

August 2018 Newsletter

The Pigeon Genetics Newsletter, News, Views & Comments. The Pigeon Genetics Newsletter, News, Views & Comments.

(Founded by Dr. Willard .F. Hollander)

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The following is a report from the University of Utah regarding "PATTERN" in Pigeons.

***TOPIC* : Introgression of regulatory alleles and a missense coding mutation drive plumage pattern diversity in the rock pigeon. (forwarded to us with permission by Gary Young).**

Anna I Vickrey, Rebecca Bruders , Zev Kronenberg, Emma Mackey, Ryan J. Bohlender, Emily T. Maclary, Raquel Maynez, Edward J. Osborne, Kevin P. Johnson, Chad D. Huff, Mark Yandell, Michael D. Shapiro, (University of Utah U.S.A. , University of Texas, U.S.A., University of Illinois Urbana - Champaign, U.S.A.) Research Article July 17 , 2018 .

Abstract

Birds and other vertebrates display stunning variation in pigmentation patterning, yet the genes controlling this diversity remain largely unknown. Rock pigeons (*Columba livia*) are fundamentally one of four color pattern phenotypes, in decreasing order of melanism: T-check, checker, bar (ancestral), or barless. Using whole-genome scans, we identified *NDP* as a candidate gene for this variation. Allele-specific expression differences in *NDP* indicate *cis*-regulatory divergence between ancestral and melanistic alleles. Sequence comparisons suggest that derived alleles originated in the speckled pigeon (*Columba guinea*), providing a striking example of introgression. In contrast, barless rock pigeons have an increased incidence of vision defects and, like human families with hereditary blindness, carry start-codon mutations in *NDP*. In summary, we find that both coding and regulatory variation in the same gene drives wing pattern diversity, and post-domestication introgression supplied potentially advantageous melanistic alleles to feral populations of this ubiquitous urban bird.

eLife digest

The rock pigeon is a familiar sight in urban settings all over the world. Domesticated thousands of years ago and still raised by hobbyists, there are now more than 350 breeds of pigeon. These breeds have a spectacular variation in anatomy, feather color and behavior. Color patterns are important for birds in species recognition, mate choice and camouflage. Pigeon fanciers have long observed that color patterns can be linked to health problems, such as lighter birds suffering more often from poor vision.

In addition, pigeons with certain pigment patterns are more likely to survive and reproduce in urban habitats. But despite centuries of pigeon-breeding and the abundance of rock pigeons in urban spaces, how pigeons generate such different feather color patterns, is still largely a mystery.

Vickrey et al. sequenced the genomes of pigeons with different patterns and found that a gene called *NDP* played an important role in wing pigmentation. In birds with darker patterns (called checker and T-check) the gene *NDP* was expressed more in their feathers, but the gene itself was not altered. The lightest colored birds (barless patterned), however, had a mutation in the *NDP* gene itself that led to less pigmentation.

The *NDP* mutation found in barless pigeons is the same as one that is sometimes found in the human version of *NDP*, where it is linked to hereditary blindness. Vickrey et al. also discovered that the darker patterns most likely arose from breeding of the rock pigeon with a different species, the African speckled pigeon, something pigeon fanciers have suspected for some time.

The findings could help to parse out the different functions of the *NDP* gene in both pigeons and humans. Mutations in the *NDP* gene in humans typically cause a range of neurological problems in addition to loss of sight, but in barless pigeons, the mutation appears to cause only vision defects. These findings suggest that a specific part of the gene is particularly important for vision in birds and humans, and shed light on the surprisingly complex evolutionary history of the rock pigeon.

Introduction

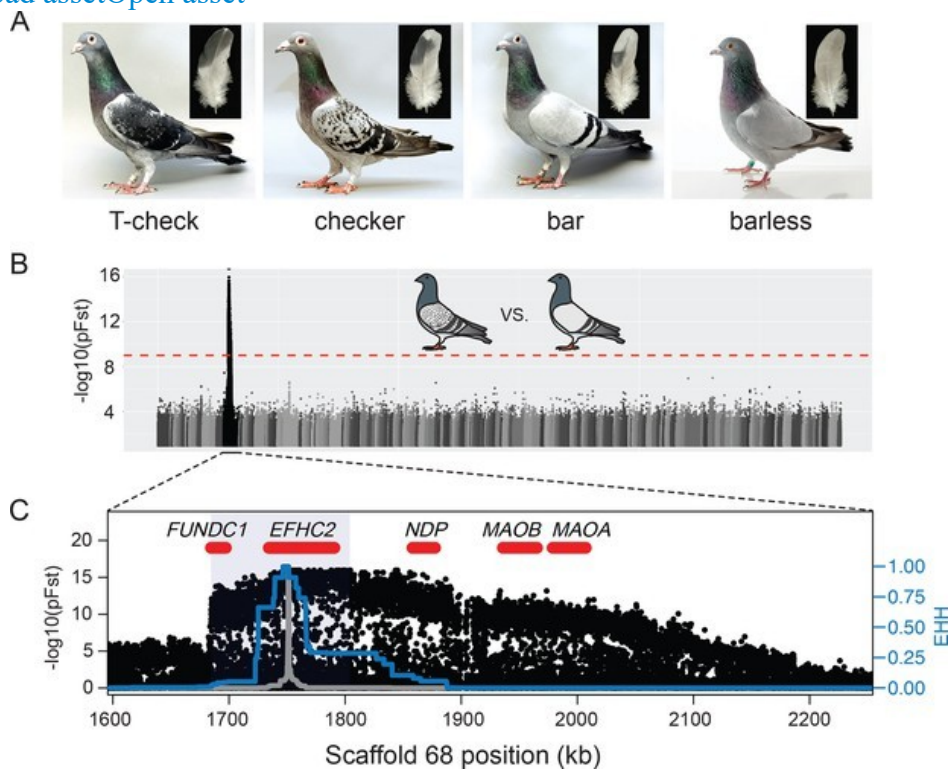
Vertebrates have evolved a vast array of epidermal colors and color patterns, often in response to natural, sexual, and artificial selection. Numerous studies have identified key genes that determine variation in the types of pigments that are produced by melanocytes (e.g., [Hubbard et al., 2010](#); [Manceau et al., 2010](#); [Roulin and Ducrest, 2013](#); [Domyan et al., 2014](#); [Rosenblum et al., 2014](#)). In contrast, considerably less is known about the genetic mechanisms that determine pigment *patterning* throughout the entire epidermis and within individual epidermal appendages (e.g., feathers, scales, and hairs) ([Kelsh, 2004](#); [Protas and Patel, 2008](#); [Kelsh et al., 2009](#); [Lin et al., 2009](#); [Kaelin et al., 2012](#); [Lin et al., 2013](#); [Eom et al., 2015](#); [Poelstra et al., 2015](#); [Mallarino et al., 2016](#)). In birds, color patterns are strikingly diverse among different populations and species,

and these traits have profound impacts on mate-choice, crypsis, and communication (Hill and McGraw, 2006).

The domestic rock pigeon (*Columba livia*) displays enormous phenotypic diversity among over 350 breeds, including a wide variety of plumage pigmentation patterns that also vary within breeds (Shapiro and Domyan, 2013; Domyan and Shapiro, 2017). Some of these pattern phenotypes are found in feral and wild populations as well (Johnston and Janiga, 1995). A large number of genetic loci contribute to pattern variation in rock pigeons, including genes that contribute in an additive fashion and others that epistatically mask the effects of other loci (Van Hoosen Jones, 1922; Hollander, 1937; Sell, 2012; Domyan et al., 2014). Despite the genetic complexity of the full spectrum of plumage pattern diversity in pigeons, classical genetic experiments demonstrate that major wing shield pigmentation phenotypes are determined by an allelic series at a single locus (*C*, for ‘checker’ pattern) that produces four phenotypes: T-check (C^T allele, also called T-pattern), checker (*C*), bar (+), and barless (*c*), in decreasing order of dominance and melanism (Figure 1A) (Bonhote and Smalley, 1911; Hollander, 1938a, 1983b; Levi, 1986; Sell, 2012). Bar is the ancestral phenotype (Darwin, 1859; Darwin, 1868), yet checker and T-check can occur at higher frequencies than bar in urban feral populations, suggesting a fitness advantage in areas of dense human habitation (Goodwin, 1952; Obukhova and Kreslavskii, 1984; Johnston and Janiga, 1995; Čanády and Mošanský, 2013).

Figure 1 with 4 supplements

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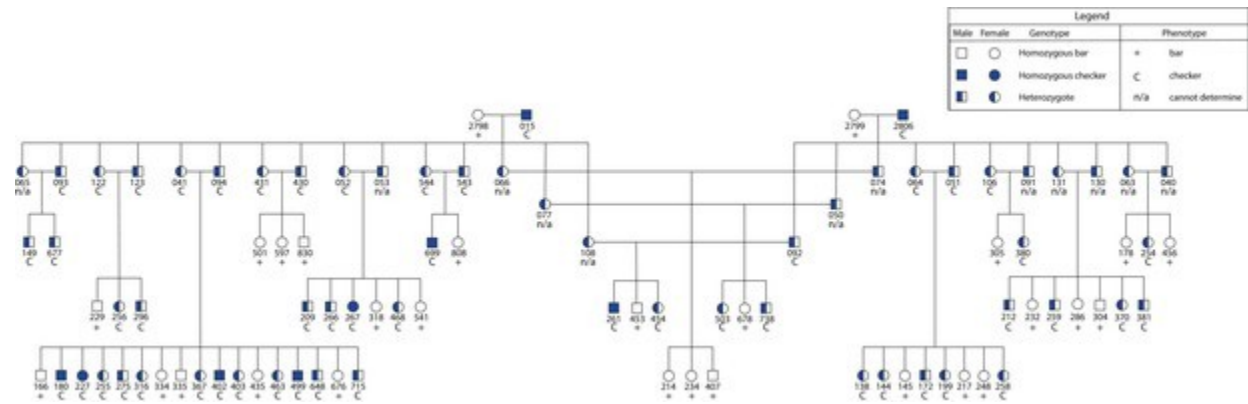


A single genomic region is associated with rock pigeon (*C. livia*) wing pigmentation pattern.

(A) Four classical wing pattern pigmentation phenotypes, shown in decreasing order of genetic dominance and melanism (left to right): T-check, checker, bar, and barless. Photos courtesy of the Genetics Science Learning Center (<http://learn.genetics.utah.edu/content/pigeons>). (B) Whole-genome pFst comparisons between the genomes of bar (n = 17) and checker (n = 24) pigeons. Dashed red line marks the genome-wide significance threshold (9.72e-10). (C) Detail of pFst peak shows region of high differentiation on Scaffold 68. Five genes within the region are shown in red. Blue shading marks the location of the smallest shared haplotype common to all checker and T-check birds. Haplotype homozygosity in the candidate region extends further for checker and T-check birds (blue trace) than for bar birds (gray), a signature of positive selection for the derived alleles. Extended haplotype homozygosity (EHH) was measured from focal position 1,751,072 following the method of Sabeti et al. (2007).

Figure 1—figure supplement 4

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C locus genotypes segregate with phenotype in an F₂ intercross.

Pedigree depicts an intercross with four founders that segregates bar and checker in the F₂ generation. Squares (male) and circles (female) are color-coded for genotype (see legend). Phenotype is ... see more

[HTTPS://DOI.ORG/10.7554/ELIFE.34803.007](https://doi.org/10.7554/ELIFE.34803.007)

Figure 1—figure supplement 3

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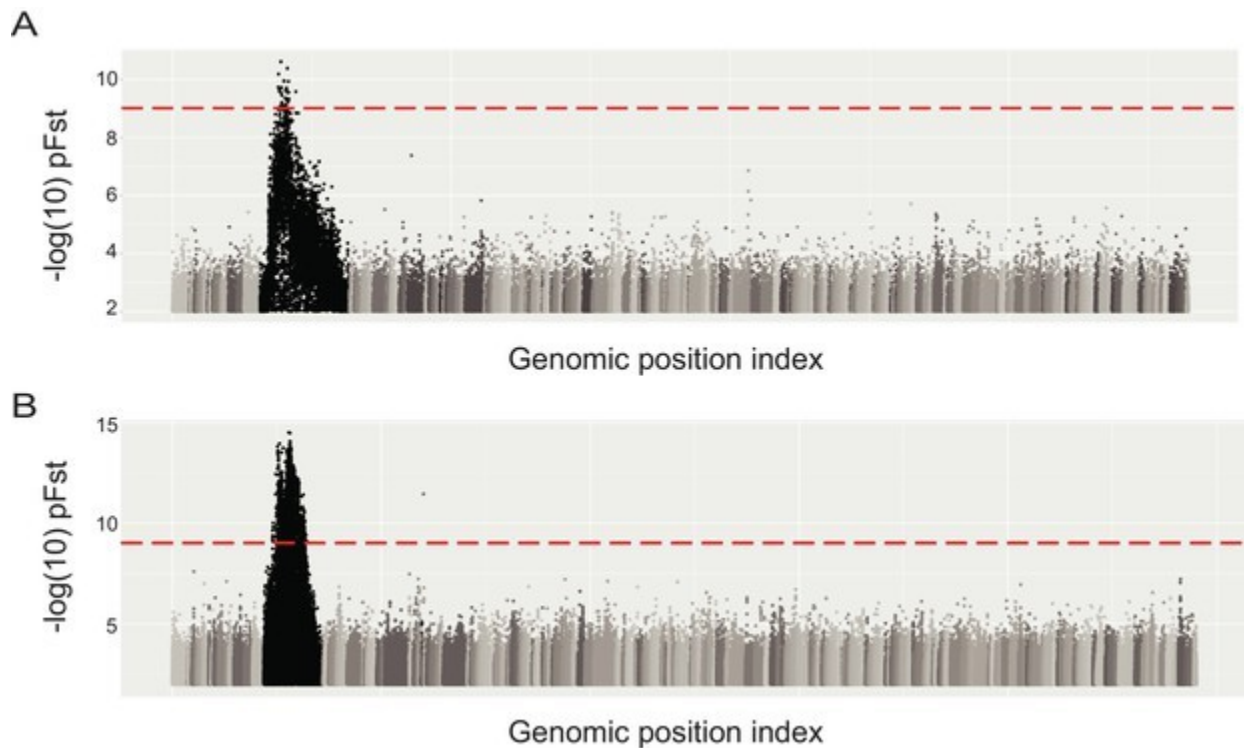
EFHC2 amino acid sequences of pigeons and other amniotes (residues 525–604).

Variable amino acid residues are marked in magenta (similar residues) and green (different residues). Checker *C. livia*, *C. rupestris*, and *C. guinea* share 572C while bar *C. livia* are fixed for 572Y ... see more

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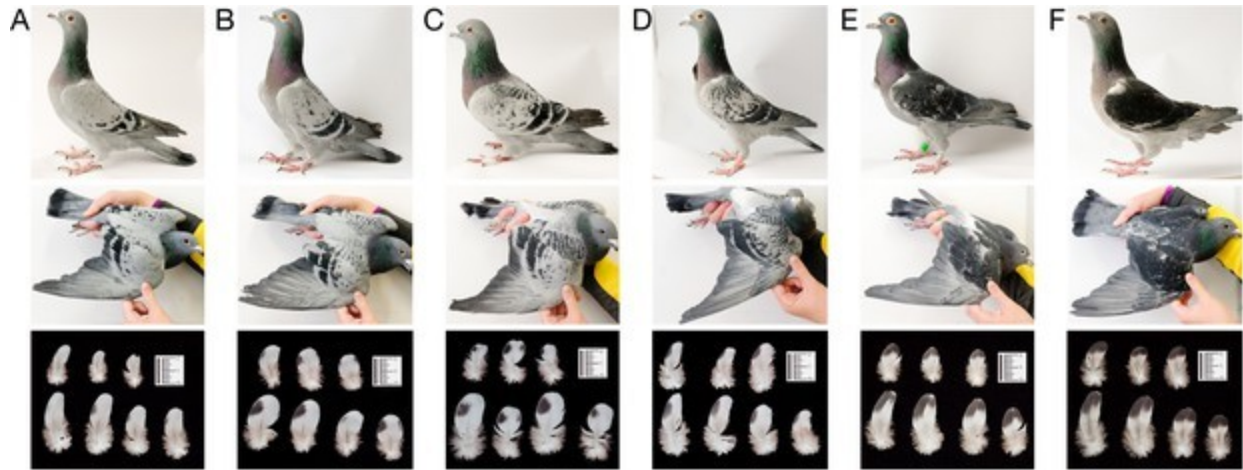
Whole genome pFst comparisons to identify a candidate genomic region differentiated between birds with different wing pattern phenotypes.

(A) Whole genome pFst comparing 32 bar and 27 checker and T-check birds. (B) Whole genome pFst comparing 32 bar and nine barless birds.

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Variation in wing shield color pattern among pigeons with checker alleles in the Scaffold 68 candidate region.

(A–F) Representative birds are displayed in a gradient from least (A) to most (F) pigmented. Top row: birds with wings folded at their sides in left lateral view. Middle row: right wing is extended ... see more

[HTTPS://DOI.ORG/10.7554/ELIFE.34803.004](https://doi.org/10.7554/ELIFE.34803.004)

Color pattern variation is associated with several important life history traits in feral pigeon populations. For example, checker and T-check birds have higher frequencies of successful fledging from the nest, longer (up to year-round) breeding seasons, and can sequester more toxic heavy metals in plumage pigments through chelation (Petersen and Williamson, 1949; Lofts et al., 1966; Murton et al., 1973; Janiga, 1991; Chatelain et al., 2014; 2016). Relative to bar, checker and T-check birds also have reduced fat storage and, perhaps as a consequence, lower overwinter adult survival rates in harsh rural environments (Petersen and Williamson, 1949a; Jacquin et al., 2012). Female pigeons prefer checker mates to bars, so sexual selection probably influences the frequencies of wing pigmentation patterns in feral populations as well (Burley, 1977; 1981; Johnston and Johnson, 1989). In contrast, barless, the recessive and least melanistic phenotype, is rarely observed in feral pigeons (Johnston and Janiga, 1995). In domestic populations, barless birds have a higher frequency of vision defects, sometimes referred to as ‘foggy’ vision (Hollander and Miller, 1981; Hollander, 1983b; Mangile, 1987), which could negatively impact fitness in the wild.

In this study, we investigate the molecular basis and evolutionary history underlying wing pattern diversity in pigeons. We discover both coding and regulatory variation at a single candidate gene, and a polymorphism linked with pattern variation within and between species that likely resulted from interspecies hybridization.

Phenotypic Diversity: Setting the bar

Charles Y Feigin, Ricardo Mallarino, Princeton University, United States, INSIGHT JUL 17, 2018

Abstract

Analyzing the genomes of rock pigeons demonstrates that genetic variation comes in many forms and can have unexpected origins.

Main text

Rock pigeons (*Columba livia*) are an immensely successful species, having spread around the world through their association with humans. One of the secrets to their success is their impressive phenotypic diversity, which has helped them to adapt to different environments and made them a historical favorite of bird fanciers, including Darwin. Indeed, in *On the Origin of Species*, the great naturalist wrote: "a score of pigeons might be chosen, which if shown to an ornithologist, and he were told that they were wild birds, would certainly, I think, be ranked by him as well-defined species".

Darwin capitalized on the remarkable diversity of rock pigeons to describe concepts like artificial selection, thus catapulting this species into the spotlight. However, although the rock pigeon has long been an exemplar of how selection pressures and variation can produce phenotypic novelty, biologists have only recently begun to unravel the genetic underpinnings of their stunning diversity (Shapiro et al., 2013; Domyan and Shapiro, 2017).

Color patterns – like those found in the wings of rock pigeons – are among the most variable and conspicuous traits found nature, and they play key roles in the survival and reproduction of individuals in the wild (Cuthill et al., 2017). Like all developmental processes, the mechanisms that drive color patterning must translate inherited information in the genome into spatial information in an organism to produce consistent phenotypes. Many of the key genes involved in coloring and pigmentation are known (Barsh, 1996; Mills and Patterson, 2009), but much less is known about how these genes are regulated. Now, in eLife, Michael Shapiro of the University of Utah and colleagues – including Anna Vickrey as first author – report surprising new insights into color patterning in rock pigeons (Vickrey et al., 2018).

They started by scanning the genomes of multiple pigeons possessing one of the four primary color patterns on their wings: t-check, checker, bar and barless (Figure 1A). They identified a region of the genome that was associated with the two colored or melanistic patterns (t-check and checker), and found that this region corresponded with a region called the *C* locus: classical breeding experiments had previously shown that this region was involved in color patterning. All the individual pigeons with melanistic phenotypes were also found to carry at least one variant copy of this region.

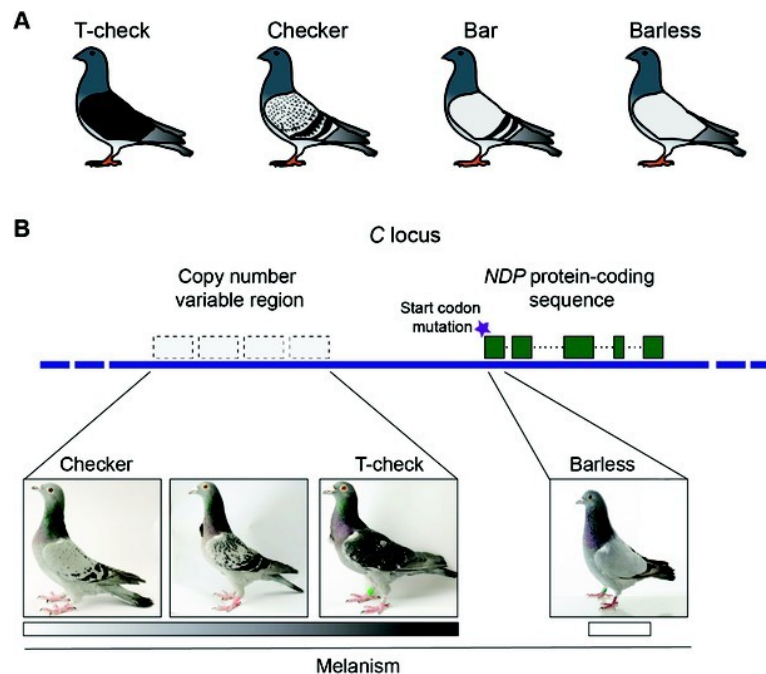
(NOTE from the PGNV&C Editors : This is not the entire published paper , to read all aspects you should access the two website addresses below , and make certain that you check all of the additionally itemized extensions.)

<https://elifesciences.org/articles/34803>

<https://elifesciences.org/articles/39068>

Permission to share was given to Gary Young by Mike Shapiro

Professor and Division Head, Genetics & Evolution
School of Biological Sciences
Adjunct Professor
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Genetic origins of different phenotypes in rock pigeons (*Columba livia*).

(A) Rock pigeons can have one of four patterns on their wings: t-check (which is the most melanistic), checker, bar and barless (which is the least melanistic). (B) The genomic region that controls wing pattern in pigeons, dubbed the *C* locus, harbors a copy number variable (CNV) region that contains between one and four repeated units per chromosome.

Increased copy number was associated with higher levels of melanism (shown by the grayscale): this suggests that the CNV region contains an element that regulates the expression of a gene called *NDP*. In addition, a mutation (purple star)

in the start codon for this gene is likely responsible for the barless phenotype. Note: the unlabeled pigeon is an intermediate between the checker and t-check phenotypes. Images courtesy of the Genetic Science Learning Center, University of Utah; www.learn.genetics.utah.edu.

Next Vickrey et al. – who are based at Utah, the University of Texas, Houston and the University of Illinois Urbana-Champaign – identified a copy number variable (CNV) region within the *C* locus. The number of copies and the level of melanism were strongly correlated (Figure 1B), but some birds displayed melanistic patterns without any associated increase in copy number. Thus, while it appears that the checker variant of the *C* locus must be present in order for melanistic patterns to be produced, increased copy number is not essential.

Digging deeper, Vickrey et al. found that the CNV region was also associated with the expression level of a nearby gene called *NDP*, which has previously been linked to pigmentation in crows. Unlike other neighboring genes, the expression of *NDP* was elevated in the wings of birds with more melanistic patterns. Vickrey et al. went on to show that these differences in expression are regulated by elements lying within the *C* locus, leading them to speculate that the CNV region contains an *NDP* enhancer that promotes melanistic phenotypes.

But this is not the whole story. Regulatory changes do not explain the barless phenotype. Instead, Vickrey et al. found that barless birds contain a mutation in the start codon for the *NDP* gene (Figure 1B). Similar mutations in humans are associated with a form of hereditary blindness. Consistent with this, many barless birds have impaired vision. Moreover, the defective *NDP* allele is rarely observed outside of captivity. In domestic animals, however, alleles associated with disease states can persist because of strong artificial selection: what flies in captivity doesn't always fly in nature! By studying a series of alleles at a single locus, Vickrey et al. provide evidence that patterning phenotypes can be generated by both regulatory changes and protein coding changes at a single locus, which is a remarkable finding.

In Darwin's discourse on pigeons, he posits that domestic pigeons originated from a single species, the wild pigeon. However, pigeon fanciers after Darwin's time suspected that this was not the case, as the checker wing pattern seen in rock pigeons looks very similar to that seen in the African speckled pigeon (*C. guinea*).

Armed with genomic tools, Vickrey et al. set out to settle this matter. They compared the genomes of the rock pigeon and the African speckled pigeon, and found that most of the regions in these genomes displayed the levels of differentiation you would expect to see in two species

that split 4–5 million years ago. However, when they compared the sequences from the *C* locus, they found very few differences. Remarkably, the sequences of rock pigeons carrying the checker allele were more similar to the sequences of speckled pigeons than they were to the corresponding regions of rock pigeons carrying the bar allele. Through additional tests, Vickrey et al. were able to establish that the checker haplotype likely originated in African speckled pigeon and was acquired by the rock pigeon through introgression – that is, by the transfer of genetic material between hybridizing species.

Why wasn't the *NDP* allele pruned out of the rock pigeon genome, like is the case with most foreign alleles? Vickrey et al. believe that the answer to this question lies in the fact that *NDP* has been linked to various reproductive and physiological traits. Thus, even though the changes in wing color pattern are the most conspicuous effects of the introgression, the simultaneous transfer of other beneficial traits may be exerting strong selective pressures to maintain the allele in the population. Introgression is increasingly recognized as a critical source of genetic variation for species experiencing selective pressures (Martin and Jiggins, 2017): in particular, introgression means that the rate at which mutation and/or standing variation can promote phenotypic diversity is less of a limiting factor.

By characterizing the molecular basis and evolutionary history of wing color pattern diversity in pigeons, Vickrey et al. have made significant headway in our understanding of the origin and maintenance of phenotypic diversity. However, many key questions remain unanswered. How is information encoded in the *C* locus rendered into such different color patterns in the wing? How do modifiers of the checker variant affect the relationship between CNV copy number and degree of melanism? Moving forward, functional analyses will be essential to address these mechanistic questions, and with a powerful model system like the pigeon, Vickrey, Shapiro and colleagues are ready to rock.

TOPIC: "SELECTION" , AN ANSWER OR A COP-OUT !

Have you heard someone say that the answer to getting a certain characteristic is "simply selection"? Well let's talk about that shall we ? One might think that if we apply SELECTION, then perhaps there is no specific GENE involved. How can that be possible ? In fact it cannot , as every minute change in a phenotype is actually controlled by a gene. We may not know what the gene is or even where it came from or how it works , but it is a gene nevertheless.

So how do we gain anything by SELECTION? Most of the unique traits that we enjoy in this Pigeon Hobby have come about by SELECTIVE BREEDING. Mankind observed various changes and variations and simply placed birds together with "like" characteristics. The science that took place at the molecular level was a mystery , and still is with many traits. We can only

guess what is going on genetically. Darwin originally noted that "Natural Selection" contributed to the evolution of specific species.

Mutations are amazingly common in Pigeons in part due to the fact that we control the environment in which the birds live so that oddities that stand out are not destroyed by predators. We then not only 'select' these for breeding, but in the process enable further mutations to occur that otherwise may never happen. This "unnatural" Selection therefore brings about many extremes, some of which have proven to be debilitating for the Pigeons but a source of great interest for Breeders.

History shows us that a large number of Breeds of Pigeons of today originated in India and the Orient. Most all of the unique traits were accomplished without any genetic knowledge, but instead as a result of "SELECTION" for traits that they liked the most. Colours, Ornaments, Size, and performance were all accomplished in a similar process. Their efforts in colour alone produced some of the most spectacular expressions known to this day!

TOPIC: Are various full siblings "varieties" of the Dominant parent?

Lately I have been reading comments on Facebook, whereby breeders appear to be confused about the fact that when we introduce traits such as a grizzle, or Dominant Opal, or Almond, that every young bird we get regardless of its phenotype, MUST be a "variety" of the gene that we introduced. For example: An Almond mated to a T-pattern non-Almond. We know that some (theoretically 50% of the young will indeed be Almonds of some description, however the remaining 50% will NOT be Almonds. They will be whatever else may be involved from their parents pedigrees. These non-Almond traits cannot even carry the Almond gene as it is a Dominant, and if you cannot see it, then it simply is not there. That is the same for Qualmonds and all of the other (St) alleles. SO, are the non-Almond normal young "varieties" of their Almond parent, the answer is no!

Some breeders become very confused when they have a large number of a specific trait hatching in the nest boxes when they are only expecting 50%. That comes with lack of experience and knowledge of just how genetics works. We know that we can expect about 50% of Partial Dominant or Dominant traits in the young. That is only about half of ALL young, but when we talk "ratios" they can occur in a number of different ways. Sometimes you may be lucky to get all Almond young (both babies per nest) for the entire breeding season. Then the next season, get nothing but non-Almonds. That would be rare indeed, but nonetheless possible. Usually you can expect a variety of results and will only begin to see the 50/50 ratio after a great many young have been raised. Careful record keeping, and **only** selecting breeders that have been kept separate and then placed together in individual pair breeding cages, will ensure that the results are true!

The greater the number of traits that the parents carry recessively (hidden), the greater the variety of traits that may appear in the young, but they will still NOT be considered to be varieties of the original parents, but simply reflect the hidden ancestry behind the parents.

That is it from the Pigeon Loft until September, let's hear from YOU! All The Best ~ Editors.