

Estimated breeding values for better selection

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Hip and elbow dysplasia are common, multifactorial hereditary diseases leading to osteoarthritis in dogs. Many countries have well-established scoring systems and breeding limitations to combat the problem, but selection based on phenotypes often tends to be slow, even ineffective. *Comparative analyses of genetic trends and prospects for selection against hip and elbow dysplasia in 15 UK dog breeds* was written by Thomas W. Lewis, Sarah C. Blott and John A. Woolliams and published in BMC Genetics journal in 2013. The goal was to compare the state of different breeds after decades of phenotype selection, and to shed light on the possible increased benefits estimated breeding values (EBV) can give to selection against hip and elbow dysplasia. Using regression models, EBV is calculated from generations of pedigree data to give a more complete image of an individual's genotype. As such, estimated breeding values are an effective way to evaluate genetic risk and to enhance selection intensity for the improvement of hip and elbow scores in dogs.

Lewis et al. (2013) begin by introducing the basic functions of hip and elbow dysplasia: Hip dysplasia is a degenerative joint disorder that starts with laxity of the joint and develops into osteoarthritis over time due to poor joint function. Elbow dysplasia is not just that, but is rather an umbrella term to describe a variety of abnormalities in the elbow. Due to this, the heritability of elbow dysplasia has traditionally been estimated lower than for hip dysplasia. (Lewis et al. 2013) Heritability is an important term, and is described well in U.S. National Library of Medicine Genetic Home Reference web page: "Heritability does not indicate what proportion of a trait is determined by genes and what proportion is determined by environment. So, a heritability of 0.7 does not mean that a trait is 70% caused by genetic factors; it means that 70% of the variability in the trait in a population is due to genetic differences among [individuals]." The estimates of heritability for elbow dysplasia range from 0.10 to 0.38, and 0.20 to 0.43 for hip dysplasia. These numbers are moderate and include datasets from various scoring systems. (Lewis et al. 2013)

Next, Lewis et al. (2013) introduce the idea of estimated breeding value (EBV), a tool for improving selection strength. They describe how pedigree data is used to compute a best linear unbiased predictor (BLUP) for each individual, giving a better estimate on its genetic risk (and with it, its "true breeding value") better than its phenotype could. Fifteen popular dog breeds were selected for the study, and their hip and elbow scores along with their pedigree data was retrieved from the British Veterinary Association and UK Kennel Club files. In total, 142 287 hip scores and 13 908 elbow scores across all fifteen breeds were used in this study (Lewis et al. 2013). This is

definitely impressive but also shows the difference in popularity between hip and elbow scoring. The most prominent feature of BLUP is the possibility to factor in those environmental (fixed) effects presented as modifiers of heritability in a previous paragraph. In this study the fixed effects used were sex, inbreeding coefficient, age, date of birth and year of evaluation. The model dynamically adjusts according to the emerging differences in these chosen factors. For example male dogs are more often affected by elbow dysplasia than females (Hou et al. 2013), making poor joints on a female a somewhat bigger negative effect on the EBV of the individual. Similarly, age is a big factor in evaluation and due to the degenerative nature of hip and elbow dysplasia can be the difference between what is considered “healthy” and “sick” joints. (Hou et al. 2013).

The results of the study support the benefit of EBV in breeding selection. Lewis et al. (2013) arrived at a mean heritability of 0.38 for hip dysplasia across all breeds. Without EBV, nearly every breed was found to have improved hip scores over time, even if the improvement rates per year were rather poor and equal to “excluding between less than 2% and less than 18% of the highest risk animals from breeding”. With elbows the selection intensity was even lower, and similar to removing only around 4-8% of the highest risk animals from the breeding population. As a key result of this study, Lewis et al. (2013) present that the selection accuracy was always higher than expected when EBV was used instead of phenotypical selection. Even when the breeding animals themselves had not been scored for hips or elbows, calculating EBV from parental phenotypes along with other pedigree data was sufficient for significant progress. It was concluded that the selection accuracy with EBV would always be higher than just by looking at an individual breeding animal's scores. Improved selection accuracy directly leads to faster and more effective genetic progress, and along with it, naturally phenotypical progress as well. (Lewis et al. 2013)

The authors give a compelling example of the benefits of increased selection pressure. Lewis et al. (2013) state that under the current breeding methods of always selecting for phenotypically “above average” breeding animals, it would take 30 to 300 years to achieve the same progress in joint health as could be achieved in just 9-18 years using EBV. Lewis et al. (2013) further suggest that if kennel clubs published estimated breeding values, the information would become widely available for breeders who need it, and it could also help prospective dog owners with evaluating risks in various available litters, indirectly encouraging breeders to make better choices as well. Naturally phenotyping – the screening and scoring of hips and elbows – should continue with the same, if not increased activity as before, as these raw data are always crucial for the accuracy of EBV calculation and important for the owners of individual animals. (Lewis et al. 2013)

Finally, the paper gives interesting commentary on breeding populations with small numbers. Lewis et al. (2013) acknowledge that it is difficult to select against genetic disorders in a small breed where phenotyped individuals are scarce. Only choosing from the few scored animals can lead to a decline in genetic diversity, making a small population especially vulnerable to over-contribution of certain individuals through the use of “popular sires”. Lewis et al. (2013) are adamant in their stance that selection against hereditary illnesses should still take place even in small populations, but some sort of balance must be reached between control of genetic risks and increased inbreeding. Focusing on eradicating a specific genetic illness by sacrificing genetic diversity will only lead to the emergence of new genetic illnesses. (Lewis et al. 2013)

This study is overall an excellent, direct look into the benefit of estimated breeding values. The dataset was large, but the authors did recognize that it would not be possible to collect this much data in most dog breeds. Because EBV can address any trait, and there are several other degenerative joint diseases than just hip and elbow dysplasia, it could be argued the paper could have addressed only the hips as the most common one. The authors did reference to a possible comorbidity between these two genetic problems, but were not able to find a statistically significant one (Lewis et al. 2013). They also used a rather limited number of fixed effects in their calculations, with the human factor – the person evaluating the images – missing entirely. Finally, while reading the text it became clear that the authors were as enamored by the possibilities of estimated breeding values as the author of this critical summary. The facts that even unphenotyped individuals can be assigned EBVs through family, and that EBV can be calculated for nearly every measurable trait tempts imagination and hopefully inspires many dog breeders. Breeding with EBV is already commonplace in livestock, and after reading this article it is definitely easier to see that there is no reason why it could not work in a pet species as well.

References:

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