ISCA DATABASE LINKS:

<http://www.ncbi.nlm.nih.gov/dbvar/studies/nstd37/download/?type=i>

<http://www.ncbi.nlm.nih.gov/dbvar/studies/nstd37/>

<https://www.iscaconsortium.org/index.php/faq>

dbVAR

<http://www.ncbi.nlm.nih.gov/dbvar>

Copy loss = deletion?

Copy gain = duplication?

Perché su ClinVar separa queste 4 classi

Definizione: Analysis of individual human genomes has revealed an unexpected amount of variability in human populations. The most common form of genetic variation involves small changes in the genetic code that alter a single base pair. Other [types of mutations](http://www.nature.com/scitable/topicpage/Genetic-Mutation-1127) range from small insertions to large[chromosomal rearrangements that can be detected cytogenetically using a microscope](http://www.nature.com/scitable/topicpage/Chromosome-Abnormalities-and-Cancer-Cytogenetics-879). The term "copy number variation" refers to an intermediate-scale genetic change, operationally defined as segments greater than 1,000 base pairs in length but typically less than 5 megabases, which is the cytogenetic level of resolution. CNVs include both additional copies of sequence (duplications) and losses of genetic material (deletions). Because CNVs change the structure of the genome, such mutations, together with inversions and translocations, are collectively classified as forms of genome structural variation. Recently, scientists have come to appreciate that CNVs account for much of human variability.

Sembra quindi che copy loss = deletion e copy gain = duplication, quindi perché clivar le ha splittate????