Recommendations for the description of RNA sequence variants

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Since references to WWW-sites are not yet acknowledged as citations, please mention den Dunnen JT and Antonarakis SE (2000). Hum.Mutat. 15:7-12 when referring to these pages.

Contents

RNA level

(suggestions extending the published recommendations NEW *in italics)*

Designations at RNA-level, similar to those at protein level, describe the consequence of a change; the origin lies at DNA-level. Sequence

changes at RNA level should be given ***only when RNA was experimentally analysed***. Sequence changes at RNA level are described as those at the DNA level with the following modifications / additions;

- an "***r.***" is used to indicate that a change is described at RNA level
- nucleotides are designated by the bases (in lower case); r.78u>a denotes that at nucleotide 78 a U is changed to an A
  - a (adenine)
  - c (cytosine)
  - g (guanine)
  - u (uracil)
- **more than one transcript**
  when one change affects RNA-processing, yielding two or more transcripts, these are described between square brackets, separated by a ","-character (*see Discussion*)
  NOTE: two transcripts usually means that also two different proteins are produced (*see Recommendations*)
  - r.[=, 73_88del] denotes the nucleotide change c.76A>C causing the appearance of two RNA molecules, one normal transcript (r.=) and one containing a deletion of nucleotides 73 to 88 (shift of the splice donor site to within the exon)
  - r.[76a>c, 73_88del] denotes the nucleotide change c.76A>C causing the appearance of two RNA molecules, one carrying variant 76a>c and one containing a deletion of nucleotides 73 to 88 (shift of the splice donor site to within the exon)
  - r.[88g>a; 88_89ins88+1_88+45] denotes the nucleotide change c.88G>A causing an insertion of the intronic nucleotides 88+1 to 88+45 (shift of the splice donor site to an intronic position)
  - r.[=, 88_89ins88+1_88+10; 88+2u>c] denotes the intronic variant c.88+2T>C causing the appearance of two RNA molecules, one normal (r.=) and one containing an insertion of the intronic nucleotides 88+1 to 88+10 with the nucleotide change 88+2t>c
  - r.[11712del, 11712delc;11715dup] describes the effect of variant c.11712delC (APOB gene) yielding two RNA molecules, one with this deletion (r.11712del) and one containing an extra A nucleotide (r.11712delc;11715dup) reopening the reading frame in a low proportion of the transcripts
- ***effect on RNA***
  *when changes are reported at RNA level that have been **deduced only** (i.e. without any experimental proof) this should be clearly indicated. The suggested description is **between brackets**, to indicate that it is a deduction only. For changes where an effect on RNA is very likely (e.g. when the splice site is directly affected) or can reasonably be expected (e.g. when the promoter is affected) this may be indicated in the description. In such cases, although RNA has not been analysed, since these changes are likely to be pathogenic one is tempted to list these changes and indicate the deduced effect. Suggested descriptions are;*
  - *unknown effect*
    - *r.? - RNA has not been analysed, an effect is expected but difficult to predict (e.g. when the transcription start site, the polyA-addition signal or the polyA-addition site is changed)*
    - *r.(?) - RNA has not been analysed but an effect other then that caused by the change at DNA level is not expected*
    - *r.(=) - RNA has not been analysed, but no change is expected*
  - *probable effect*
    - *r.(76a>u) - RNA has not been analysed, the change expected is r.76a>u (can also be described as r.(?))*
  - *amount of RNA*

changes which affect the promoter of a gene and/or the transcription initiation site (cap site) may affect the amount of RNA produced. Similarly, a deletion of the promoter / exon 1 region usually has the effect that no RNA is produced (or that other promoters are activated).

- *r.0 - no RNA can be detected*
- *r.0? - probably no RNA is produced*

- *effect on splicing*
  - *r.spl? - the change is expected to affect splicing, e.g. when the splice donor or splice acceptor site is changed*
  - *r.(spl?) - the change might affect splicing, e.g. changes close to the splice donor or splice acceptor site or in the first or last nucleotide of an exon*

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