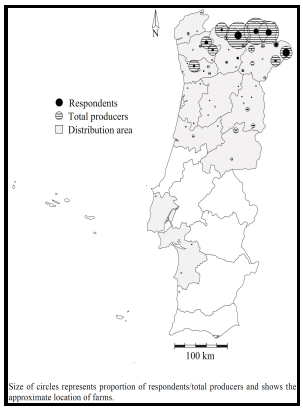
Pig genetic resources in Europe - characterisation and conservation

Wageningen Pers - Assignment Essays



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The child was homozygous for the delta-F508 mutation associated with haplotype B; the father was a compound heterozygote for this mutation and a second CF mutation associated with haplotype C. The presence of microscopic nephrocalcinosis in 3 patients less than 1 year of age suggested to these authors that the mutation in cystic fibrosis involves a primary abnormality of renal calcium metabolism. Moreover, at our academic service, we have our own plagiarism-detection software which is designed to find similarities between completed papers and online sources.

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Infertility suggested that characteristics of cervical mucus may account for infertility in females with cystic fibrosis. A mutation detection rate of 99.

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The patients presented with intestinal obstruction and required surgical resection of a thickened and narrowed area of the colon. In type II, fibroblast metachromasia is present in both vesicles and granules and is evenly distributed through the cytoplasm; mucopolysaccharide content of the cells is markedly increased.

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Delta-F508 gene deletion in cystic fibrosis in Southern Europe.

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Attempting total ascertainment of cases in white children born alive in Ohio during the years 1950 through 1953, estimated the phenotype frequency to be about 1 in 3,700, a value only about one-fourth that of some earlier estimates.

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This is the basis of the folkloric anecdote that the midwife would lick the forehead of the newborn and, if the sweat tasted abnormally salty, predict that the infant was destined to die of pulmonary congestion and its side effects.

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Sequencing of the ENaC subunits revealed heterozygous mutations in the SCNN1A and SCNN1B genes in 4 patients, respectively, whereas the remaining patient was heterozygous for a mutation in both SCNN1B and SCNN1G. Similar analysis in American black families suggested that multiple mutant alleles are found in this population. Pig Model generated pigs with a targeted disruption of both CFTR alleles.

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Considering that the haplotype background of the mutations that most often cause cystic fibrosis in Europe is different from that of non-CF chromosomes, reasoned that these haplotype backgrounds might be found at high frequencies in populations in which CF was currently not common; thus, such populations would be candidates for the place of origin of CF mutations. Cystic fibrosis CF is classically described as a triad of chronic obstructive pulmonary disease, exocrine pancreatic insufficiency, and elevation of sodium and chloride concentration in sweat. The mice survived and showed functional correction of ileal goblet cell and crypt cell hyperplasia and cAMP-stimulated chloride secretion.

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