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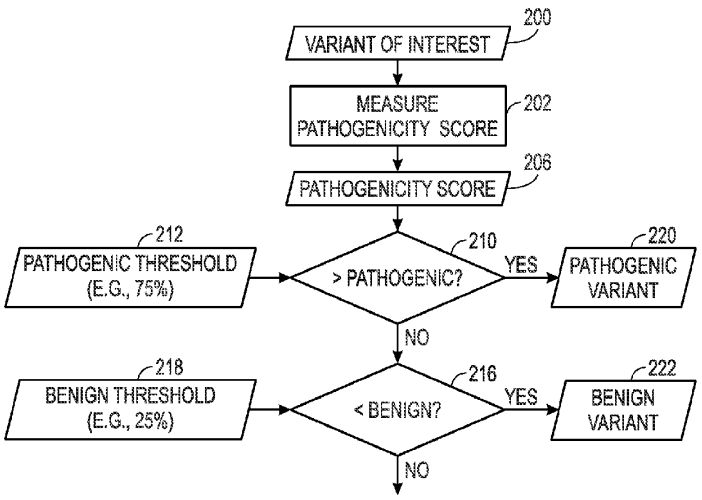

FIG. 7

(57) Abstract: Derivation and use of pathogenicity scores (206) for gene variants are described herein. Applications, uses, and variations of the pathogenicity scoring process include, but are not limited to, the derivation and use of thresholds (212, 218) to characterize a variant as pathogenic or benign, the estimation of selection effects associated with a gene variant, the estimation of genetic disease prevalence using pathogenicity scores (206), and the recalibration of methods used to assess pathogenicity scores (206).

[Continued on next page]

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