



## A snapshot of some pLI score pitfalls

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Titre	A snapshot of some pLI score pitfalls
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Auteur	Ziegler, Alban [1], Colin, Estelle [2], Goudenège, David [3], Bonneau, Dominique [4]
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Mots-clés	exome sequencing analysis [5], loss of function [6], pLI score [7]
Résumé en anglais	<p>The pLI score reflects the tolerance of a given gene to the loss of function on the basis of the number of protein truncating variants, that is, the frameshift, splice donor, splice acceptor, and stop-gain variants referenced for this gene in control databases weighted by the size of the gene and the sequencing coverage. It is frequently used to prioritize candidate genes when analyzing whole exome or whole genome data. We list here the main pitfalls to consider before using this score. Concrete illustrations are given for each of these pitfalls.</p>
URL de la notice	<a href="http://okina.univ-angers.fr/publications/ua19792">http://okina.univ-angers.fr/publications/ua19792</a> [8]
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### Liens

- [1] <http://okina.univ-angers.fr/publications?f%5Bauthor%5D=34150>
- [2] <http://okina.univ-angers.fr/e.colin/publications>
- [3] <http://okina.univ-angers.fr/publications?f%5Bauthor%5D=29951>
- [4] <http://okina.univ-angers.fr/d.bonneau/publications>
- [5] <http://okina.univ-angers.fr/publications?f%5Bkeyword%5D=28639>
- [6] <http://okina.univ-angers.fr/publications?f%5Bkeyword%5D=28640>

- [7] <http://okina.univ-angers.fr/publications?f%5Bkeyword%5D=28641>
- [8] <http://okina.univ-angers.fr/publications/ua19792>
- [9] <http://dx.doi.org/10.1002/humu.23763>
- [10] <https://onlinelibrary.wiley.com/doi/abs/10.1002/humu.23763>
- [11] <http://www.ncbi.nlm.nih.gov/pubmed/30977936?dopt=Abstract>

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