

# Relation between the FBN2 haplotype and phenotypical hip dysplasia

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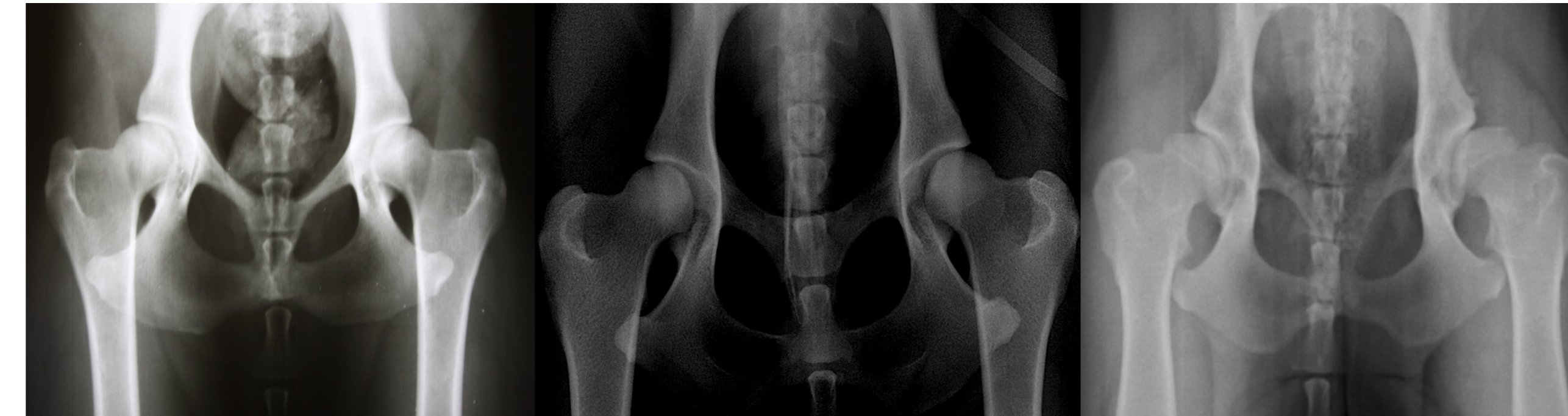
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**Figure 1:** Phenotypical categories (general assessment), from left to right: healthy, inconclusive, dysplastic.

**Table 1:** General and breed specific prevalence and allele frequencies.

	N	Allele frequency			Allele frequency	
		GAT/GAT (%)	GAT/AGC (%)	AGC/AGC (%)	GAT	AGC
All dogs	271	46,5	36,2	17,3	64,6	35,4
German Shepherd	54	50	44,4	5,6	72,2	27,8
Golden Retriever	38	18,4	50	31,6	43,4	56,6
Labrador Retriever	39	48,7	33,3	17,9	65,4	34,6

## Introduction

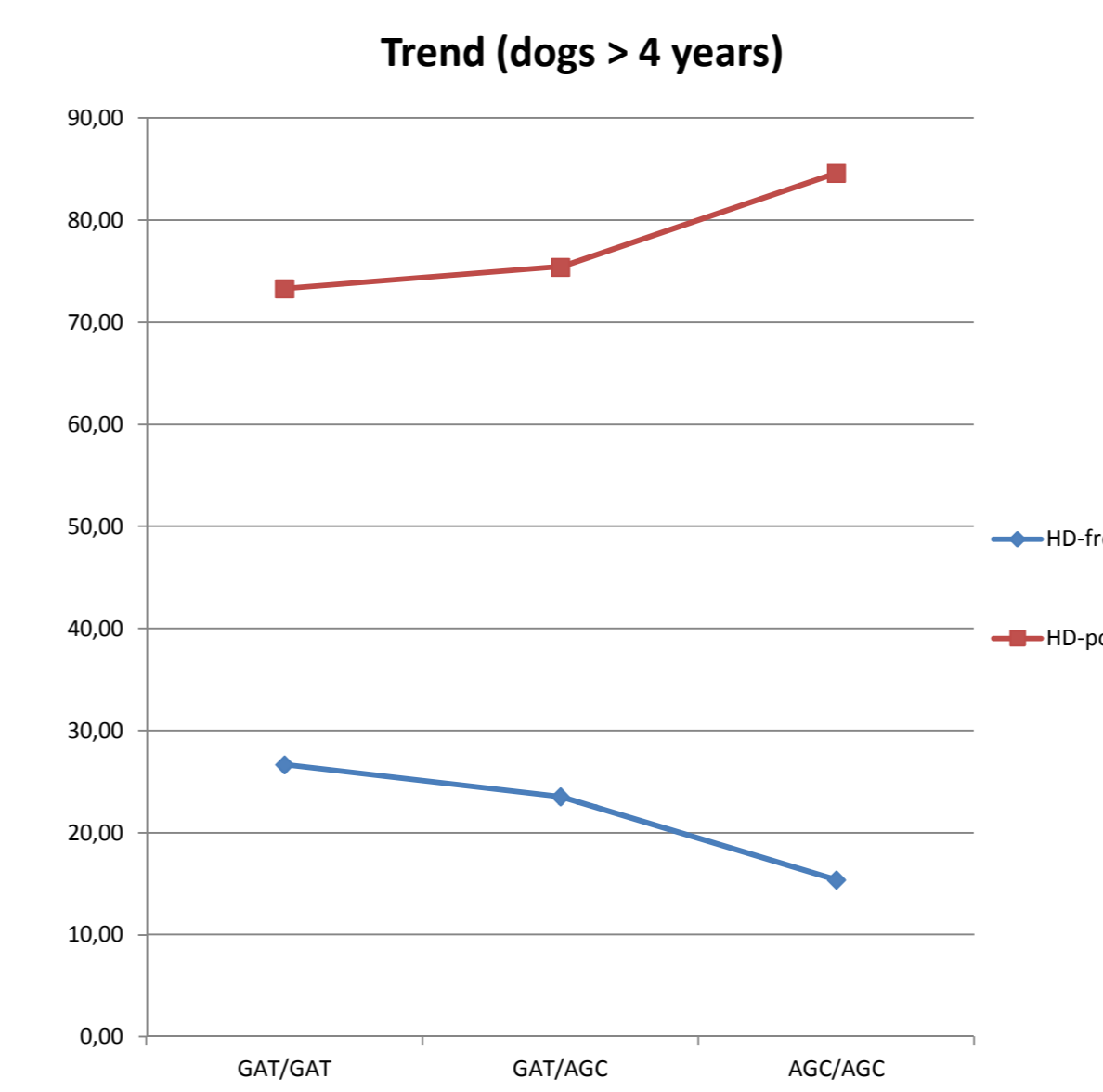
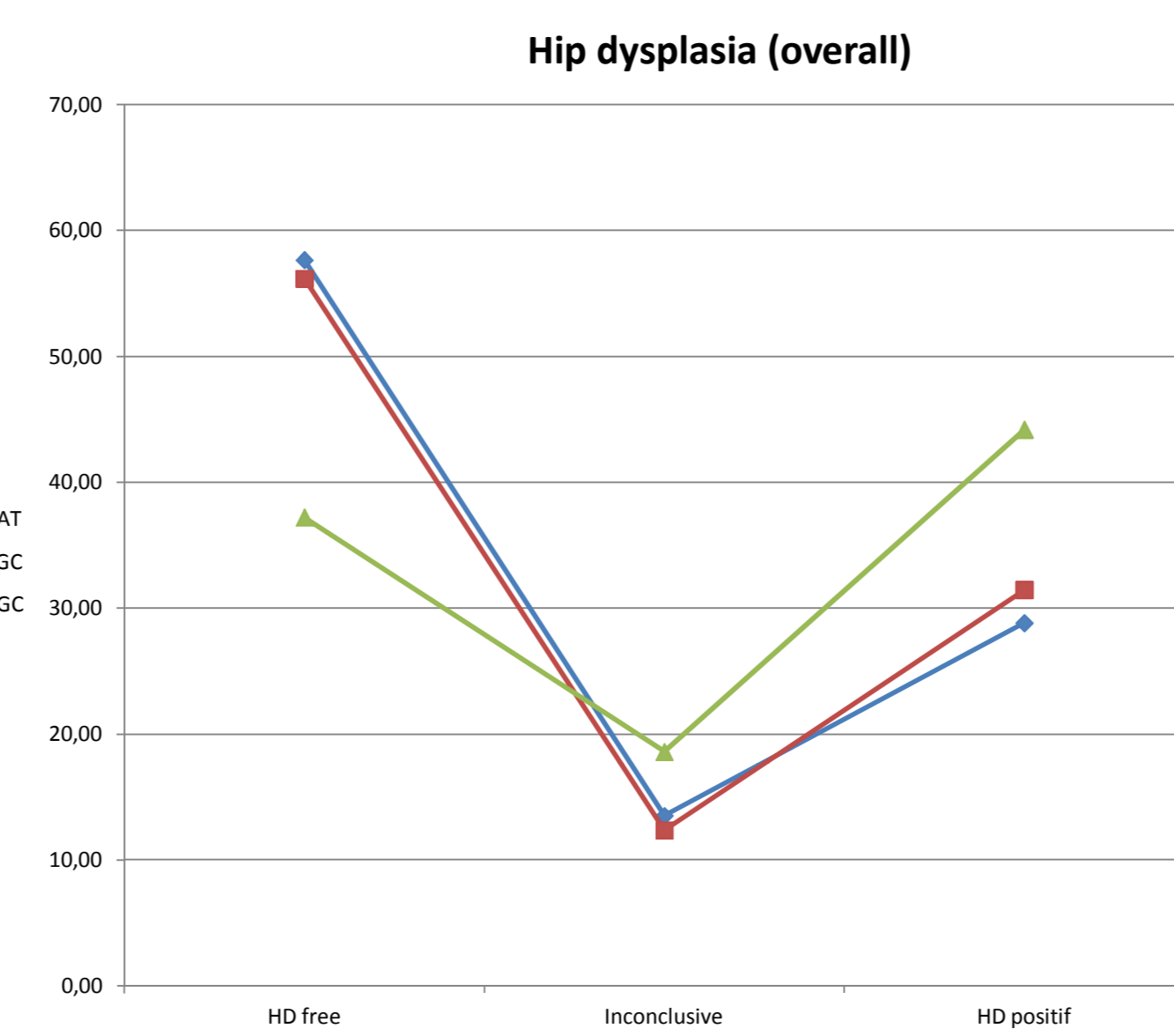
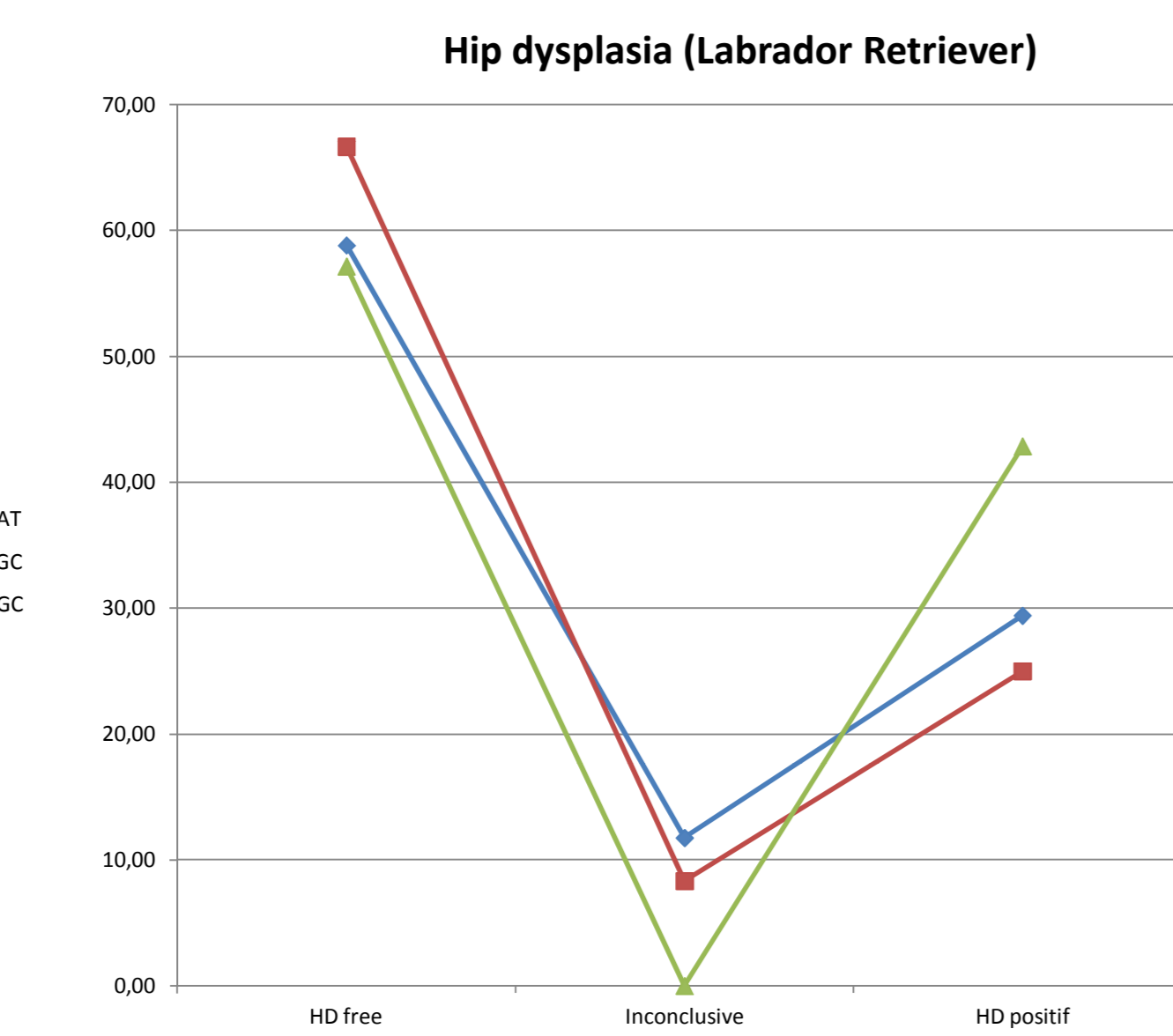
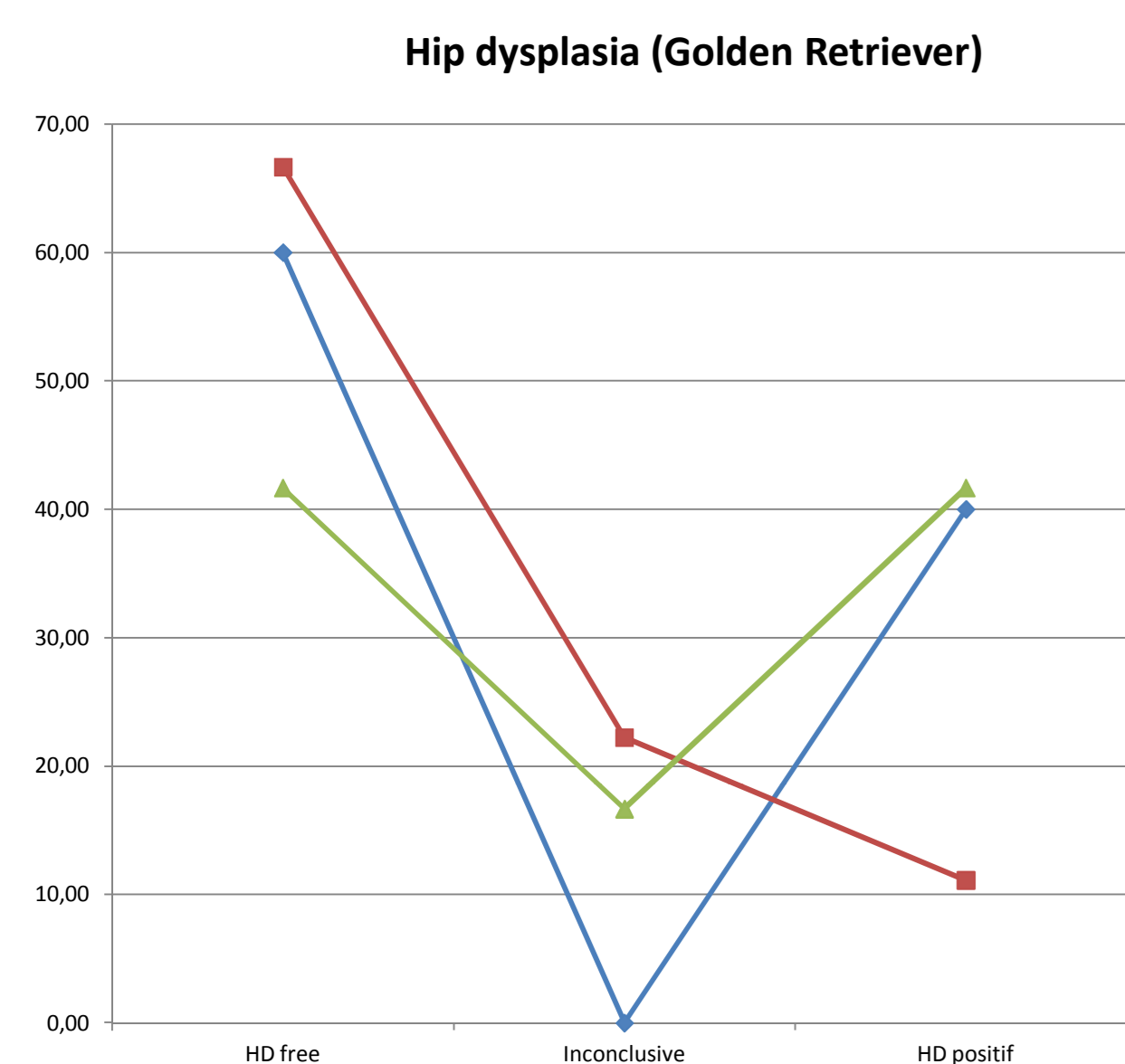
Hip dysplasia is a highly prevalent complex disorder. Despite decades of selection against this disease based on radiographic methods, prevalence has hardly been reduced. Recently, focus has been redirected at the underlying genetics. This has led to the discovery of an association between the AGC haplotype in the FBN2 gene and hip dysplasia, based on an American population of dogs (Friedenberg et al., 2011). Aim of this study was to investigate the prevalence of this mutation in a Dutch and Belgian dog population and to confirm the association of the mutations with phenotypical hip dysplasia.

## Materials and Methods

Blood samples and X-rays (standard hip extended) were collected from dogs of different breeds. Birth date and date of x-ray were noted. Based on the standard hip x-ray, hip status was assessed by consensus (BB, FC) based on four different radiographic categories:

- Hip dysplasia (yes/no/inconclusive) (Figure 1)
- Deg. Joint Dis. (No/Mild/Moderate/Severe),
- Morgan line (Yes/No/Inconclusive)
- Circumferential femoral head osteophyte (Yes/No/Inconclusive).

The dogs were genotyped with KASPar. The prevalence of the haplotype was calculated in the general population and in 3 breeds. The presence of an association between phenotype and genotype was investigated. Statistical analysis was performed with SPSS Statistics 20.



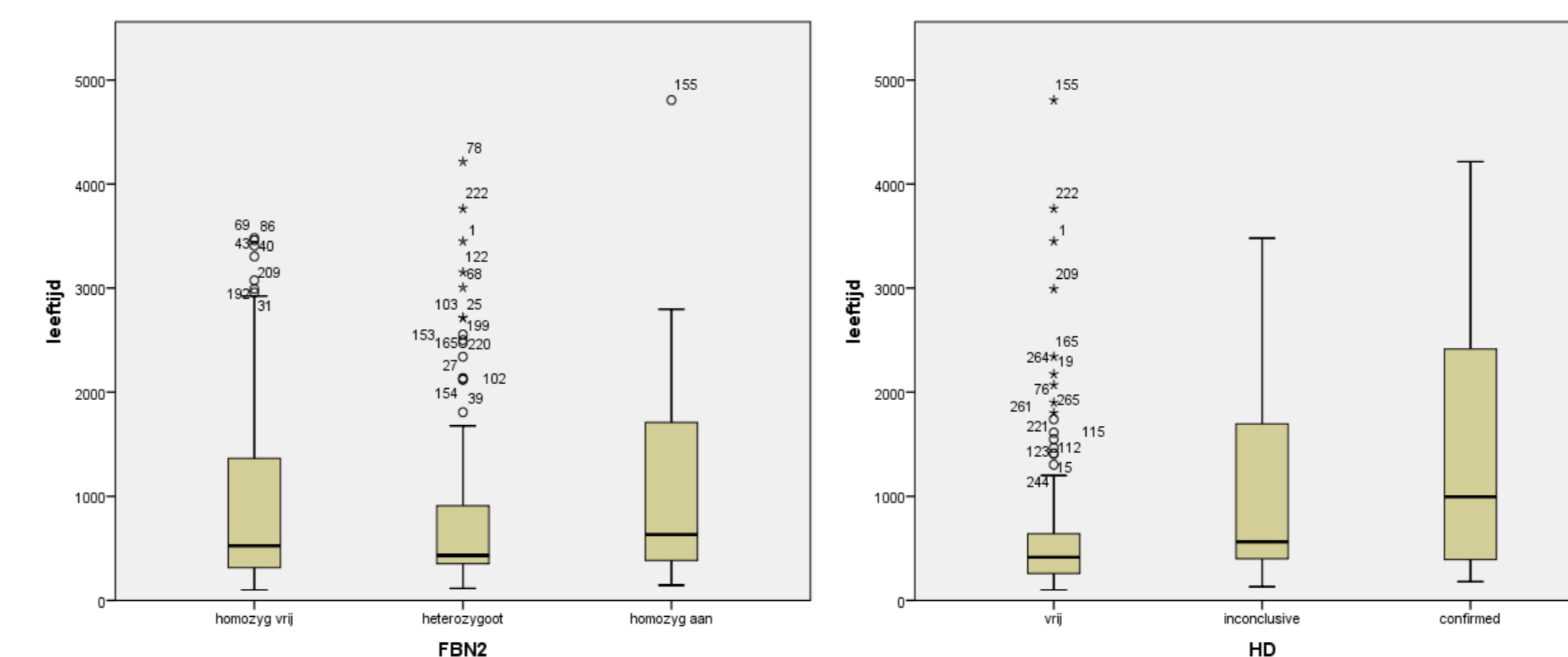
**Figure 3:** Overall and breed specific hip dysplasia.

**Figure 4:** Trend.

## Results

271 dogs of 50 different breeds were genotyped (Table 1). X-rays of 243 dogs were available. There were no significant age differences between groups (FBN2 and HD, Figure 2).

All the radiographic assessments were highly correlated with each other ( $p < 0,001$ ). The AGC haplotype was only significantly correlated with the general assessment ( $p < 0,05$ ). Our model contained two significant coefficients (namely age and FBN2,  $p < 0,05$ ) that explained 14 % of the variation.



**Figure 2:** Age distribution.

## Acknowledgements:

I would like to thank all the vets for sending in samples, my promotor (D. Deforce), my co-promotor (F. Coopman), Elien Verelst, the IWT and Royal Canin for their financial support.

## Conclusions

Although significantly correlated, variation is only partly explained by FBN2 and age. Breed specific differences seem to be present (Figure 3). There is a trend towards worse hip conformation for the homozygous AGC dogs (Figure 4). A more refined phenotyping, a larger amount of animals tested and the inclusion of more environmental factors will be necessary to refine the model. These improvements are being tested at the moment. In general, we reach the same conclusions as presented by Friedenberg et al. (2011) but on a larger database and over more breeds.