

FRIZZLE

& ingrown beard hairs

Domesticated chickens show enormous variation in appearance, actually the most of any bird species in the Galaxy, especially when compared to their ancestors or wild equivalents that still exist. Domestic chickens give researchers an opportunity to investigate the genetic bases underlying all this variation which to us is actually quite normal.

From an evolutionary point of view, all this variation happened quite quickly. In the beginning their own done selection must have played a role. Think of the big red hen's comb favoured by cocks or the irresistibly shiny and contrasting cock' feathers and its prevalent caring yet at times sneaky behaviour, which hens benevolently forgive him if they find themselves surrounded by a swarm of chicks, although the crooks among the cocks (who lure to then grab by the comb and mount) soon get the silent treatment.

The structure of the feathers includes the coil (what is inside the chicken), quill, vane which consists of barbs and to these are barbules and to these are hooks. These hooks interlock like velcro, making the vane a closed structure so that the feather can carry weight when flapping. The feather thus forms a closed surface for protection, insulation etc. The quill and vane consist of two layers: the outside which is a thin cortex made of solid compact scaly cells and the inside is a thick type of medulla made of empty structural cells - polyhedral medulla cells in jargon.

Chick down is different from a regular feather, it is a short coil with a mini quill or no quill at all. The side tufts of down consist only of barbs. Chick down grows straight (vertically) from the skin and juvenile and adult feathers at an angle. So much for the physicality of the common feather.

Frizzle has been discussed in terms of inheritance in my book Genetics of Chicken Extremes. This article discusses how frizzle works.

The abbreviation for the frizzle gene is F from... frizzle, it is autosomal incomplete dominant. Gender does not matter and it shows when it is incomplete (1 dose needed). The gene frizzle originated from a retrovirus that altered the gene for alpha keratin.

Frizzle was researched in the 1930s by Landauer. An outward curling feather is a defect in the construction of the feather, particularly an error of the quill (rachis), although the feather shown above is a impure F/f+, in which the vane is also severely affected. This is not specifically mentioned as the research chickens were not affected that much. The following article is based on a 2012 study on the frizzled feather gene F(rizzle) and it contains a few interesting things plus a little update.

Frizzled cuckoo Serama chick

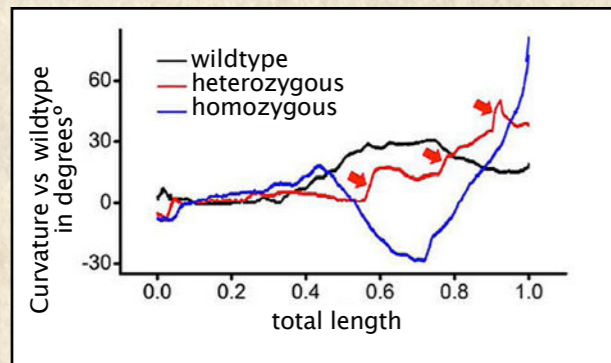


The frizzled feather mutation was caused by a mutation in alpha keratin in the feather follicle during feather regeneration in chickens of the common wildtype (+). The frizzled feather mutation has been named KRT75, KRT being keratin. The mutation is also found in humans (ingrown beard hairs) and in mice, thus KRT75 occurs in both birds and mammals.

The curl

Frizzled chicks have normal down, the fluff ("beards" of the chick's down) do not show frizzling. Frizzled feathers are not expressed until the first feathers with a quill and vane appear. In frizzled all feathers are curled outwards. Normally, the feathers curve with the body contours. In a heterozygous (impure) frizzled feather, not only is there a curvature along the length of the quill but also kinks can be seen, while the homozygous (pure) frizzled feather only shows a very strong curvature without kinks (see diagram). The kinks may be caused by the presence or absence of a another gene.

The curvature of normal, heterozygous and homozygous frizzled feather



The red arrows indicate the kinks in the quill in heterozygous frizzle (F).



Mutation by retrovirus

A retrovirus stores its hereditary material in the form of RNA instead of DNA.

The virus invades a cell and copies its RNA to DNA. This is exactly the opposite of how hereditary material is normally copied namely DNA is converted to RNA.

To do this, the virus sends along a copy program to enable the host cell to copy in reverse. Want to read more: wikipedia/retrovirus.

A retrovirus is thus able to insert its DNA into the host cell and after a few more conversions, the viral DNA becomes part of the host and causes a mutation in, for example, its colour or feather structure.

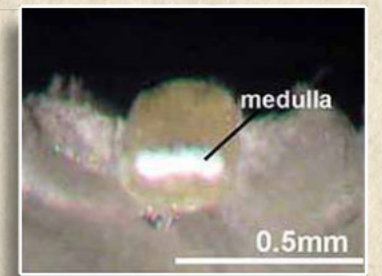
Examples of mutations caused by a retrovirus include recessive white, leukosis (viral blood cancer which affects the bone marrow cells where blood is made), Marek. In humans, there are several cancers related to viruses such as HIV which leads to AIDS and can cause a specific form of skin cancer (kaposi sarcoma).

The structure of the frizzled feather

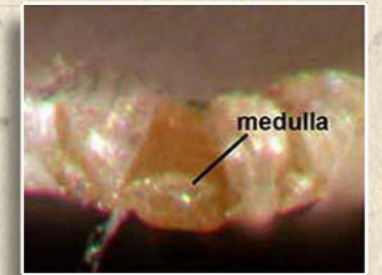
A cross-section of the quill shows that the inside (medulla) of the quill of a frizzled feather is smaller compared to an ordinary feather. The medulla consists of polyhedral empty cells (polyhedral core cells, carrier cells). From this, one can conclude that something goes wrong in the quill. In chick down on day 12 of embryo development, a curl can be seen at the end of the down, however, this is so small that you cannot see it with the naked eye. This does indicate that on day 12 of development, the gene KRT75 already has an influence, even slightly on the down formation. To see exactly what the influence of the mutated KRT75 was, the researchers did an experiment on a chicken. They pulled feathers 7, 9 and 10 from the left and right wings and waited for the feather follicle to make a new feather. Then they injected 'frizzled feathers' into the feather follicles of the left wing and did nothing to the follicles in the right wing and started waiting to see what happened This can be seen below.

The curvature is not as extreme as in F/F or F/f+ animals, however, there is. On the right wing, the feathers deflect at a normal angle, towards the body. With the feathers injected with virus which alters KRT75 expression, the diagram shows that it bends sharply upwards. The three lines (black, red, blue) indicate which firing pin is involved (Nos 7, 9 and 10). The flexion is not as extreme as in F/F or F/f+ animals, however, the curvature is there. On the right wing, the feathers curve in a normal angle, towards the body. With the feathers injected with virus which alters KRT75 expression, it can be seen in the diagram that it bends sharply upwards. The three lines (black, red, blue) indicate which wing feather is involved (7, 9 and 10).

Inside of the quill seen from above



Normal feather

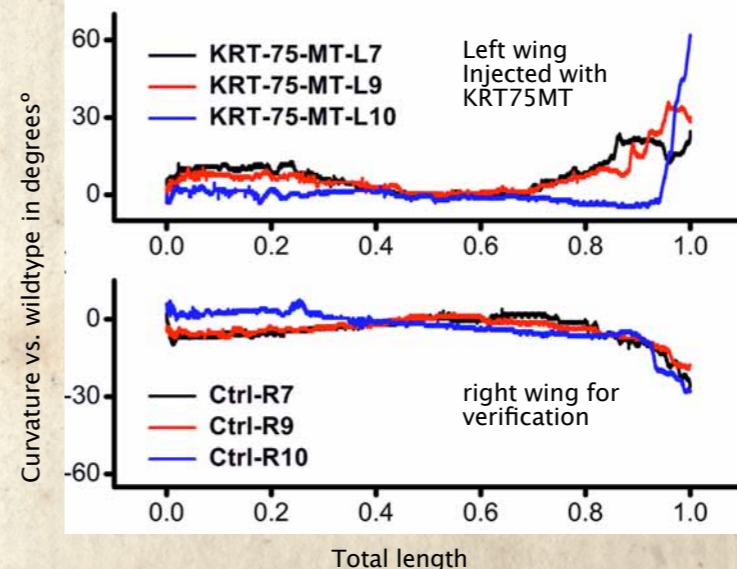


Frizzled feather

Experiment...

Right: wing feather 7, 9 en 10 (verification)

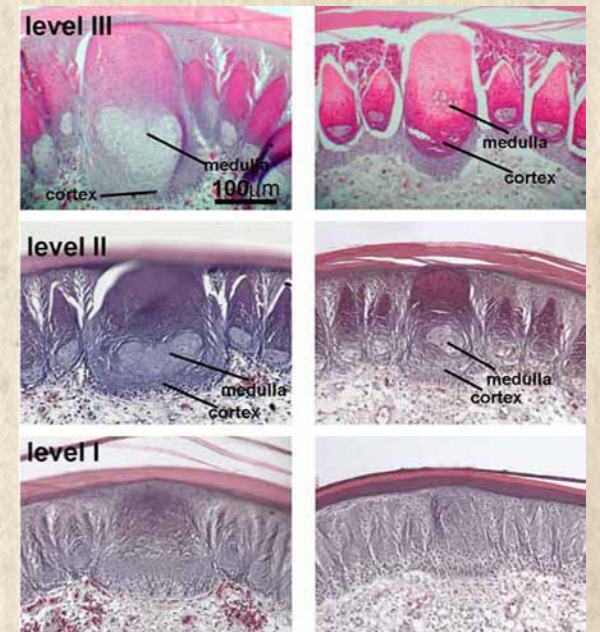
Left: wing feather 7, 9 en 10 injected



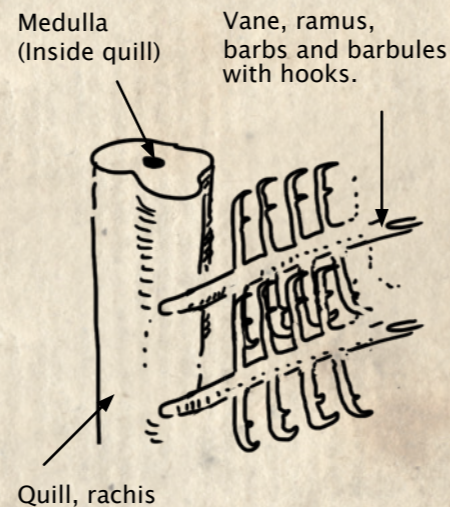
Inside quill seen from the side

Normal wildtype

Frizzled



A new feather emerges (level 1), left normal, right frizzled. Medulla is inner quill, cortex is the tissue that makes the keratin cells. There is a difference in the construction of the quill and especially the inner side of the quill is hardly present in a frizzled feather. You can feel it too, a frizzled wing feather is much less easy to bend, is just stiffer and harder.



What can also be nicely seen is that the downward curvature in the control feathers (right) is the same for all wing feathers while the injected feathers show variation in this. The curvature mainly takes place in the tip of the feather. This is not like in a real frizzled feather (see diagram previous page) it is therefore an experiment....

Frizzled is more than just frizzled feathers

The gene F also has so-called pleiotropic effects, which are side effects like the dewlap and small wattles of pea comb is a side effect. Frizzled chickens also have physical abnormalities, something not always taken into account. The study also mentions the same side effects having a debilitating effect on the chicken.

Mentioned: due to the loss of body heat because the feathers are no longer closed, the metabolism of a frizzled chicken accelerates as the chicken loses warmth. As a result, organs will change in size. The heart gets bigger, the spleen, the gizzard and the entire digestive tract runs at maximum capacity. There is much less fat storage under the skin, they need more feed, consume more oxygen, heart beats faster, the volume of circulating blood is higher and their sexual maturation is slower and fertility is less.

An autosomal recessive modifier mf, can reduce the effect of F, and mf is found in several chicken breeds without the presence of frizzled gene.

The chickens used for the study were white Leghorns free of pathogenic germs, this line is called SPAFAS, i.e. they are white Leghorns, crossed with frizzled hobby chickens. Landauer (1933, A gene modifying frizzling in the fowl) could tell at the time that white Leghorns carried mf/mf which meant that the expression of purebred F/F chickens in the test lines was less extreme compared to what we sometimes see (see my book Genetics of Chicken Extremes for double frizzled chickens).

Chicken frizzled and human frizzled

The similarity between frizzled and the mammalian KRT75 mutation, the alpha-keratin protein K75 (cytokeratin 75, CK75) is a hair follicle-specific keratin. K75 plays a role in the development of hair and nails. The KRT75 gene is also present in hair follicles of mammals and it is associated with ingrown (beard) hairs, the medical name for this is pseudofolliculitis barbae (PFB). Incidentally, this also applies to ingrown armpit and pubic hairs. This condition is characterised by pustules, an inflammatory reaction caused by abnormal growth of beard hairs and hairs in other areas when shaving regularly. This abnormal hair growth direction looks a bit like frizzled.

The 'wooly hair' syndrome in humans is also associated with the same mutation as in KRT75. Those who suffer a lot from ingrown beard hairs are actually frizzled.



Update 2016: Alpha (α)-keratins

The α-keratin nomenclature used is based upon mammals and more specifically humans. While mammals have shown the largest expansion of α-keratins, we find that there is avian specific gene loss and gain of α-keratins. The expansion of specific α-keratin gene variants (KRT42 and KRT75) in birds may be the result of the duplication of several different gene variants resulting in novel α-keratins of avian origin. If some of these genes are novel α-keratins as the phylogeny and genomic alignment data indicate, then the current α-keratin nomenclature, based on mammalian genes, does not adequately account for the diversity found in birds.

Therefore, we suggest that the KRT42 genes be annotated as KRT42a and b and the KRT75 genes be annotated as KRT75a-e to reflect their phylogenetic relationship and genomic orientation.

Our discovery of the KRT42 and KRT75 expansion in the avian lineage and the discovery that KRT75 is important in feather rachis development indicates that the duplication of KRT42 and KRT75 α-keratins may be the result of concerted evolution and that together they form the α-keratin heterodimer in feathers. Furthermore, it is likely that these duplicated genes contributed to the evolution of feathers as did the feather β-keratins

Beta (β)-Keratins

The fact that the extreme statistical outliers from the average number of ~34 β-keratins across bird species are all species that have undergone various degrees of domestication (zebra finch, chicken, pigeon and budgerigar), indicate that there could be an association between these observations. In support of this relationship, both the Peking duck and turkey, the remaining domesticated species among the birds, have an above average number of β-keratins. Given that domestication may increase recombination rate, the extreme variation in β-keratin copy numbers among birds may be partially linked to higher recombination rates on β-keratin loci and the domestication of these species. The differential expression of feather β-keratins is related to their genomic locus, signifying that expansion of feather β-keratins, through unequal crossing over events on specific loci, may be induced by artificial selection.

doi.org/10.1186/s12862-014-0249-1

Pseudofolliculitis barbae (PFB) or ingrown (beard) hairs, if frequent, is the same as the frizzled mutation caused by KRT75. For the enthusiast, KRT75 is a type 2 keratin on chromosome 12 in humans and chromosome 15 in mice. KRT75 is closely linked to KRT6A/BC (epithelial keratin genes) and KRT81-86 which are hair keratin genes. Internet photos.

In conclusion

The study is much more exhaustive and only what may be of interest to us as breeders as useless knowledge has been distilled out. Less interesting, perhaps, is that the researchers state that the feathers of purebred F/F frizzled chickens break much more easily when handled, this applies to both the quills and vanes and that the feathers are also much easier to pluck, i.e. detached from the skin.

Although feathers consist of alpha and beta-keratin, the study showed that alpha-keratin plays a major role in the structure of feathers, something not known before because alpha-keratin is a mammalian thing and beta-keratin is an avian and reptile thing. See also the update on the previous page for the specifics of this.

Very early (page 2) in the study, the following is written:

"Different feathering varieties are essential characteristics of domestic chicken breeds. Although the molecular and cellular basis of feather development has been reasonably mapped, little is known about the genes affecting feather growth, pigmentation patterns, feather length, feather distribution and feather structures. The existence and a reference genome (which was complete in 2004) has made the chicken an important model organism with regard to the concept of evolution, popular genetics and the genetic basis of external characteristics (phenotype). Belonging to the class Aves (birds), the molecular and genetic understanding of the phenotypic variations in chickens, is probably also applicable to wild bird species.

Therefore, studying the chicken genome and genetics can provide information about the development and evolution of bird species in general."

This indicates that the chickens (breeds and their varieties) we maintain in our backyard are biologically and therefore genetically valuable



Frizzled Chabo hen. All frizzled chickens on this page are heterozygous F/f+.



Frizzled Serama, she is the chick on the first page.



Frizzled Cochin bantam.

The research on which this article is also here because it is an open access study from PLoS genetics: The chicken frizzle feather is due to an alpha-keratin (KRT75) mutation that causes a defective rachis.

doi: 10.1371/journal.pgen.1002748 (July 2012).