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(54) Title: PREDICTING GENE VARIANT PATHOGENICITY

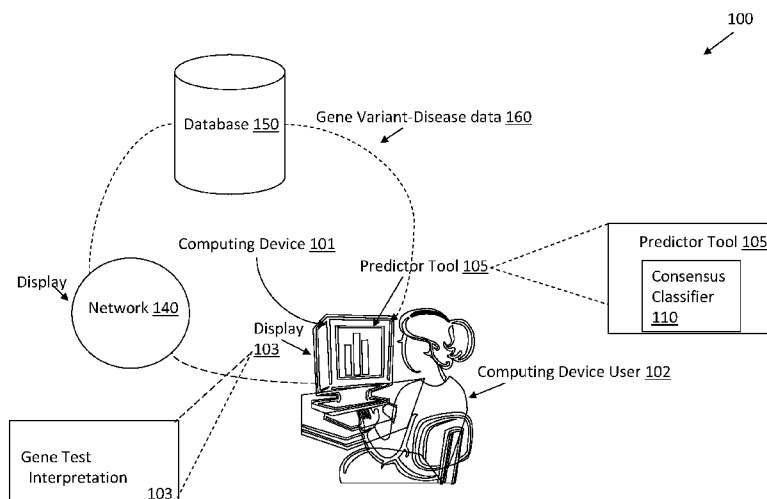


FIG. 1

(57) Abstract: A computer-implemented, gene-specific prediction tool for classifying and interpreting gene tests is described. The prediction tool includes a predictor using a consensus framework. The predictor employs a weighted metric of existing and complementary prediction algorithms and calculated reference intervals of disease outcomes to calculate a consensus score used in interpreting gene tests.





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**A. CLASSIFICATION OF SUBJECT MATTER****G06F 19/10(2011.01)i**

According to International Patent Classification (IPC) or to both national classification and IPC

**B. FIELDS SEARCHED**

Minimum documentation searched (classification system followed by classification symbols)

G06F 19/10; G06F 19/00; G06Q 50/00; G01N 33/50; C12Q 1/68; G06N 5/02

Documentation searched other than minimum documentation to the extent that such documents are included in the fields searched

Korean utility models and applications for utility models

Japanese utility models and applications for utility models

Electronic data base consulted during the international search (name of data base and, where practicable, search terms used)

eKOMPASS(KIPO internal) &amp; Keywords: gene, genetic, predict, analysis

**C. DOCUMENTS CONSIDERED TO BE RELEVANT**

Category*	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
A	US 2009-0177496 A1 (TUCK EDWARD F. et al.) 09 July 2009 See abstract; claims 1, 28, 47-49; and figure 34.	1-20
A	US 2004-0137436 A1 (BRENDAN LARDER et al.) 15 July 2004 See abstract; claim 4; and figures 10A-10B.	1-20
A	US 2006-0278241 A1 (GUALBERTO RUANO) 14 December 2006 See abstract; claims 1, 9.	1-20

 Further documents are listed in the continuation of Box C. See patent family annex.

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**PCT/US2012/037844**

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