



Flowchart for application of functional codes PS3 and BS3 via protein function for missense variants (left) and small deletions (right)

*Eligible functional studies include Kato (PMID: 12826609), Funk (PMID 39774325), Giacomelli (PMID: 30224644), Kotler (PMID: 29979965), Kawaguchi (PMID: 16007150), as well as other assays considered to provide relevant functional information after expert interpretation, as previously reported (PMID: 33300245)