

L^AT_EX 项目模板

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目录

目录	3
第一章 General	5
1.1 General recommendations	5
第一部分 附录	7
1.2 元素周期表	9

第一章 General

Since references to web sites are not yet acknowledged as citations, please mention Den Dunnen et al. (2016) HGVS recommendations for the description of sequence variants: 2016 update. Hum.Mutat. 25: 37: 564-569 when referring to these pages. Note that although the examples on these pages mainly give examples for human (Homo sapiens), the recommendations can be applied to all species.

1.1 General recommendations

1. all variants should be described at the most basic level, **the DNA level**. Descriptions at the RNA and/or protein level may be given in addition.
 - descriptions should make clear whether the change was **experimentally determined or theoretically deduced** by giving predicted consequences in parentheses
 - descriptions at RNA/protein level should describe the changes observed on that level (RNA/protein) and not try to incorporate any knowledge regarding the change at DNA-level (see Questions below)
2. all variants should be described in relation to an **accepted reference sequence** (see Reference Sequences).
 - the reference sequence file used should be **public** and **clearly described**, e.g. NC_000023.10, LRG_199, NG_012232.1, ENST00000357033, NM_004006.2, NR_002196.1, NP_003997.1, etc.
 - when variants are not reported in relation to a genomic reference sequence from a recent genome build, the preferred reference sequence is a **Locus Reference Genomic sequence (LRG)**
 - when no LRG is available, one should be requested (see Reference Sequences).
 - the reference sequence used must contain the residue(s) described to be changed.
 - a letter prefix should be used to indicate the type of reference sequence used. Accepted prefixes are;
 - “g.” for a **genomic reference sequence**
 - “c.” for a **coding DNA reference sequence**
 - “n.” for a **non-coding DNA reference sequence**

- “r.” for an **RNA reference sequence (transcript)**
 - “p.” for a **protein reference sequence**
 - numbering of the residues (nucleotide or amino acid) in relation to the reference sequence used should follow the approved scheme (see Numbering)
3. 3’ rule: for all descriptions the most 3’ position possible of the reference sequence is arbitrarily assigned to have been changed
- the 3’ rule also applies for changes in single residue stretches and tandem repeats (nucleotide or amino acid)
 - the 3’ rule applies to ALL descriptions (genome, gene, transcript and protein) of a given variant
4. descriptions at DNA, RNA and protein level are clearly different:
- **DNA-level** 123456A>T (see Details): number(s) referring to the nucleotide(s) affected, nucleotides in **CAPITALS** using **IUPAC-IUBMB assigned nucleotide symbols**
 - **RNA-level** 76a>u (see Details): number(s) referring to the nucleotide(s) affected, nucleotides in **lower case** using **IUPAC-IUBMB assigned nucleotide symbols**
 - **protein level** Lys76Asn (see Details): the amino acid(s) affected in **3- or 1-letter** followed by a number IUPAC-IUBMB assigned amino acid symbols * **three-letter amino acid code is preferred** (see Standards)
5. prioritisation: when a description is possible according to several types, the preferred description is: (1) deletion, (2) inversion, (3) duplication, (4) conversion, (5) insertion
- when a variant can be described as a duplication or an insertion, prioritisation determines it should be described as a duplication
6. only approved **HGNC gene symbols** should be used to describe genes or proteins

第一部分

附录

1.2 元素周期表

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