

## Incontinentia Pigmenti in Horses

Rachel Towers. Funding support from the Ectodermal Dysplasia Society and Newcastle University.

A family of horses have an illness similar to Incontinentia Pigmenti (IP), with a strong family history suggestive of an inherited cause. We are undertaking a study to try and discover the genetic mistake causing the condition in these horses. Non-human examples for IP research include the mouse and the dog; a horse example for IP has not been identified and may provide an avenue for research into larger animals in the future. IP in humans is characterised by skin lesions, where lesions change over time and eventually disappear.



In addition to skin lesions patients with IP can develop abnormalities in their teeth, hair, nails, eyes and central nervous system.

This family of horses develop skin lesions consistent with IP in humans; they also have some microscopic skin features of IP that can't be seen by the naked eye. In addition abnormalities of teeth, nails, hair and eyes are present in these horses.

60-80% of human patients with IP have a specific genetic mistake<sup>1,2,3</sup>. We have started investigating the sequence of this gene in affected horses. We have also begun to investigate whether this gene works differently in abnormal skin lesions compared to unaffected skin. We haven't yet identified a specific genetic mistake; however some results may suggest abnormal functioning of the gene in affected skin. It is too early to say whether these results suggest a mistake in this gene is responsible for the condition in these horses.

We hope to do further work comparing the genes in affected and unaffected horses within this family to identify genes that are only present in affected horses. This provides an exciting and promising opportunity to discover the genetic mistake causing IP in this horse family, which may help us to better understand the condition and possible treatments in humans.

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