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Mutalyzer 2.0 β-2
released on 16 Aug 2010

HGVS nomenclature version 2.0

Nomenclature check

Please insert the mutation name using the HGVS format:
<Accession Number>.<version number>(<Gene symbol>):<sequence type>.<mutation>

Example: AB026906.1:c.274G>T

NM_000059.3(BRCA2):c.9382C>T

Submit   Clear field

Exon 25 (last exon is 27)

Mutalyzer output:

0 Errors, 0 Warnings.

79310   *79335*   *79360*

Overview of the raw variants:

Raw variant 1: substitution at 9609
AATTGCTGCAAGCAACCTCCAGTGG C GACCAGAATCCAAATCAGGCCTTCT
AATTGCTGCAAGCAACCTCCAGTGG T GACCAGAATCCAAATCAGGCCTTCT

Description relative to transcription start:
(Not for use in LSDBs in case of protein-coding transcripts).

NM_000059.3:n.9609C>T

OH 17/8/10

Affected transcripts:

NM_000059.3(BRCA2_v001):c.9382C>T

NP_000050.2

Affected proteins:

NM_000059.3(BRCA2_i001):p.(Arg3128*)

Detailed information about the selected transcript and predicted protein:

Reference protein:

```
  1   MPIGSKERPT FFEIFKTRCN KADLGPISLN WFEELSSEAP PYNSEPAEES EHKNNNYEP
 61   LFKTPQRKPS YNQLASTPII FKEQGLTLPL YQSPVKELDK FKLDLGRNVP NSRHKSLRT
121   KTKMDQADDV SCPLLNSCLS ESPVVLQCTH VTPQRDKSVV CGSLFHTPKF VKGRQTPKH
181   SESLGAEVDP DMSWSSSLAT PPTLSSTVLI VRNEEASETV FPHDTTANVK SYFSNHDES
241   KKNDRFIASV TDSENTNQRE AASHGFGKTS GNSFKVNSCK DHIGKSMPNV LEDEVYETV
301   DTSEEDSFSL CFSKCRTKNL QKVRTSKTRK KIFHEANADE CEKSKNQVKE KYSFVSEVE
361   NDTDPLDSNV ANQKPFESGS DKISKEVVPS LACEWSQLTL SGLNGAQMEK IPLLHISSC
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