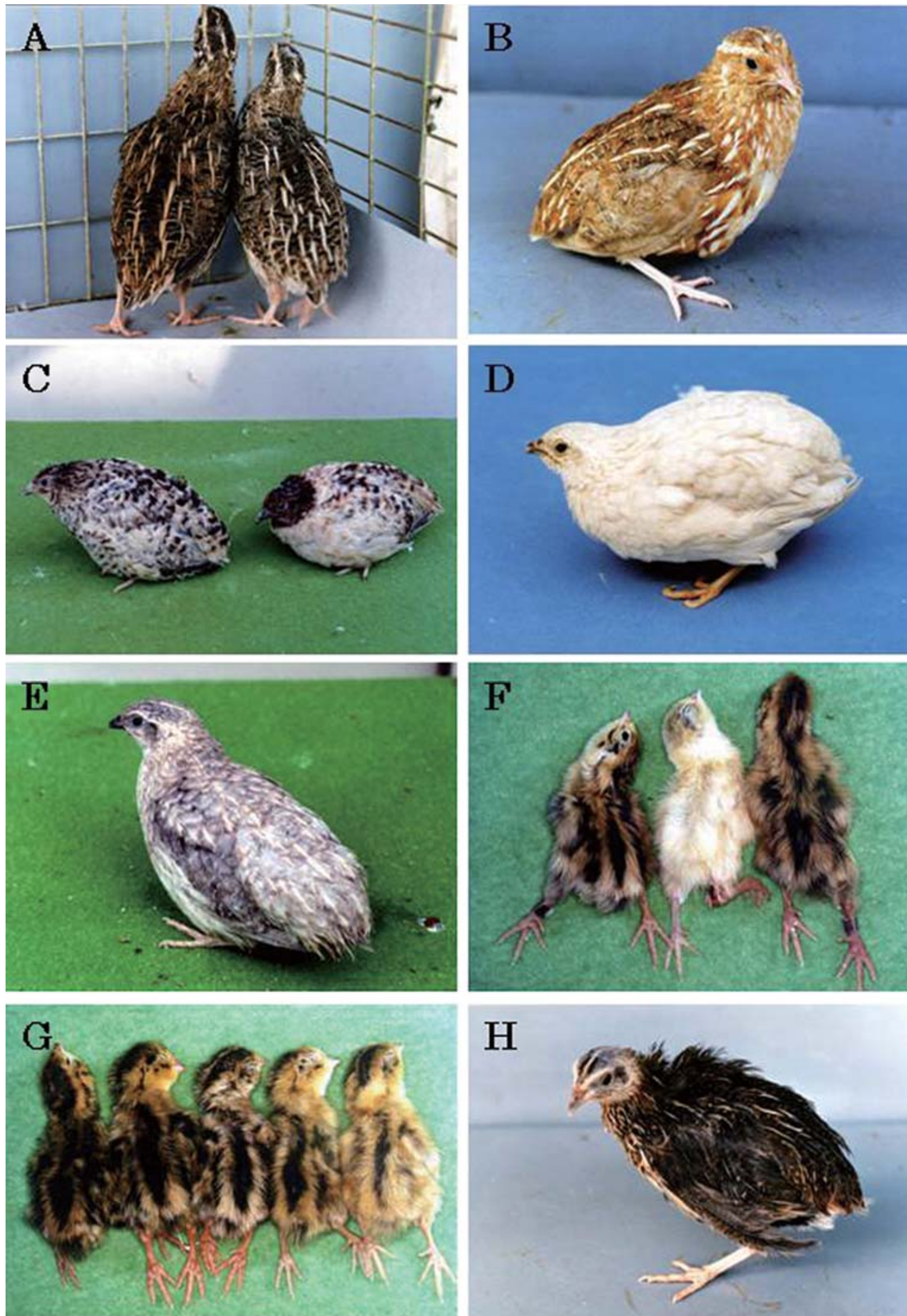

Fig. 1. **Several morphological and behavioral mutants in Japanese quail.** (A) Comparison of the wild-type (left) and light-down (L/l^+) mutant (right) quail. (B) A male of the orange (*or/or*) mutant quail. (C) A female (left) and male (right) of the pansy (d^{ps}/d^{ps}) mutant quail. (D) The homozygote for the white mutation (*W/W*). (E) The heterozygote for the white mutation (W/w^+). (F) Homozygous (middle) and heterozygous (left and right) chicks for the white (*W*) mutation. (G) Wild-type (left) and mutant chicks (others) heterozygous for the *W* mutation. The neonatal mutant plumage is variable in color. (H) The fray mutant (Fr/fr^+, *mod/mod*) with mild expressivity. (I) The fray mutant with intermediate expressivity. (J) The fray mutant with severe expressivity. (K) A black-at-hatch mutant male. (L) The ventral surface of the partial featherlessness mutants. (M) Comparison of the normal (left) and stumpy-limb (*sl/sl*) mutant (right) quail. (N) The back-drawer (*bkd-1/bkd-1, bkd-2/bkd-2*) mutant exhibiting severe abnormality. (O) The heterozygote (left) and homozygote (right) for the silver (*B*) mutation. (P) The red-eyed brown (=sex-linked cinnamon, al^c) mutant.

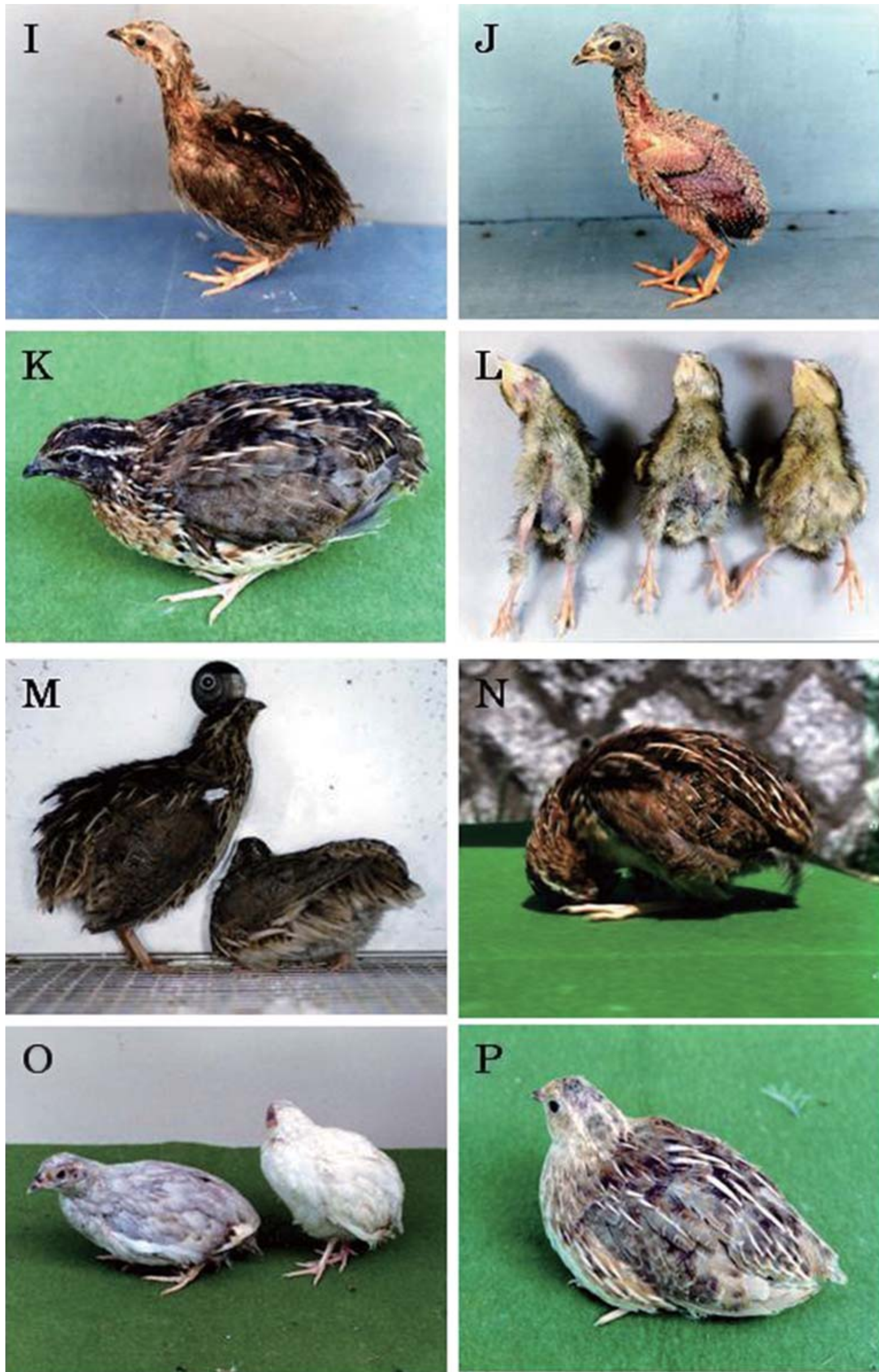

Fig. 1. **(continued)**

similar to that of the sex-linked cinnamon. The roux plumage shows significant association with 3% lower body weight and 3% less abdominal fat pad. Egg production ability is normal, although egg weight is 2% lower (Minvielle *et al.,* 1999).

Rusty (*RU*R*) (Minvielle *et al.,* 2005a): This autosomal recessive plumage color mutant shows reddish-brown (rusty) color all over the body. The individual contour feathers show rusty color in the pennaceous portion. However, the plumulaceous portion shows a slate like color as in the wild type, differing from the roux mutant (Minvielle *et al.,* 2000b, 2003) in which rusty color is seen all over the contour feather. The rusty color is heavier in the rusty mutant than in the roux mutant. Hatchability of the rusty mutant is normal.

White (*W*) (Wakasugi and Kondo, 1973): This is an autosomal dominant plumage color mutation. The mutant name is assigned to the plumage appearing in the homozygotes (Fig. 1D). Very few homozygotes for this mutation survive to adulthood and show white plumage all over the body. However, according to the observations made by the author, the white is not pure white but has extremely diluted gray tinges in the outer margin of individual contour feathers. The adult plumage color of the heterozygotes, which is fully viable, is brownish gray with the wild-type plumage pattern (Fig 1E). The density of the color is variable from individual to individual. Some are similar to that of the wild type and cannot be distinguishable from the wild-type plumage. In the most grayish case, the wild-type plumage pattern becomes obscure. The chicks homozygous for this mutation have dusty creamy yellow plumules accompanied by dark skin and crooked toes (Fig. 1F). Almost all homozygotes die around hatching. In the heterozygous chicks, the plumage pattern is the same as that of the wild type, but black and tan stripes seen in the wild type are replaced with grayish and dusty yellowish stripes, respectively (Fig 1G). According to Cheng and Kimura (1990), the white mutant is homologous to the autosomal dilute mutant (Somes, 1984) and the dominant dilute mutant (Truax and Johnson, 1979).

Plumage System Mutations

Curly (*CU*C*) (Minvielle *et al.,* 2005a): This is an autosomal recessive mutation. The adult plumage of the curly mutant shows a fluffier appearance than the wild-type plumage. The curly phenotype results from the connected calamus of the growing wing feathers at chick days. The expressivity of the mutant plumage is variable and the penetrance of the mutant gene is not complete. Postnatal survivability of this mutant is normal.

Fray (*Fr, mod*) (Tsudzuki *et al.,* 1998c): This mutant is characterized by curly, short, and sparse plumules in chicks and underdevelopment of barbs and barbules in adults. There is a large variability in the degree of severity of the mutant phenotype (Fig. 1H, I, J). In the most severe case (Fig. 1J), both chicks and adults have no plumules or feathers and are almost totally bare. This mutant trait is controlled by the combination of an autosomal dominant mutant gene (*Fr*) and another autosomal recessive modifier gene (*mod*). The *Fr* gene has a complete lethal effect in the homozygous condition. The recessive modifier (*mod*) is epistatic to *Fr,* and the homozygous state of the modifier allows the expression of the *Fr* gene; that is, only the Fr/fr^{+} *mod/mod* genotype exhibits the fray mutant phenotype. Furthermore, the *Fr* and *mod* are closely linked on the same chromosome (Tsudzuki *et al.,* 1998c). The fray mutant is similar to the defective feathers mutant in the phenotype and mode of inheritance of two-locus control (Fulton *et al.,* 1983). However, the two loci controlling the defective feathers mutant trait seem to be located at different chromosomes (Fulton *et al.,* 1983). Therefore, the two mutants are different.

Partial featherlessness (*Pf*): In 1995, the author discovered phenotypically unusual chicks among the black-at-hatch (*Bh*) mutant (Minezawa and Wakasugi, 1977) quail (Fig. 1K) flock kept by the Department of Laboratory Animal Science, Faculty of Agriculture, Osaka Prefecture University. In the mutant chicks, feather buds did not extend in the limited areas of the hind head, back, abdomen, thigh, and chin (Fig. 1L). Although such chicks showed high mortality, some survived to adulthood and were able to reproduce. No feathers grew in the areas that were bare at the neonatal chick days. From the matings with the wild-type colored normal birds and the birds that have the black-at-hatch plumage and partial featherlessness, wild-type colored chicks that have partial featherlessness appeared with low incidence, although partial featherlessness was nearly always exhibited accompanied by black-at-hatch plumage. This result strongly suggests that the mutant locus controlling the partial featherlessness is closely linked to the *Bh* locus, and that the gene is an autosomal dominant.

Eggshell Color Mutations

Celadon (*ce*) (Ito *et al.,* 1993): The name was assigned to this eggshell color mutation because of the similarity of the eggshell color to that of the porcelain in the time of the Sung, China. This mutation is controlled by an autosomal recessive gene and is hypostatic to the white eggshell (*we*) mutation (Poole, 1964). The celadon eggshell possesses the pigments protoporphyrin and biliverdin, as does the wild-type eggshell. However, the amount of the protoporphyrin in the celadon eggshell is much smaller than in the wild type (5.6% of the wild type). The biliverdin content of the celadon eggshell is 44% of that in the wild-type eggshell.

Morphogenetic Mutations

Ear tuft (*hfd*) (Tsudzuki and Wakasugi, 1988b): This is a morphogenetic mutation controlled by an autosomal recessive gene *hfd* (hyomandibular furrow closure defect) with incomplete penetrance. The *hfd* is a bottom recessive

allele at the *hfd* locus (Tsudzuki and Wakasugi, 1989). This mutation is characterized by ear-opening abnormality and an ear tuft, which is composed of an epidermal peduncle and covering feathers, protruding within or below the abnormal ear opening. Abnormal ear openings are oval-shaped and accompanied by fissures of varying size on the ventral margin. The incidence of the ear tuft and ear-opening abnormality is 33% and 42% respectively in the *hfd* homozygotes. These abnormalities are attributed to the hyomandibular furrow closure (or visceral arch) defect occurring at five days of incubation. The incidence of the hyomandibular furrow closure defect is 91% in the homozygotes. However, the defect that occurred at five days of incubation is rapidly repaired by six days of incubation in approximately a half of the affected embryos, which results in the low (42%) incidence of the ear-opening abnormality at later stages (Tsudzuki and Wakasugi, 1988d). The early embryonic abnormalities occurred around the visceral arch region also give rise to head skeletal abnormalities; that is, partial deletion or irregularity is seen in the mandible, basiparasphenoid, quadratojugal, squamosal, and quadrate bones. The incidence of the head skeletal abnormalities is 87% in the *hfd* homozygotes (Tsudzuki and Wakasugi, 1988c).

Hereditary multiple malformation (*hmm*) (Tsudzuki *et al.,* 1998a): This autosomal recessive mutation is the first polydactyl mutation found in Japanese quail. No mutant embryos survive beyond 15 days of incubation, with the peak of death at the sixth day of incubation. Mutant embryos at late incubation stages (13–15 days) show an early embryo-like body shape, and have no plumules, although feather buds are seen in their body. In the mutant head, the eyelid is left wide open. The upper and lower beaks are greatly deformed and set apart. The wings and legs are stumpy and showed syndactylous polydactyly. In the abdomen, a part of the ventriculus, liver, and small intestine protrudes out of the umbilicus region. The skeleton of the mutant embryos shows underdevelopment and/or morphogenetic abnormalities all over the body. In the mutant skeleton, ossification is generally delayed, and cartilage is predominant throughout the body except the head. Delay of the ossification is most remarkable in the fore and hind limbs.

Short beak (*sbk*) (Tsudzuki *et al.,* 1998b): This mutant was found in 1994. The majority of the short beak mutants die at the late embryonic stages and within three days of hatching. In this mutant, the length of the beak, shanks, and digits is shortened to approximately 90% of that of the wild type, although there is no significant difference in body weight between the mutant and wild type. Irrespective of the shortening, the *sbk* shank is thickened to approximately 110% of the wild type. The shortened beak shows no parrot-like shape, contrasting with other poultry condrodystrophic mutants (Hays, 1944; Collins *et al.,* 1968; Nestor, 1978; Hermes *et al.,* 1990; Tsudzuki *et al.,* 1991). Some of the *sbk* mutants (5.3%) reach sexual maturity and are able to reproduce. This mutant is not allelic to a similar mutant, stumpy limb (*sl*) (Tsudzuki, 1995a)

Short lower beak (*slb-1, slb-2*) (Nakane and Tsudzuki, 1998c): This mutant was found in 1996. This morphogenetic mutant dies during late embryonic stages. The mutant embryos at 15 days of incubation are first characterized by a shortened lower beak. The severity of the shortening varies largely. In the most severe case, the length of the lower beak is approximately one third of the upper beak, and in mild cases the lower beak is slightly shorter than the upper beak. Almost all mutant embryos keep the mouth wide open. In the skull of the mutant embryos, the Meckel's cartilage of the mandible is abnormally bent downward in its proximal portion, and ossification of the mandibular bone occurs around the abnormal Meckel's cartilage, which seems to be responsible for the shortened lower beak and the opened mouth. In addition to these abnormalities, this mutant shows hypoplasia or partial lack of the hyoid apparatus, abnormal formation and/or fusion of the sternal ribs, and increased number of cervical vertebrae. This mutant trait seems to be controlled by two autosomal recessive genes that are linked on the same chromosome, the recombination rate of which is approximately 30%. However, further genetic studies will be needed to confirm the mode of inheritance of this mutation.

Stumpy limb (*sl*) (Tsudzuki *et al.,* 1991; Tsudzuki, 1995 a): This mutation was discovered in 1985 as an embryonic completely lethal mutation. However, changing of genetic backgrounds gave rise to viable adult mutants (Fig. 1M) (Tsudzuki, 1995a). Late embryos and chicks are characterized by brachycephaly, short beak, thick neck, and short and thick extremities. At the skeleton level, the mutant exhibits globular skull, unusual curvature of the *Processus palatinus maxillaris* of the upper beak, and shortening and thickening of the appendicular bones. Some embryos show a bending of the humerus, femur and/or tibiotarsus. Abnormalities are more conspicuous in the leg bones than in the wing bones. In the UOP-WT genetic background (Ito and Tsudzuki, 1994), 56% of the affected embryos could hatch out of the eggshell with no assistance, and could walk in a relatively smooth manner. 73% of the chicks that could hatch died at various ages before reaching maturity with a mortality peak at three days of age. Of these, the birds that survived for relatively long periods started showing difficulty in walking around two weeks of age and subsequently died at various ages. The remaining (27%) survived maturity, but the birds that sexually matured were only 12% of the hatched mutant chicks. Although reproductive ability of these sexually matured mutant birds varied from individual to individual due to the degree of the difficulty in walking, spermatogenesis and oogenesis seemed to progress normally in the gonads of the sexually developed *sl* mutants.

Throat tuft (hfd^{Tt}) (Tsudzuki and Wakasugi, 1989): This mutant shows ear-opening abnormality and a feathered tuft as seen in the ear-tufted (*hfd*) mutant (Tsudzuki

and Wakasugi, 1988b), and is autosomally recessive to the wild type and dominant over the *hfd* mutation. As seen in the case of the *hfd,* the penetrance of the hfd^{Tt} allele is also not complete. The tuft originates in the throat region and is composed of an epidermal peduncle and covering feathers. Some birds possessing these throat tufts exhibit abnormal ear-openings, in which either a wide cleft or a short fissure is formed at the ventral margin. The incidence of the throat tuft and ear-opening abnormality is 50% and 17%, respectively. Throat tufts are usually accompanied by normal ear openings, contrasting with the case of the ear-tuft mutant. Furthermore, the position from which peduncles protrude in the hfd^{Tt} mutant is different from that of the *hfd* mutant. There is also a difference in abnormal morphology of the ear opening between the two. The ear-tuft mutants exhibit a fissure below the ear opening, whereas the throat-tuft mutants express a wide cleft, much wider than the fissure of the ear-tuft mutants, at the ventral margin. Throat tufts and ear-opening abnormalities are also attributed to visceral arch defects seen in the embryos at five days of age, as in the case of the ear-tuft mutants. The incidence of the defects is 38%, greatly lower than that of the ear-tuft mutant. Incomplete closure of the hyomandibular furrow is also repaired in some affected embryos. However, abnormal morphology in the visceral arch regions is different between the two mutants, which leads to partial deletion or irregularity in the head skeleton of the throat-tuft mutant with different severity and incidence of each bone from the case of the ear-tuft mutant (Tsudzuki and Wakasugi, 1990b). Irrespective of the different incidence of each bone abnormality, the whole incidence of the head skeletal abonormality of the throat-tuft mutant is 88%, similar to that (87%) of the ear-tuft mutant (Tsudzuki and Wakasugi, 1990a, b). Comparative studies on the head skeletal abnormalities in the ear-tuft and throat-tuft mutants suggested that the *hfd* gene influences the dorsal region of the visceral arches with stronger intensity than its allele hfd^{Tt}. In contrast, hfd^{Tt} appears to have a greater effect than *hfd* on developmental events associated with the third arch and the ventral or median ventral regions of the first and second arches (Tsudzuki and Wakasugi, 1990 b). Interestingly, the incidence of the head skeletal abnormality is significantly lower in the hfd/hfd^{Tt} heterozygotes than in the hfd/hfd and hfd^{Tt}/hfd^{Tt} homozygotes (Tsudzuki and Wakasugi, 1991).

Zazen (*Zn, moz*) (Nakane and Tsudzuki, 1998b): This mutation was found in 1996. The mutant name is based on the leg condition that is similar to the figure of religious meditation (=zazen) in the Zen sect of Buddhism. This mutant shows complete lethality at the late embryonic stages. The 15-day mutant embryos cross their legs in the front of the abdomen around the yolk sack. In about half the mutant embryos, the crus is joined to the shank at an abnormal angle, which results in the shank turning toward the backside. In almost all (94%) of the mutant embryos, the eyelid is left wide open. In addition to the leg and eye abnormalities, some mutants show shortening of the lower beak with upward bending of its tip, lack of the edges around the basal part of the upper beak, and lateral curvature of the upper beak. In the legs, all the mutant embryos have full-length fibula from the stifle articulation to the tibiotarsal-tarsometatarsal articulation. The femur of the mutant embryos is articulated with the proximal end of the fibula, whereas in the normal embryos the femur is articulated with the tibia. In the mutant head, the maxillary and the palatine processes of the premaxilla are extremely reduced in length, which seems to result in the lack of the basal edges of the upper beak. Almost all zazen mutants show lack of the parietal and abnormally formed ribs. The joint between the sternal and vertebral ribs is fused and/or ossified. The vertebral ribs gain or lose one rib in number, and the ribs themselves are abnormally shaped and/or deformed. This mutation seems to be controlled by two genes; one is an autosomal dominant mutant gene (*Zn*) and the other is a sex-linked recessive modifier (epistatic) gene (*moz*) that allows the expression of the *Zn* in the *moz/moz* state. However, further genetic studies will be necessary to confirm the mode of inheritance of this mutation.

Behavioral or Muscular Mutations

Back drawer (*bkd-1, bkd-2*) (Tsudzuki and Wakasugi, 1988a): The onset of the expression of the abnormal behavior varies from hatching to eight weeks of age. All individuals that exhibit the abnormality at hatching die within five days. Severely affected birds crouch and draw backward with the head pressing on the floor and the beak pointing toward the abdomen (Fig. 1N). Such birds occasionally roll forward. Intermediately affected birds usually walk forward with neck bending downward and occasionally draw backward or roll forward. Mildly affected birds direct their heads downward slightly and neither draw backward nor roll forward. The mild and intermediate abnormalities appear before and/or after the period of the severe abnormality. Some severely affected birds survive for a long period comparable to normal birds, with gradual recovery from the abnormality. The reproductive ability of the mutants is reduced in both sexes, particularly the laying ability of females. Although this mutation is controlled by two independent autosomal recessive genes, Tsudzuki and Wakasugi (1988a) did not propose a gene symbol for this mutation. Later, Cheng and Kimura (1990) assigned the symbols *bkd-1* and *bkd-2* to the genes.

Circular (*cr*) (Mizutani, 2000): This behavioral mutant shakes its neck and subsequently shows a turning movement right or left. Although the cause of the abnormal behavior is unknown, it has been revealed that in the mutant brain the total amount of proteins is less than that of the normal quail. This mutation is controlled by an autosomal recessive gene. So far, no gene symbol has been assigned to this mutation. Here, the author tentatively assigns the symbol *cr* to this mutation.

Hypotrophic axonopathy (*hax*) (Mizutani *et al.,* 1992): This is an autosomal recessive mutation characterized by neurofilament deficiency. The quail that shows the neurofilament deficiency (hypotrophic axonopathy) is usually known as quiver (Quv) quail (Yamasaki *et al.,* 1991; Mizutani, 2002). The mutants exhibit quivering at the neonatal stage. In mildly affected birds, the quivering is observed only in the head and neck, whereas in severely affected birds it is manifested in the entire body. This symptom is more marked when the affected birds are frightened by some visual or auditory stimuli. Fertility, hatchability, and posthatch mortality are 24.5%, 43.5%, and 19.8%, respectively. Light microscopic morphometry revealed that the mutant has significantly smaller cross-sectional areas of the cervical spinal cord and the optic and sciatic nerves. Furthermore, electron microscopic morphometry indicated that in the cervical spinal cord, myelinated axons in the affected birds are significantly smaller in size than normal, though greater in density. Electron microscopically and immunohistochemically, neurofilaments are not detected in the axons or neuronal cell bodies (Yamasaki *et al.,* 1991). Later on, through electrophoresis and Western blot analysis, Yamasaki *et al.* (1992) revealed that low, middle, and high molecular mass neurofilament subunits are markedly deficient in the brain, cervical spinal cord, and sciatic nerves of the affected quail. Immunohistochemically, the spinal cord of the mutant has no immunoreactive products corresponding to low molecular mass neurofilament.

Muscular disorder (*Md*) (Braga *et al.,* 1995a, b): The mutant quail with this disorder is usually known as LWC quail (Mizutani, 2002). This mutation is characterized by generalized myotonia, muscle stiffness, and muscle weakness, and is classifiable as a type of progressive muscular disorder. Affected birds are externally identified by their inability to lift their wings upward and by their inability to stand upright when placed on their dorsum. These symptoms are clinically observed as early as 28 days of age. The mutant skeletal muscles include characteristic histological lesions found in sarcoplasmic masses, ringed fibers, internal migration of nuclei, and fiber size variation. These lesions appear first in the pectoral region and later in the muscles of the thoracic and pelvic limbs. This mutation is an autosomal dominant with homozygous lethality, but no gene symbol has been assigned to this mutation. Here, the author tentatively assigns the symbol *Md* to this mutation.

Other Mutations

Clench (*cl*) (Nakane and Tsudzuki, 1998a): This mutation was discovered in 1995. The abnormality exhibited by this mutant is limited to the toes. The mutants have rigidly clenched toes in both legs at hatching, which gives rise to a human fist-like appearance in the distal end of the legs. Both chicks and adults can stand and walk. In spite of such abnormality, this mutant can reproduce normally.

Diabetes insipidus (*di*) (Minvielle *et al.,* 2007a): This is the first nephrogenic diabetes insipidus mutation reported in Japanese quail. This mutation is inherited as an autosomal recessive trait. Minvielle *et al.* (2007a) found the quail showed excessive drinking and excessive urination, and carried out experiments on plasma levels and brain mRNA contents for avian Arg vasotocin. As a result, they found that these were affected little by the mutation, however plasma avian Arg vasotocin was 13-fold higher and brain mRNA contents were significantly increased in both normal and mutant quail following a 24-hour water deprivation. The mutant quail shows similar performance to the normal quail in the traits such as egg production and quality, feed intake, and gross carcass traits. However, residual feed consumption is higher in the mutant.

Practical Utilization of Quail Mutants

Among the Japanese quail mutants that are listed in Table 1 or described in this article, black-at-hatch (Fig. 1 K), glycogenesis type-II, hypotrophic axonopathy, imperfect albinism, muscular disorder, and silver (Fig. 1O) have been frequently utilized as biomedical animal models, as reported in Ono and Wakasugi (1983a, b), Kubota *et al.* (1995), Satoh *et al.* (1997), Shiojiri *et al.* (1999), and Niwa *et al.* (2002) for the black-at-hatch; Matsui *et al.* (1983), Tsujino *et al.* (1997, 1998), Kikuchi *et al.* (1998), Yang *et al.* (1998), Lin *et al.* (2002), and McVie-Wylie *et al.* (2003) for the glycogenesis type-II; Zhao *et al.* (1993, 1994, 1995), Takahashi *et al.* (1994, 1995), Hasegawa *et al.* (1994, 1996), Hirai *et al.* (1999a, b), Toyoshima *et al.* (2000) and Sheykholeslami *et al.* (2001) for the hypotrophic axnopathy; Dkhissi *et al.* (1994, 1996) and Schrodl *et al.* (2005) for the imperfect albinism; Braga *et al.* (1995a, b) and Tanaka *et al.* (1996a, b) for the muscular disorder; and Araki *et al.* (1998, 2002), Tsukamoto *et al.* (1999) and Kawaguchi *et al.* (2001) for the silver. Besides the mutants mentioned above, other mutants are also potentially powerful resources in biomedical fields to elucidate the regulatory mechanisms for pigmentation, morphogenesis, nervous systems, and metabolism.

In addition to these biomedical uses, the growth and productive ability have also been investigated for the recessive white (Petek *et al.,* 2004), roux (Minvielle *et al.,* 1999, 2000a), and yellow (Minvielle *et al.,* 2007b) from the view point of possible use in agricultural (industrial) fields.

Modern Advances of Molecular Genetics for Japanese Quail

Development of DNA Markers and Genetic Linkage Maps

In 2000, an integrated comprehensive linkage map was constructed in chickens (Groenen *et al.,* 2000; Schmid *et al.,* 2000) on the basis of modern molecular techniques, including 51 linkage groups. This generally resulted from the development of DNA markers, especially with the development of a large number of microsatellite DNA markers. Today, over 900 microsatellite loci have been mapped in the integrated map with 31 chromosomes and

22 linkage groups (Schmid *et al.,* 2005). On the other hand, in 2000, only 4 linkage groups (3 autosoms and Z chromosome) were known in Japanese quail, with a few loci in each linkage group, due to the absence of DNA markers (Wakasugi and Kondo, 1973; Ito *et al.,* 1988a, b; Cheng and Kimura, 1990; Shibata and Abe, 1996).

To obtain microsatellite markers for Japanese quail, a few trials were performed by adapting already developed chicken microsatellite markers to the genome of Japanese quail (Pang *et al.,* 1999; Inoue-Murayama *et al.,* 2001; Kayang *et al.,* 2003). From these trials however, it was concluded that the chicken microsatellite marker primers for polymerase chain reaction (PCR) are not efficient in amplifying Japanese quail microsatellites, therefore microsatellite markers for Japanese quail should be generated from Japanese quail genome itself. Kayang *et al.* (2000, 2002, 2004) developed 100 microsatellite markers directly from the Japanese quail genome and constructed a first-generation microsatellite linkage map that included 72 microsatellite markers (Kayang *et al.,* 2004). The total length of the map was 576 cM, with an average spacing of 10 cM between loci. Besides the microsatellite linkage map, Roussot *et al.* (2003) developed a number of amplified fragment length polymorphism (AFLP) markers and constructed an AFLP-based linkage map with 41 linkage groups (39 autosomal, and Z and W chromosomes), in which the number of AFLP markers included, the total length of the map, and the average spacing between markers were 258, 1516 cM, and 7.6 cM, respectively. Later, Kayang *et al.* (2006) integrated the microsatellite map of Kayang *et al.* (2004) and the AFLP map to one linkage map of 904.3 cM, with an average spacing between loci of 9.7 cM. Furthermore, Kayang *et al.* (2006) assigned the Japanese quail chromosomes (CJA) to chicken chromosomes (GGA) by a comparative mapping between the two species. As a result, CJA01 to 07, 09, 10, 13, 14, 18, 20, 27 and Z have been assigned to GGA 01 to 07, 09, 10, 13, 14, 18, 20, 27 and Z, respectively.

Besides the linkage map of Kayang *et al.* (2006), Kikuchi *et al.* (2005) constructed a genetic linkage map based on a large number of AFLP markers, together with several orthologous chicken microsatellite markers to Japanese quail and two phenotipic loci, and tried some comparisons between the Japanese quail AFLP linkage groups and chicken chromosomes. The total length of the AFLP linkage map was 2816 cM with an average marker interval of 5.5 cM. Subsequently, Sasazaki *et al.* (2006a) improved the AFLP map by increasing the number of AFLP markers. Moreover, in addition to the AFLP markers, Sasazaki *et al.* (2006a) developed expressed sequence tag (EST) markers by using chicken gene sequence information on the web site, and located the EST markers on the AFLP map. Mannen *et al.* (2005) also developed microsatellite markers on the basis of the sequence of cDNA, and mapped them on the same AFLP map. With the mapping information of the EST markers and the microsatellite markers derived from cDNA, Mannen *et al.* (2005) and Sasazaki *et al.* (2006a) assigned some Japanese quail AFLP linkage groups to chicken chromosomes. In addition, with the same reference family used by Kikuchi *et al.* (2005), Mannen *et al.* (2005), and Sasazaki *et al.* (2006a), Sasazaki *et al.* (2006b) mapped the orthologous genes/ESTs of chickens on the Japanese quail genome, and made direct comparisons between the Japanese quail linkage groups and chicken chromosomes. However, there are some discrepancies between the studies (Kikuchi *et al.,* 2005; Mannen *et al.,* 2005; Sasazaki *et al.,* 2006a, b) in assigning Japanese quail AFLP linkage groups to chicken chromosomes. Apart from the correctness of the assignment, the AFLP linkage map, which was first reported by Kikuchi *et al.* (2005), has become a map of 3199 cM in total length, with an average marker interval of 5.0 cM, by the work of Sasazaki *et al.* (2006b). This map is composed of 1995 markers, including 1933 AFLP markers, three phenotypic loci, and 59 genes/ESTs. These are assigned to 66 linkage groups including W chromosome (Sasazaki *et al.,* 2006b). As mentioned earlier, there are at present two main genetic linkage maps for Japanese quail in the world. It is desired that the two maps should be integrated in the near future.

Linkage Groups for Phenotypic Loci, Functional Genes, and ESTs

Table 3 summarizes linkage relationships between loci (markers) that were revealed on the basis of modern molecular techniques, along with classical linkage relationships. As mentioned earlier, in 2000 there were only three autosomal linkage groups and a Z-chromosamal linkage group composed of a small number of classical mutant phenotypic loci and biochemical trait loci, and the locus order in each linkage group, and the chromosome number, were unclear. The linkage relationships that had been known in 2000 are: extended brown (*E*) and phosphoglucose isomerase (*PGI*) (Ito *et al.,* 1988a); panda (*s*), serum albumin (*ALB*), and vitamin D binding protein (*GC*) (Ito *et al.,* 1988b; Shibata and Abe, 1996); Yellow (*Y*) and white breasted (*wb*) (Cheng and Kimura, 1990); and imperfect albinism (*al*) and sex-linked brown (*br*) (Wakasugi and Kondo, 1973). Today, the number of linkage groups that include phenotypic traits, genes, and ESTs is approximately 20, and the chromosome number has already been assigned to the linkage groups in most cases. However, it is difficult to declare the exact number of linkage groups, because there are some discrepancies in the assignment of linkage groups to chromosomes as mentioned before. The classical phenotypic loci that have been mapped with DNA markers are black-at-hatch plumage (*Bh*), panda plumage (*s*), yellow plumage (*Y*), hypotrophic axonopathy (*hax*), transferring (*Tf*), haemoglobin (*Hb-1*), and prealbumin-1 (*Pa-1*). They, respectively, were mapped on chromosomes/linkage groups CJA 01 (Niwa *et al.,* 2003; Miwa *et al.,* 2005), CJA04 (Miwa *et al.,* 2006), CJA20 (Miwa *et al.,* 2005; Kayang *et al.,* 2006), JQG05 (Kikuchi *et al.,* 2005), CJA09 (Miwa *et al.,*

Table 3. **A list of linkage groups for phenotypic traits, genes, and ESTs in Japanese quail**

Chromosomes/ linkage groups[1)]	Loci/markers mapped[2)]	References
CJA01	*Bh, Pf, SEMA3E, IFR1, HAL, FLIK, DMD, LOC395530, ChEST83p14, GLUR4/D, WNT11, TYR*	Niwa *et al.* (2003), Miwa *et al.* (2005), Kayang *et al.* (2006), Sasazaki *et al.* (2006b), This article
CJA02	*CVIPR, PON2, ER81, AHR, LOC395386, PRL, ChEST293k8, IKAROS, LOC421033, CDH2, LOC420199, MT3-MMP, CDH2, VIPR, BCL2, PRKDC, ROCK1, ZFP161, CATA6, CEBPD, RGS20, MOS, PENK, COPS5, GDAP1, EYA1, PKIA*	Shibusawa *et al.* (2001), Sasazaki *et al.* (2006b), Kayang *et al.* (2006)
CJA03	*LOC396025, UGP2, LOC396240, LOC395531, LOC396192, TLX, LOC421887*	Sasazaki *et al.* (2006b)
CJA04	*s, ALB, GC, BTK, LOC396173, PGK, IRF-2, RPK-1, PCM-1, CEX2*	Ito *et al.* (1988b), Shibata and Abe (1996) Miwa *et al.* (2006), Sasazaki *et al.* (2006b)
CJA05	*DKK3, IGF2, TN1, IP3KA, PSEN1, ChEST56312, MJD1, STK29, CKB*	Sasazaki *et al.* (2006b), Kayang *et al.* (2006)
CJA06	*LOC423612, LOC395602, LOC395834, GDNFRALPHA, BEK*	Sasazaki *et al.* (2006b)
CJA07	*LOC396420, LOC396548, GCG, GIRK1, LOC395400, ABCB6*	Sasazaki *et al.* (2006b), Kayang *et al.* (2006)
CJA08	*LOC395269, LOC424533, LOC424547, LOC395940, DAB1, LEPR*	Sasazaki *et al.* (2006b)
CJA09	*Tf*	Miwa *et al.* (2005), Kayang *et al.* (2006)
CJA14	*Hb-1*	Miwa *et al.* (2005)
CJA18	*FASN*	Kayang *et al.* (2006)
CJA20	*Y, wb*	Cheng and Kimura (1990), Miwa *et al.* (2005) Kayang *et al.* (2006)
QL13	*Pa-1*	Miwa *et al.* (2005)
JQG01	*JGH39, JQH6* (*TNA*), *LOC418843*	Mannen *et al.* (2005), Sasazaki *et al.* (2006a)
JQG02	*LOC421033, CDH2*	Sasazaki *et al.* (2006a)
JQG03	*EST4*	Sasazaki *et al.* (2006a)
JQG05	*hax* (*Quv or NEFL*)	Kikuchi *et al.* (2005)
JQG06	*DKK, LOC423356*	Sasazaki *et al.* (2006a)
JQG08	*LOC416999*	Sasazaki *et al.* (2006a)
JQG15	*LOC419683*	Sasazaki *et al.* (2006a)
JQG19	*JQE3-2*	Mannen *et al.* (2005)
JQG42	*EST9*	Sasazaki *et al.* (2006a)
JQG66	*JQE3-1*	Mannen *et al.* (2005)
unknown	*E, PGI*	Ito *et al.* (1988a)
Z	*al, br*	Wakasugi and Kondo (1973), Minvielle *et al.* (2000b)

[1)] There are some discrepancies between studies in assignment of linkage groups to chromosomes. JQG01 corresponds to CJA01 or 02. JQG02 corresponds to CJA02 or 05. Similarly, JQG05 to CJA05 or 22; JQG06 to CJA05, 06, or 07; and JQG08 to CJA07, 08, or 15.
[2)] Including microsatellite markers derived from genes and ESTs.

2005; Kayang *et al.,* 2006), CJA14 (Miwa *et al.,* 2005), and QL13 (Miwa *et al.,* 2005). In the future, the number of mutant phenotypic loci, genes, and ESTs that are mapped will dramatically increase by using DNA markers and their linkage maps, along with the use of comparative mapping methods between Japanese quail and other species, especially chickens.

Quantitative Trait Locus (QTL) Mapping

In chickens, the first report for QTL mapping appeared in 1998 on the basis of microsatellite DNA markers (Vallejo *et al.,* 1998; van Kaam *et al.,* 1998). Since then, about 700 QTLs have been discovered for economic traits of chickens (Abasht *et al.,* 2006); for example, for growth and carcass traits (Abasht and Lamont, 2007; Rao *et al.,* 2007), meat quality traits (Nadaf *et al.,* 2007), egg quality and egg production traits (Schreiweis *et al.,* 2006; Wright *et al.,* 2006), disease resistance or immune response traits (Cheng *et al.,* 2007; Heifetz *et al.,* 2007), and behavior traits (Buitenhuis *et al.,* 2004; Jensen *et al.,* 2005). The author also reported chicken QTLs for shank length and body weight, which are related to the broiler industry (Tsudzuki *et al.,* 2007).

Contrastingly, in Japanese quail there are at present only a few reports concerning QTLs, although QTL mapping is possible with DNA markers in addition to the mapping of loci for qualitative traits. Beaumont *et al.* (2005) tried to identify QTLs for body weight and 11 kinds of fearfulness-related traits with AFLP markers developed by Roussot *et al.* (2003), and detected experiment-wide significant QTLs for five traits; namely, body weight at 2 weeks of age, the logarithm of tonic im-

mobility, the number of indictions, the number of jumps, and dejections. Furthermore, Minvielle *et al.* (2005b) carried out QTL analysis using microsatellite markers developed by Kayang *et al.* (2004) for 13 traits responsible for growth, feed consumption, egg production, tonic immobility, and body temperature. They found experiment-wide significant QTLs for five traits, *i.e.* egg clutch length, eggshell weight, egg number, feed intake, and residual feed intake.

Causative Genes for Mutant Phenotypes

With the exception of two examples, the causative genes for mutant phenotypes of Japanese quail were unclear at the molecular level in the 20th century, although detection of causative genes has continually been reported on and after 2006. The first and second Japanese quail mutations, on which molecular basis of the abnormality was discovered in the last decade of the 20th century, are "hypotrophic axonopathy (*hax*)" (Mizutani *et al.,* 1992) and "silver (*B*)" (Homma *et al.,* 1969).

The *hax* mutant quail is characterized by quivering behavior and neurofilament deficiency in the axons of the cervical spinal cord and the optic and sciatic nerves (Yamasaki *et al.,* 1991). Ohara *et al.* (1993) found a nonsense mutation in the neurofilament-L (NF-L) gene of the *hax* mutant quail. A point mutation from C to T resides at the nucleotide residue 352, resulting in a stop codon at the position corresponding to amino acid residue 114 of the NF-L protein. The expression level of NF-L mRNA in the *hax* brain drastically decreases to $<5\%$ of that in the wild-type quail.

The homozygote for the silver mutation (*B/B*) has pure white plumage and eye defects (Homma *et al.,* 1969). In the *B/B* eyes, transdifferentiation from the retinal pigment epithelium into the neural retina occurs at the central area (Fuji and Wakasugi, 1993). Mochii *et al.* (1998) indicated that the microphthalmia-associated transcription factor (*Mitf*) gene mutation is responsible for the silver mutation. They revealed that there are two kinds of nucleotide changes leading to amino acid changes in the *Mitf* gene by complete sequencing of the cDNA from the *B/B* retinal pigment epithelial cells. One leads to a substitution from histidine to arginine in the basic region of MITF protein, and the other is a two-base deletion resulting in frame shift and a premature termination of the polypeptide in the region following the zipper region. In spite of having such mutations, the retinal pigment epithelial cells of the *B/B* embryos express normal size *Mitf* mRNA, but do not express normal MITF protein, which causes a partial loss of function of the transactivation of the promoter of the 115-kDa melanosomal matrix protein (*mmp 115*) gene, a kind of pigment-cell-specific genes.

Other Japanese quail mutants, for which the causative molecular mutations were revealed in this century, are extended brown (*E*), pansy (d^{ps}), panda (*s*), imperfect albinism (*al*), sex-linked cinnamon (al^{c}), roux (*BR*R*), yellow (*Y*), and recessive black (*Y*RB*).

The extended brown mutant shows blackish (or dark brownish) plumage throughout the body (Somes, 1979; Truax and Johnson, 1979). Nadeau *et al.* (2006) sequenced an 85 bp segment of the 945-bp single coding exon of melanocortin 1 receptor (*MC1R*) gene of the extended brown from a French flock, and observed perfect association between a non-synonymous substitution (Glu 92Lys) and the mutant plumage. The "black" plumage color mutation (Chikamune and Kanai, 1978; Somes, 1984) also exhibits similar phenotype to the extended brown. Nadeau *et al.* (2006) also sequenced extended brown *MC1R* from a Japanese flock, and found the same substitution as in the case of the extended brown from a French flock. The "extended brown" from a Japanese flock examined by Nadeau *et al.* (2006) is thought to be the "black" mutant discovered in Japan. Thus, it is very likely that the extended brown and black are the same mutants.

Pansy (d^{ps}), which exhibits tri-colored mutant plumage, is a bottom recessive allele at the black (*D*) locus (Tsudzuki and Wakasugi, 1987; Tsudzuki *et al.,* 1990). Additionally, it has been revealed that the pansy has a mutation in the *MC1R* locus (Wada and Tsudzuki, unpublished data; accession no. AB201636 in DDBJ). The pansy mutant has insertion of two extra nucleotides (CG) at codon 26 (nt76insCG), which leads to a premature stop codon and produces truncated transcript by a shift of the reading frame. The redhead mutant (e^{rh}) (Truax *et al.,* 1979) is similar to the pansy in the phenotype, and it is phenotypically unclear whether the two are identical mutants. It is desired to investigate *MC1R* mutation in the redhead, which will precisely determine the sameness or difference between the two mutations.

Panda (*s*) (Mizutani *et al.,* 1974) and dotted white (s^{dw}) (Tsudzuki *et al.,* 1992) mutants show white plumage all over the body with colored spots. The panda is dominant over the dotted white (Tsudzuki *et al.,* 1993). Based on the mapping result of the panda (*s*) locus on Japanese quail chromosome 4 (CJA 4) and the comparative mapping between Japanese quail and chickens, Miwa *et al.* (2006) first suggested the endothelin receptor B (*EDNRB 2*) gene to be a candidate gene for the *s* locus. According to this finding, Miwa *et al.* (2007) subsequently confirmed that the causative gene for the *s* locus is *EDNRB2.* The panda phenotype is associated with a missense mutation at the nucleotide position 995 (c. 995G>A) in the *EDNRB 2,* which leads to amino acid substitution (arginine to histidine change) at the position 332 (p. R332H). On the other hand, no amino acid substitution was found for the dotted white phenotype. The expression level of the *EDNRB2* mRNA was lower and drastically lower in the panda and dotted-white mutants respectively, than in the wild-type quail. In spite of the depression of *EDNRB2* mRNA expression level and the amino acid change in EDNRB2 protein, no significant difference was observed in the number or migration pattern of neural crest cells between the wild-type and panda quail in 2.5-day embryos.

Imperfect albinism (*al*) and sex-linked cinnamon (al^c) (Fig. 1P) are plumage color mutations and alleles at the *al* locus on Z chromosome. The al^c is dominant over the *al* (Cheng and Kimura, 1990). Gunnarsson *et al.* (2007) found that the solute carrier family 45, member 2, protein (*SLC45A2*) gene, is the causative gene for the mutations at the *al* locus. In the imperfect albinism, a transversion from G to T resides at the splice acceptor site just preceding exon 4. The splice site mutation leads to an in-frame skipping of exon 4 of *SLC45A2,* and 47 amino acids are missing in the mature protein. Gunnarson *et al.* (2007) also observed an association of a transition from C to A at nt 287 (Ala72Asp) in exon 1 of *SLC45A2* with the sex-linked cinnamon allele.

The roux (*BR*R*) is a recessive allele to the brown mutation at the brown (*br*) locus on Z chromosome, and exhibits slightly lighter brown color than the brown (Wakasugi and Kondo, 1973; Minvielle *et al.,* 2003). Nadeau *et al.* (2007) revealed that the roux phenotype is expressed by the non-synonymous point mutation that occurred at the nucleotide position 845 (c. 845T>C) of the tyrosinase-related protein 1 (*TYRP1*) gene. The 845T >C results in a change of phenylalanine to serine at amino acid 282 (p. Phe282Ser). The expression level of *TYRP1* is the same between the dorsal skins of the wild-type and roux quail, suggesting that the mutation is not due to a regulatory mutation.

Yellow is a plumage color mutant that shows wheat-straw color throughout the body with basically the same markings as the wild type (Homma, 1967). Nadeau *et al.* (2008) revealed that the yellow is a mutation of the regulatory region of agouti signaling protein (*ASIP*) gene, and is not a mutation caused by coding sequence variation at *ASIP.* The yellow mutation lacks almost the entire coding sequence of two upstream loci, hnRNP associated with lethal yellow (*RALY*) and eukaryotic translation initiation factor 2B (*EIF2B*), which results in the production of a new transcript, because *ASIP* expression is controlled by the *RALY* promoter. These genotypic conditions in the yellow mutant quail are quite similar to those in the yellow mutant (A^y) mouse (Michaud *et al.,* 1993). Unlike the yellow mouse however, no significant difference was observed in the ASIP mRNA expression level in the skins of the yellow and wild-type quail (Nadeau *et al.,* 2008).

Besides the study of Nadeau *et al.* (2008) for the yellow mutant quail, Hiragaki *et al.* (2008) also investigated the coding sequence of the *ASIP* gene in the yellow, fawn-2 (Tsudzuki *et al.,* 1996), and recessive black (Fujiwara *et al.,* 2005) mutants and wild-type quail. The three mutants are alleles at the *Y* locus, and the order of dominance is fawn-2>yellow>wild type>recessive black (Tsudzuki *et al.,* 1996; Hiragaki *et al.,* 2008). The yellow mutant showed no variation in the coding region, which is consistent with the result of Nadeau *et al.* (2008). The recessive black showed 8-bp deletion (c. 373–380 del), causing a frame shift that changes the last six amino acids, including a cysteine residue, and removed the normal stop codon. On the other hand, the fawn-2 had two synonymous single nucleotide polymorphisms (c. 31T>C and c. 100C>T), which raised the possibility that there might be another regulatory mutation in the fawn-2 as seen in the yellow. The expression level of the recessive black *ASIP* is significantly lower than that of the wild type, whereas that of the fawn-2 is drastically higher than the wild-type and yellow quail. The wild-type and yellow quail showed a similar level of expression of *ASIP* to each other as observed by Nadeau *et al.* (2008).

As mentioned earlier, genetic studies of Japanese quail accelerated at the beginning of the 21st century at the molecular level. In future, more numbers of DNA markers and genes both for qualitative and quantitative traits will be mapped on the further integrated map, and further numbers of causative genes for mutant phenotypes will be revealed. Such work will raise the value of Japanese quail to a higher level as a research model in the biomedical fields, and as a bird of economic importance.

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