Ambry Genetics® | Ambry Classifi™

Ambry Multigenic[1] Copy Number Variation Classification Scheme

Classification Score				
Benign ≤-4 points	Likely Benign -3 to -2 points	Uncertain Significance -1 to +5 points	Likely Pathogenic +6 to +9 points	Pathogenic ≥ +10 points

	Point Range	Code	Criteria
CLINICAL	0.25 to 10.00+	CLN_PHE	**Proband Counting** (unrelated affected individuals without an alternate molecular basis for disease)
	1.25 to 10.00+	CLN_DNV	***de novo* Variant** (with confirmed or unconfirmed parental relationships)
	-8.00 to 4.00	CLN_SEG	**Segregation** (cosegregation or lack of segregation with disease in affected family members)
	-8.00 to 6.00	CLN_CCR	**Case-Control Studies** (significant disease association or no association with disease)
GENE CONTENT	-2.00 to 10.00	GEN_DEL	**Genes Contained within a Copy Number Loss** (assessment of the number and identity of impacted genes)
	-2.00 to 10.00	GEN_DUP	**Genes Contained within a Copy Number Gain** (assessment of the number and identity of impacted genes)
FREQUENCY	-8.00 to -1.00	POP_FRQ	**Population Frequency** (general population or subpopulation frequency too high to be pathogenic based on disease prevalence and penetrance)

[1]The Ambry multigenic copy number variation classification scheme is intended for use with copy number gains or losses that involve multiple genes.

One Enterprise, Aliso Viejo, CA 92656 USA Toll Free +1.866.262.7943 Fax +1.949.900.5501 **ambrygen.com**

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