Colour Genetics

*Australian alpaca breeder **Elizabeth Paul** has been trying to unravel the mysteries of colour inheritance in alpacas for some years and her book 'The Alpaca Colour Key' which she publishes herself in now available in the UK. In this issue we publish an introduction to the book and to colour genetics to be followed in our Summer 2005 magazine with an article on blue eyed whites.*

BASIC GENETICS:

Genetics is the science of studying and predicting patterns of **inheritance**. Any discussion on colour inheritance requires a short list of essential definitions for a basic genetic vocabulary, to give an understanding of the genetic mechanisms involved.

The **cell** is the basic unit of life. Large complex animals such as alpacas and humans are made up of millions of cells. The cell contains a **nucleus,** which is composed of **DNA** or deoxyribonucleic acid. DNA is the biological **code,** which contains all the information required to create the animal. The code is set in the form of individual pieces of information, called **genes**. Genes are organised into long threads of DNA, called **chromosomes**, which are found in pairs within the nucleus. Genes also occur in pairs, and the members of each pair are called **alleles**. Each gene pair has a specific position on a given chromosome. Each **species** of animal has a particular **number** of chromosome pairs that sets them apart from other species. Humans have 23 pairs of chromosomes, and alpacas have 37 pairs.

Reproduction in higher mammals involves **duplication** and then **halving** of the chromosome pairs, so that each sperm or egg contains only half the original number of chromosomes. At **fertilization**, the original number of chromosomes is restored, with half of the genetic material coming from the mother, and half from the father. The new baby has the same number of chromosome pairs as its parents, but it is not a clone of either, as the recombination of any two halves is an entirely **random** event. Each mating event is therefore a completely **separate** event from the one before it, and the one that will come after it. This is the basis of variation within a species.

The **genotype** is the full complement of gene pairs for the alpaca. This is **fixed** from the moment of fertilization of an egg by a sperm, and cannot be altered (except by gene technology). The **phenotype** is the physical expression of those genes, which is what we can see or measure on the alpaca.

The expression of those genes can be altered by any number of **environmental** effects, such as time, temperature, day length, chemicals, different foods and so on. Alleles in the genotype are said to be **dominant**, when they are **always** expressed in the phenotype. Dominant alleles require only **one** copy of the gene in the genotype to be expressed. If two dominant alleles are present, one is usually inactive. Alleles that require **two** copies of the gene in the genotype, to be expressed in the phenotype, are called **recessive**. When the alleles in the genotype are the **same** as each other, they are called **homozygous**. When they are **different**, they are called **heterozygous**.

Usually when we talk about the "genotype" of an animal, we are really only referring to a few genes whose expression we are interested in, eg fleece type or colour.

GENE SERIES

There appear to be three main series of genes, which interact in various combinations to control pattern and colour in mammals. The first is called Extension, which controls the other two. The second is Agouti and the third is the black/brown locus. Dr Phillip Sponenberg of the USA considered that the Agouti locus was the most important locus for determining coat colour; and proposed a series of phenotypes to fit with observed patterns and colours in alpacas.

There is a complex interdependence between the first two series, in that Agouti alleles require the presence of Extension wild type allele for their full expression.

The various alleles at the Extension locus either extend or reduce the amount of eumelanin in the coat. These alleles give a uniform coat colour, with no shading or variation. They are both antagonistic and epistatic to Agouti series alleles. There are three main alleles. The Extension series is designated by the letter E with superscripts of D, dominant black; +, wild type; and ee, recessive red.

Agouti is the reverse of Extension, in that it runs from light to dark, but it requires the presence of the Extension wild type allele for expression of the Agouti alleles. The Agouti series is designated by the letter A. The top dominant for the Agouti series is the lightest allele for that species. It may be called "dominant white".

The black/brown locus – determines whether the eumelanin present is black or its recessive brown. As these are two alleles of the one gene, the animal cannot have both black and brown together in the one coat. The Black/ brown locus is designated by the letter B, with superscripts of B (dominant black); and bb, recessive brown.

Agouti itself is a protective colouration more often seen in smaller animals, such as cats and rodents, where both red and black pigment may be found in the one hair. Guard hair is often uniformly dark. Agouti also produces the effect of the ventral part of the body being lighter than the dorsal part in many species.

In the larger grazing animals, this translates to symmetrical, fairly well defined patterns of red and black areas in the coat. Patterns with more red dominate over patterns with more black. Domestic horses and cattle with these patterns are called "bays"; they have generally redbrown bodies and black points, that is, any combination of the ears/face/mane/tail or lower legs. The shade of red, particularly in horses, can vary from light red yellow to very darkest mahogany, where the black points are sometimes hidden.
Dr Sponenberg considered that the red colour on nearly all fawn, red, tan and "brown" alpacas was phaeomelanic red, and that most alpacas described as "browns" would have black fibre on them, and would therefore be bays. The important point here is that the "brown" colour, on an animal that appears to be both black and brown, is phaeomelanic red, and not eumelanic brown.

An alpaca could be homozygous or heterozygous for each of the three gene pairs. It could be homozygous for all three genes, but it would be more likely to be heterozygous for at least one of them.

COLOUR CHARTS

Alpaca fleece colours have been described from the colour of the fleece on the blanket. These colours were originally decided by the mills that used the fleece. Patterns of colour have been largely ignored, or at least not recognised, either by alpaca breed societies or by breeders themselves. The colour charts are meaningless from the point of view of genetics. This has led to the confusion with alpaca colour genetics.

COLOUR CODE

Colour is one of the most important advantages that alpacas have over other fleece bearing animals. When I first bought into alpacas, nearly seven years ago, I fell in love with a beautiful grey female and was determined to try and breed more greys. I found no-one was able to help me do this, as the patterns of colour inheritance in alpacas seemed to be one big mystery. As a biologist with some knowledge of genetics, I felt I had the necessary background to investigate this further. What began as a simple personal goal has evolved into a seminar, titled "The Alpaca Colour Key" which I have presented regularly to Australian alpaca breeders since November 2002. I have combined models of inheritance, together with pigment studies and pedigree data, to try to form a more complete view of colour inheritance in alpacas.

PIGMENTATION

Coat colour in mammals is almost entirely dependent on the presence or absence of **melanin** pigment granules in the hair and skin. There are two types of melanin, **eumelanin** (black/brown) and **phaeomelanin**, which is yellow. Melanin is concentrated in the skin **epidermis**, at the base of **hair follicles**, and in the **retina** of the eye. Colour in these areas is determined by the size and shape, as well as the type, number and distribution of granules. Melanin is based on the amino acid **tyrosine**. Mammalian pigment granules consist of melanin, attached to a protein.

The most important role of pigment is **protection** of the animal from **UV** light, but different patterns of pigment across the animal also provides disguise protection from predators or prey, warning patterns, or sexual maturity status. Pigment also plays a role in other systems including hearing.

USES OF COLOUR

Unlike birds or even reptiles, mammals are rather restricted in their use and expression of colour. Most mammals come in drab browns and greys, with the occasional black and white combination. Many young animals have different colour patterns to the adults of their species. Very often they are spotted, striped or both. Sometimes this is for protection or concealment from predators, but it may also communicate the juvenile state to adults, causing them to modify their social responses. Adult male animals may have more intense colouration or bolder patterns than females and juveniles. This is partly for protection of the females and young, but it also advertises the male as fully mature, desirable to females and a threat to other males. Often such patterns may still be dull to our eyes, but the animals are programmed to notice subtle differences.

Colour in different patterns can also form a warning system within a group. Rabbits and many deer have a bright white under tail, which they flick to warn other members of their group of danger. Black and white skunks need no introduction; they fluff out their stripes and tail to make themselves look bigger and less of an easy target to predators. Generally, the larger the animal, the less need it has for protective colour patterns, although giraffes are perhaps the standout exception (in more ways than one!)

VARIATIONS

In the wild, the pattern and colour of a particular species may be very constant, or it may be quite variable. Sometimes the variability has little consequence to the animal's survival; and sometimes it may be lethal. At a glance, all zebras look the same, but each individual zebra has a pattern of stripes as unique to itself as a fingerprint. Foals and their mothers know each other's stripe patterns, the same as a human child knows its own mother's face in a crowd. A zebra born without stripes, if ever there was one, would very likely be abandoned by its mother, since it has no stripes to initiate her recognition response. The striping on zebras, while it does not conceal them on the open plains, confuses an approaching predator with a dazzle of shifting lines. An all white or all black zebra would easily stand out in such a situation.

All white predators are also at a disadvantage, unless they happen to be polar bears in the Arctic wilderness. White tigers are relatively common in zoos and theme parks, but they are almost non-existent in the wild. They all trace back to one white male, which was taken from the wild in the 1950's and bred to yellow females. Eventually some of his mates produced several white cubs. There is also at least one group of very pale or white lions. As lions are social cats, they have been able to survive as a group, where one on its own might not.

Animals that vary too much from the norm may also be unable to attract a mate because of the sexual requirements of that species. Leopards however, have a melanistic form, which is no bar to either hunting or to mating (probably because leopards are most active at night). Black leopards are at no particular survival disadvantage, compared to spotted ones.

Whatever the wild situation, in a domestic environment such as a farm, animals with unusual patterns or colours are more likely to be saved and nurtured, than discarded. Foxes and mink produce a range of colours when farmed, which are never seen in the wild. The usual colour for wolves is greyish, but domestic dogs come in many colours. Here also, colour is no bar to mating.

BASIC MELANIN PRODUCTION

Melanin is manufactured by special cells in the body called **melanocytes**, which arise from the neural crest area of the embryo. Cells derived from the dorsal or top part of the embryo, are the ones that will ultimately develop into melanocytes. These migrate to their destinations through the epidermis during the embryonic stages of development. Any delay in this migration may affect the final colour pattern. Pigmentation is generally more intense around the head, along the back of the neck and the top of the back, than the belly region of an animal. Mature melanocytes insert pigment granules into the base of the hair shaft as it grows out of the follicle.

CHANGES IN PIGMENTATION

Melanin production involves a number of other biochemical agents, and any alteration to the sequence, or to the components will have an effect on the final product, and therefore the colour. For example, albinism is the result of the animal's genetic inability to form tyrosinase, the enzyme required to convert tyrosine in the first step of pigment production. This is a homozygous recessive condition. The albino animal will never develop pigment, but can be shown to have melanocytes present in the skin. Other genes cause a switch between the production of eumelanin and phaeomelanin; still others cause a grouping or clumping effect of the granules themselves to produce a diluted final effect.

The level of activity of the melanocytes can also be altered, as in the change from dark summer coat to light winter coat of the snowshoe hare and Arctic fox. This change is initiated by the changing day length between summer and winter. Cold temperature can also have an effect on pigmentation. The Himalayan rabbit has a white coat, but the extremities are black, due to the lower temperature in those areas. If a Himalayan rabbits' fur is shaved off and an icepack applied to the shaved area, the hair that grows back will be black. Ageing causes a permanent reduction in melanocyte activity in the hair follicles.

Drawn from "The Alpaca Colour Key" by Elizabeth Paul November 2002.