MELANOCORTIN 1 RECEPTOR: POLYMORPHISMS ANALYSED IN ALPACA (*Lama pacos*)

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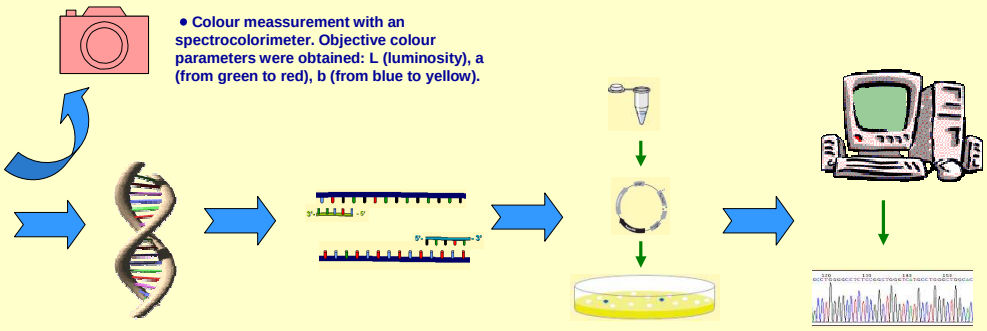
INTRODUCTION

- Coat colour is a trait of important value for alpaca fibre quality and it is determined by the pigment melanine.
- MC1R is a transmembrane receptor in melanocytes responsible for colour pigment synthesis and its gene is encoded by Extension locus.
- The knowledge of colour pattern synthesis could contribute to develop a more efficient conservation breeding programme of alpacas.

OBJECTIVE

- Characterization of polymorphisms associated to phenotype present in MC1R gene in alpacas.

METHODS



- Colour meassurement with an spectrocolorimeter. Objective colour parameters were obtained: L (luminosity), a (from green to red), b (from blue to yellow).
- DNA extraction from 16 alpaca fibre samples: 4 Black, 4 Brown, 4 Cream and 4 White.
- PCR. MC1R gene of alpaca was amplified in two overlapping fragments. Two pair of primers were used.
- Cloning. MC1R gene was cloned in a pDrive Cloning Vector.
- Sequencing. MC1R complete gene sequences were obtained.

RESULTS

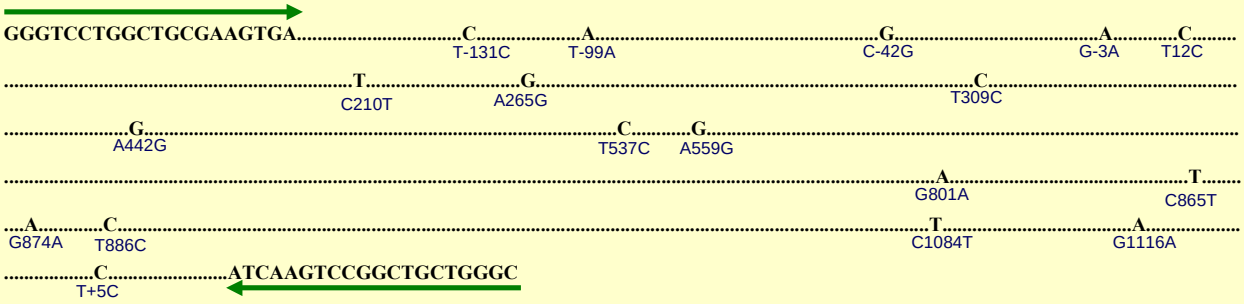


Figure 1. The figure shows the 18 polymorphisms found in MC1R gene in alpaca (Accesion number: EU220010) and the nucleotide change. Green arrows indicate forward and reverse primers.

Nucleotide	Mutation	aa Change	Effect in protein	Fibre colour
265	A/G	T/A	Polar to Non polar	4 colours
309	T/C	Silent	No	4 colours
442	A/G	V/M	Non polar	4 colours
537	T/C	Silent	No	4 colours
559	A/G	G/S	Non polar to Polar	4 colours
801	G/A	Silent	No	4 colours
1084	C/T	R/C	Polar to Non polar	4 colours
1116	G/A	Silent	No	4 colours

Table 1. The table shows the 8 polymorphisms found in MC1R gene in alpacas that had been previously described by other authors. It is shown the effect produced to the protein by the aminoacid change caused by each mutation. All polymorphisms were found in fibre samples of the four colours analysed.

Nucleotide	Mutation	aa Change	Effect in protein	Fibre colour
-131	T/C	5´UTR	No	4 colours
-99	T/A	5´UTR	No	Cream, White
-42	C/G	5´UTR	No	4 colours
-3	G/A	5´UTR	No	**Cream**
12	T/C	Silent	No	**Cream**
210	C/T	Silent	No	Cream, White, Brown
865	C/T	G/C	Polar to Non polar	**Brown**
874	G/A	V/I	Non polar	**Brown**
886	T/C	F/L	Non polar	**Brown**
5+	T/C	3´UTR	No	4 colours

Table 2. The table shows the 10 new polymorphisms found in MC1R gene in alpaca and the effect they produce to the protein by the aminoacid change caused by each mutation. The last column represents the colour of the hair sample in which the SNPs were found.

CONCLUSIONS

- 18 Single Nucleotide Polymorphisms were found in the MC1R gene in alpaca.
- 10 SNPs were new mutations while 8 SNPs had been previously described by other authors.
- Some mutations were only present in one coloured fibre samples, but futher investigation will be needed to prove if there is a relationship between specific polymorphisms and fibre colour.