LEIDEN UNIVERSITY MEDICAL CENTER

LaTeX Templates

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Introduction

For the people who use LaTeX:

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  - Presentations.
  - Posters.

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- Aims to reuse/generate as much data as possible:
  - Slide heading.
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  - Page numbers.
- The output (pdf) will always work.

LaTeX presentation

First, retrieve the template.

```
1   svn co https://www.mutalyzer.nl/svn/presentation
```

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Any future fixes / features can be added to *all* presentations without modifying any of them.

LaTeX presentation

Making a new presentation:

```
1  cd presentation/trunk
2  sh mkpres ~/presentations/myPresentation
3  cd ~/presentations/myPresentation
```

Listing 3: Make a new presentation.

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```
1  cd presentation/trunk
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```

Listing 3: Make a new presentation.

This will make a new directory myPresentation with symbolic links to files shared by all presentations and one example file: **`presentation.tex`**.

The content of your presentation goes into this file.

LaTeX presentation

Symbolic links are used to update *all* of your presentations when the template is updated.

Name	Type	Description
`presentation.tex`	file	example presentation
`beamerthemelumc.sty`	link	stylesheet
`Makefile`	link	build script
`lumc_logo.eps`	link	picture
`lumc_logo_small.eps`	link	picture
`ul_logo.eps`	link	picture
`lgtc_logo.eps`	link	picture
`gen2phen_logo.eps`	link	picture
`nbic_logo.eps`	link	picture

Table 1: Files in a presentation.

LaTeX presentation

Edit **presentation.tex**.

Variable	Example
\title	\LaTeX\ Templates
\myConference	Bioinformatics work discussion
\myDate	Tuesday, 17 May 2011
\author	Jeroen F. J. Laros
\myGroup	Leiden Genome Technology Center
\myDepartment	Department of Human Genetics
\myCenter	Center for Human and Clinical Genetics
\lastCenterLogo	\includegraphics[scale = 0.055]lgtc_logo
\lastRightLogo	\includegraphics[scale = 0.1]nbic_logo

Table 2: Configuration variables.

LaTeX presentation

Documentation of the beamer class:
https://bitbucket.org/rivanvx/beamer/wiki/Home

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Build the presentation:

```
1   make release
```

Listing 4: Builing a presentation.

You now have an additional file: **presentation.pdf**.

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Documentation of the beamer class:
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Build the presentation:

```
1   make release
```

Listing 4: Builing a presentation.

You now have an additional file: **presentation.pdf**.

Clean everything (except the source of course):

```
1   make distclean
```

Listing 5: Only keep the source.

LaTeX presentation

Example:

LaTeX presentation

Example:

This presentation.

LaTeX poster

Largely the same idea as with the presentation template:

- Retrieving the template.
- Updating.
- Creating a new poster.
- Editing the example.
- Building.
- Cleaning.

LaTeX poster

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Exceptions:

- Column width can be altered.
    - Unbalanced.

LaTeX poster

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Exceptions:

- Column width can be altered.
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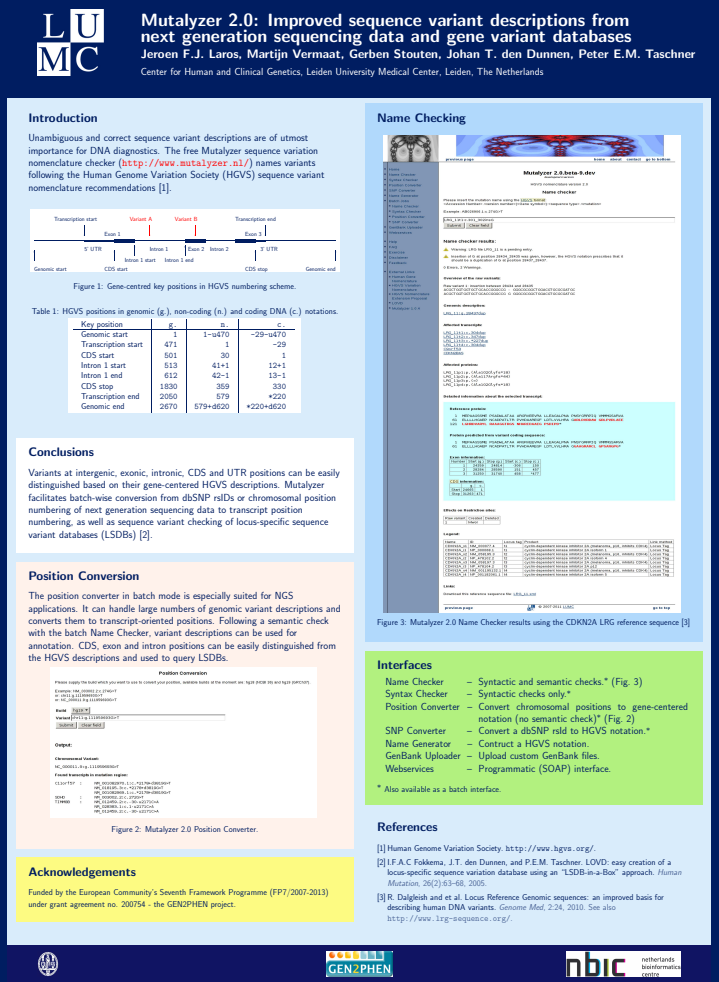
Optional if you use a bibtex bibliography database:

```
1    make bib
```

Listing 6: Generate a bibliography file.

LaTeX poster



Mutalyzer 2.0: Improved sequence variant descriptions from next generation sequencing data and gene variant databases

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Introduction

Unambiguous and correct sequence variant descriptions are of utmost importance for DNA diagnostics. The free Mutalyzer sequence variation nomenclature checker (http://www.mutalyzer.nl/) names variants following the Human Genome Variation Society (HGVS) sequence variant nomenclature recommendations [1].

Figure 1: Gene-centred key positions in HGVS numbering scheme.

Table 1: HGVS positions in genomic (g.), non-coding (n.) and coding DNA (c.) notations.

Key position	g.	n.	c.
Genomic start	1	1−u470	−29−u470
Transcription start	471	1	−29
CDS start	501	30	1
Intron 1 start	513	41+1	12+1
Intron 1 end	612	42−1	13−1
CDS stop	1930	359	330
Transcription end	2050	579	*220
Genomic end	2670	579+d620	*220+d620

Conclusions

Variants at intergenic, exonic, intronic, CDS and UTR positions can be easily distinguished based on their gene-centered HGVS descriptions. Mutalyzer facilitates batch-wise conversion from dbSNP rsIDs or chromosomal position numbering of next generation sequencing data to transcript position numbering, as well as sequence variant checking of locus-specific sequence variant databases (LSDBs) [2].

Position Conversion

The position converter in batch mode is especially suited for NGS applications. It can handle large numbers of genomic variant descriptions and converts them to transcript-oriented positions. Following a semantic check with the batch Name Checker, variant descriptions can be used for annotation. CDS, exon and intron positions can be easily distinguished from the HGVS descriptions and used to query LSDBs.

Figure 2: Mutalyzer 2.0 Position Converter.

Acknowledgements

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Name Checking

Figure 3: Mutalyzer 2.0 Name Checker results using the CDKN2A LRG reference sequence [3]

Interfaces

Name Checker	–	Syntactic and semantic checks.* (Fig. 3)
Syntax Checker	–	Syntactic checks only.*
Position Converter	–	Convert chromosomal positions to gene-centered notation (no semantic check)* (Fig. 2)
SNP Converter	–	Convert a dbSNP rsId to HGVS notation.*
Name Generator	–	Contruct a HGVS notation.
GenBank Uploader	–	Upload custom GenBank files.
Webservices	–	Programmatic (SOAP) interface.

* Also available as a batch interface.

References

[1] Human Genome Variation Society. http://www.hgvs.org/.

[2] I.F.A.C Fokkema, J.T. den Dunnen, and P.E.M. Taschner. LOVD: easy creation of a locus-specific sequence variation database using an "LSDB-in-a-Box" approach. *Human Mutation*, 26(2):63–68, 2005.

[3] R. Dalgleish and et al. Locus Reference Genomic sequences: an improved basis for describing human DNA variants. *Genome Med*, 2:24, 2010. See also http://www.lrg-sequence.org/.

Figure 1: Example poster.

Questions?

https://www.mutalyzer.nl/svn/presentation

https://www.mutalyzer.nl/svn/poster