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Aurelien Capitan, Cécile Grohs, Agnès Bonnet, Alain Pinton, Brigitte Leguienne, Daniel Le Bourhis, Olivier Bouchez, Christophe Klopp, Stephan Bouet, Per Wahlberg, et al.

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P0570 : Novel Models and Strategies to Unravel the Genetic Pathways Involved in Horn Ontogenesis

[Aurelien Capitan](#) , UNCEIA Genetics Team, Paris, France

Cécile Grohs , INRA UMR1313 Animal Genetics and Integrative Biology, Jouy-en-Josas, France

Aurélie Bonnet , INRA UMR1198 Developmental Biology and Reproduction, Jouy-en-Josas, France

Alain Pinton , INRA-ENVT UMR444 Cell Genetics, Toulouse, France

Brigitte Leguienne , UNCEIA Research and Development Team, Maisons-Alfort, France

Daniel Le Bourhis , INRA UMR1198 Developmental Biology and Reproduction, Jouy-en-Josas, France

Olivier Bouchez , INRA Genomics Facility, Castanet-Tolosan, France

Christophe Klopp , INRA SIGENAE Bioinformatics Team, Castanet-Tolosan, France

Stéphan Bouet , INRA UMR1313 Animal Genetics and Integrative Biology, Jouy-en-Josas, France

Per Wahlberg , INRA UMR1313 Animal Genetics and Integrative Biology, Jouy-en-Josas, France

Sylvain Marthey , INRA UMR1313 Animal Genetics and Integrative Biology, Jouy-en-Josas, France

Christophe Richard , INRA UCEA Experimental Unit of Bressonvilliers, Leudeville, France

Celine Chantry Darmon , Labogena, Jouy-en-Josas, France

Yves Gallard , INRA UE0326 Experimental Unit of Le-Pin-au-Haras, Exmes, France

Eric Pailhoux , INRA UMR1198 Developmental Biology and Reproduction, Jouy-en-Josas, France

Dominique Rocha , INRA UMR1313 Animal Genetics and Integrative Biology, Jouy-en-Josas, France

Didier A. Boichard , INRA UMR1313 Animal Genetics and Integrative Biology, Jouy-en-Josas, France

Andre Eggen , INRA UMR1313 Animal Genetics and Integrative Biology, Jouy-en-Josas, France

After 20 years of research neither the causal mutation for the bovine polled locus nor the genetic pathways involved in horn ontogenesis have been identified and/or published. Researchers have been faced with numerous difficulties specific to this topic: failure to fine-map the polled locus despite intensive genotyping activities in several resource populations together with several targeted sequencing efforts; lack of appropriate models i.e. horned model species make it impossible to identify functional candidate genes from previous studies; absence of candidate polymorphisms in the coding sequences of the positional candidate genes... To overcome these difficulties, we recently screened the whole French cattle population for new syndromes affecting horn development. Among the numerous cases reported, we selected seven different syndromes, all related to unique mutation events, and for which small familial structures were available. We are currently using the most recent technologies to identify the causative mutations and producing fetuses to understand horn bud programming and horn ontogenesis. This approach has already proven successful for the first two cases studied: i) identification of a 10 base pair duplication in the TWIST1 gene causing a novel type 2 scurs syndrome (Capitan *et al.*, 2011) and (ii) more recently, mapping of a 3,7 Mb deletion associated with the novel polled and multi-systemic syndrome (Capitan *et al.*, *in prep.*). Analyses on the other syndromes are ongoing and we are open to collaborations with geneticists working on this research topic.