

PgmNr 674: Greater than the sum: Thought experiment fusing HUGO Gene Nomenclature Committee assigned gene numbers into International Classification of Diseases for the coding of genetic variants.

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With the growing use of gene panels and next generation sequencing in the evaluation of human disease, gene-level description of illness is being realized on a more widespread basis. Despite this growing understanding, genetic variants and their attendant diseases remain difficult to study particularly due to a lack of pragmatic classification of genetic variants within medical nomenclature, e.g. International Classification of Diseases.

The internationally recognized HUGO Nomenclature Committee (HGNC) approves labelling of human loci to enable unambiguous scientific communication, including assignment of a unique HGNC ID number, e.g. *SYNGAP1* has been assigned the ID HGNC #11497. Even should the name or symbol of a gene change, the assigned HGNC ID remains stable. As a unique, durable identifier of a gene, it is an ideal label of pathogenic genetic variation. To code genetic variants in ICD-10, appending the HGNC ID to an ICD-10 prefix could be performed in the Q9 section (Chromosomal abnormalities, not elsewhere classified) of the Q chapter (Congenital malformations, deformations and chromosomal abnormalities), or to an unused U code chapter (U9 Germline Genetic Variants), i.e. Q91.1497 or U91.1497 for “*SYNGAP1* Pathogenic Variant Present, Germline or Postzygotic,” respectively. This methodology preserves the architecture of ICD-10 while utilizing an additional international system to remain consistent with the conventions of biomedical classification with numerous consequent benefits. Use of HGNC ID numbers within the Extension Codes of ICD-11 will then allow ongoing coding of pathogenic variants.

As such codes are entered into electronic medical records and billing databases, high-quality epidemiologic research and retrospective studies are possible. Moreover, in syndromes with multiple individual genetic causes, e.g. frontotemporal dementia or autism, gene-specific labelling can facilitate more detailed genotypic:phenotypic correlation or selective recruitment for clinical studies—a particularly important benefit with the rapid emergence of gene-specific therapies. Furthermore, this method can be adapted to add much-needed somatic mutation codes to ICD-10 as oncologic care increasingly considers personalized tumor genomics, e.g. U code chapter (U7 Somatic Genetic Variants), U79.047 “*PLAGL2* Pathogenic Variant Present, Somatic.” Use of the HGNC ID within ICD can begin a pragmatic and enduring method of incorporating genetic variation into medical coding.