

Title (en)

A PRIORITIZATION AND SCORING METHOD

Title (de)

PRIORISIERUNGS- UND REIHUNGSVERFAHREN

Title (fr)

PROCÉDÉ DE CLASSEMENT PAR ORDRE DE PRIORITÉ ET DE NOTATION

Publication

EP 4022646 A4 20221102 (EN)

Application

EP 20907928 A 20201224

Priority

- TR 201921589 A 20191225
- TR 2020051374 W 20201224

Abstract (en)

[origin: WO2021133351A1] The invention relates to a prioritization and scoring method which facilitates the interpretation of the genetic variants (in VCF file formed as a result of the bioinformatics pipeline) by the user, using machine learning for the analysis of Next-Generation Sequencing (NGS) data.

IPC 8 full level

G16H 50/20 (2018.01); **G16B 20/20** (2019.01); **G16B 40/20** (2019.01)

CPC (source: EP)

G16B 20/20 (2019.01); **G16B 40/00** (2019.01); **G16H 40/20** (2017.12)

Citation (search report)

- [I] US 2016357903 A1 20161208 - SHENDURE JAY [US], et al
- [I] IMANE BOUDELLIOUA ET AL: "DeepPVP: phenotype-based prioritization of causative variants using deep learning", BMC BIOINFORMATICS, BIOMED CENTRAL LTD, LONDON, UK, vol. 20, no. 1, 6 February 2019 (2019-02-06), pages 1 - 8, XP021269973, DOI: 10.1186/S12859-019-2633-8
- See references of WO 2021133351A1

Designated contracting state (EPC)

AL AT BE BG CH CY CZ DE DK EE ES FI FR GB GR HR HU IE IS IT LI LT LU LV MC MK MT NL NO PL PT RO RS SE SI SK SM TR

Designated extension state (EPC)

BA ME

Designated validation state (EPC)

KH MA MD TN

DOCDB simple family (publication)

WO 2021133351 A1 20210701; EP 4022646 A1 20220706; EP 4022646 A4 20221102

DOCDB simple family (application)

TR 2020051374 W 20201224; EP 20907928 A 20201224