Why use is the threshold model in comparative phylogenetics?

Felsenstein, J. 2004. "Threshold models". Inferring Phylogenies. Sinauer. Pp. 429-431.

Discusses the usefulness of the threshold model and its history in quantitative genetics. Lists three benefits over traditional Markov models of discrete character evolution for morphological characters. (1) Naturally incorporates species polymorphisms as well as fixed differences between species. A polymorphic species is a species where the species mean liability is close to the threshold and so individuals in the species occur on both sides of the threshold. (2) Species reversion to a previous state is more common in a species that recently changed as opposed to species that changed a long while ago. This is because a species that recently changed states still has a liability close to the threshold while a species that changed a long time ago may not be close to the threshold. (3) Straightforward calculation of correlations for discrete traits (i.e., the liabilities can be treated like any continuous trait) compared to Markov models where the Q matrix quickly becomes unwieldly (Pagel's correlation). Similarly, can calculate correlations between discrete and quantitative traits (i.e., between the liability and quantitative trait). One major difficulty with the threshold model in phylogenetics is that it is computationally difficult to calculate the liability values.

Maddison, W. and FitzJohn, R.G. 2015. The unsolved challenge to phylogenetic correlation tests for categorical characters. Systematic Biology 64: 127–136.

Comparative analysis of species frequently involves calculating and understanding the significance and strength of correlation between discrete characters to test functional or adaptive linkage across species. Commonly used methods that take into account phylogeny, such as Pagel's 1994 test of correlation or Maddison's 1990 concentrated changes test, can give significant results even when there is a single co-distributed change of characters on the tree. They replicate 100 data sets showing perfect single origin co-evolution for clades of 40-60 species and apply Pagel's test, all 100 data sets returned significant results ('Darwin's scenario'. A less severe example where the second character had multiple changes ('unreplicated burst') showed significant results in 83 of 100 datasets. This is problematic as the test (and other tests) is not accounting for phylogenetic pseudoreplication. There do not appear to be solutions (ie, a new test) or ways to quantify the degree of pseudoreplication 'contaminating' any given data set. Quantitative data, analyzed with PIC for example, can suffer from a similar problem, but these situations can be diagnosed and interpreted as a violation of the Brownian motion assumption. This suggests methods that rely quantitative data might be more robust in general, and point to Felsenstein's threshold model, which relies on the continuous liability to inform correlations.

Threshold model in phylogenetics.

Felsenstein, J. 2005. Using the quantitative genetic threshold model for inferences between and within species. Phil. Trans. Roy. Soc. B 360: 1427–1434.

Preliminary paper that discusses the prospects of the threshold model for between and within species inferences. Proposes the use of a MCMC importance sampling methods to approximate the likelihoods of the liabilities in the interior and for the tips (ensuring that the tip liabilities fall on the right side of the threshold). This paper is followed up with simulation test in Felsenstein (2012).

Hadfield, J.D. and Nakagawa, S. 2010. General quantitative genetic methods for comparative biology: Phylogenies, taxonomies and multi-trait models for continuous and categorical characters. Journal of Evolutionary Biology 23: 494–508.

Explicitly link phylogenetic comparative methods to quantitative genetics involving pedigrees, i.e., 'animal models'. In particular they show that internal/ancestral nodes are like missing parents and that branch lengths are equivalent to the degree of inbreeding. As a result, very efficient methods (orders of magnitude faster) developed over the decades in quantitative genetics can be directly applied to phylogenetic data. They implement this in a generalized linear mixed model framework (R package MCMCglmm) that allows both quantitative and discrete (threshold) responses. In particular they are interested phylogenetic meta-analysis (following Adams 2008), combining taxonomy with phylogeny in analysis where phylogenetic data is not available for all species, and accommodating biases in the species/trait sampling.

Felsenstein, J. 2012. A comparative method for both discrete and continuous characters using the threshold model. American Naturalist 179: 145–156.
A follow up to Felsenstein (2005), this paper illustrates how to use the threshold model for comparative methods of discrete and continuous traits. The liabilities values evolve under a Brownian motion. A MCMC expectation-maximization (EM) algorithm is used to infer the maximum likelihood inference of the evolutionary covariance among changes along the branches. At each EM iteration, a Gibbs sampler is used to infer the continuous characters and liabilities at the interior nodes. A Metropolis samplers is used for drawing the species mean liabilities at the tips, making small changes in the liabilities and accepting or rejecting them according to their conflict with the observed discrete data. In this paper, liabilities of three characters were simulated and the true covariance matrix and correlations were known. To test the method, the author used the simulated discrete data at the tips to estimate liabilities and their correlations. He also tested the method if the tip values were a mix of continuous (liabilities) and discrete. Although the simulations clustered around the true correlation, it was variable. He warns that “When the truth is a positive or a negative correlation, the inference is able to infer only a little more than the sign of the correlation correctly.” Felsenstein emphasizes the potential for the threshold model for within-species polymorphism of both discrete and continuous characters.

This lecture accompanies this paper: https://www.youtube.com/watch?v=7_L3lItQCvI

Revell, L. 2014. Ancestral character estimation under the threshold model from quantitative genetics. Evolution 68: 743–759.
Introduces a new method of ancestral state estimation using the threshold model. Using a Bayesian framework, Revell samples liabilities and threshold from their posterior distribution (MCMC) by a two part likelihood expression that includes 1) probability of the sampled liabilities for the tips and nodes in the tree under Brownian Motion and 2) the probability that the sampled tip liabilities and threshold could account for the observed discrete tip data. Revell simulates three states and four states characters with liability thresholds on 20 100-taxon trees (tips and ancestral states known). He evaluated the method by using the simulated discrete tip data and estimating liabilities and ancestral states across the tree. He evaluated if the estimated ancestral states match those of the simulated data, if the thresholds match those of the generate data. He tested if DIC can be used to determine the true liability state ordering, if Mk models can predict the ancestral state of threshold generated data, and the reciprocal. Lastly he tested the threshold model on empirical data.

Results suggest that 1) threshold model works well for ancestral state estimation when the data is generated under the threshold model, 2) DIC is a good for deciding the order of states on the liability scale, 3) Mk models don’t perform well to estimate ancestral states if the data are generated under the threshold model and 4) Mk models perform best when the data are generated under a Mk model.

Cybis, G.B., Sinsheimer, J.S., Bedford, T., Mather, A.E., Lemey, P., and Suchard, M.A. 2015. Assessing phenotypic correlation through multivariate phylogenetic latent liability model. Annals of Applied Statistics 9: 969–991. (https://projecteuclid.org/download/pdfview_1/euclid.aoas/1437397120)
Cybis et al. introduce efficient algorithms for estimating the liabilities, as a result fixed trees are not necessary, and the tree (with sequence data) and the liabilities (and correlations, thresholds etc) can be estimated simultaneously. Other program/methods outlined here require supplying a single tree or group of trees. In order to perform model testing using Bayes factors a modification to path sampling is introduced that accounts for the truncated distribution of the liabilities induced by the discrete tip states. This can be used to assess the significance of blocks of correlated characters or to assess alternative state orderings of a character. Finally, they also introduce a means of having unordered liabilities in multistate characters by adding additional dimensions to the liabilities. The method is implemented in BEAST (Drummond et al. 2012). They explore and test this method with two human pathogen examples and a floral traits example.

Hadfield, J.D. 2015. Increasing the efficiency of MCMC for hierarchical phylogenetic models of categorical traits using reduced mixed models. Methods in Ecology and Evolution 6: 706–714. (DOI: 10.1111/2041-210X.12354)
Introduces further efficiencies, based on the reduced animal model, in calculating parameter values for threshold models in a generalized linear mixed model for the MCMCglmm R package. He also relaxes the need for Pagel's λ = 1, which Felsenstein's implementation assumes. Compares performance MCMCglmm with Revell 2014's implementation in phytools (R package) and Phylip. Effective sample sizes/time were 3-6 orders of magnitude faster compared to phytools and equivalent to Phylip.