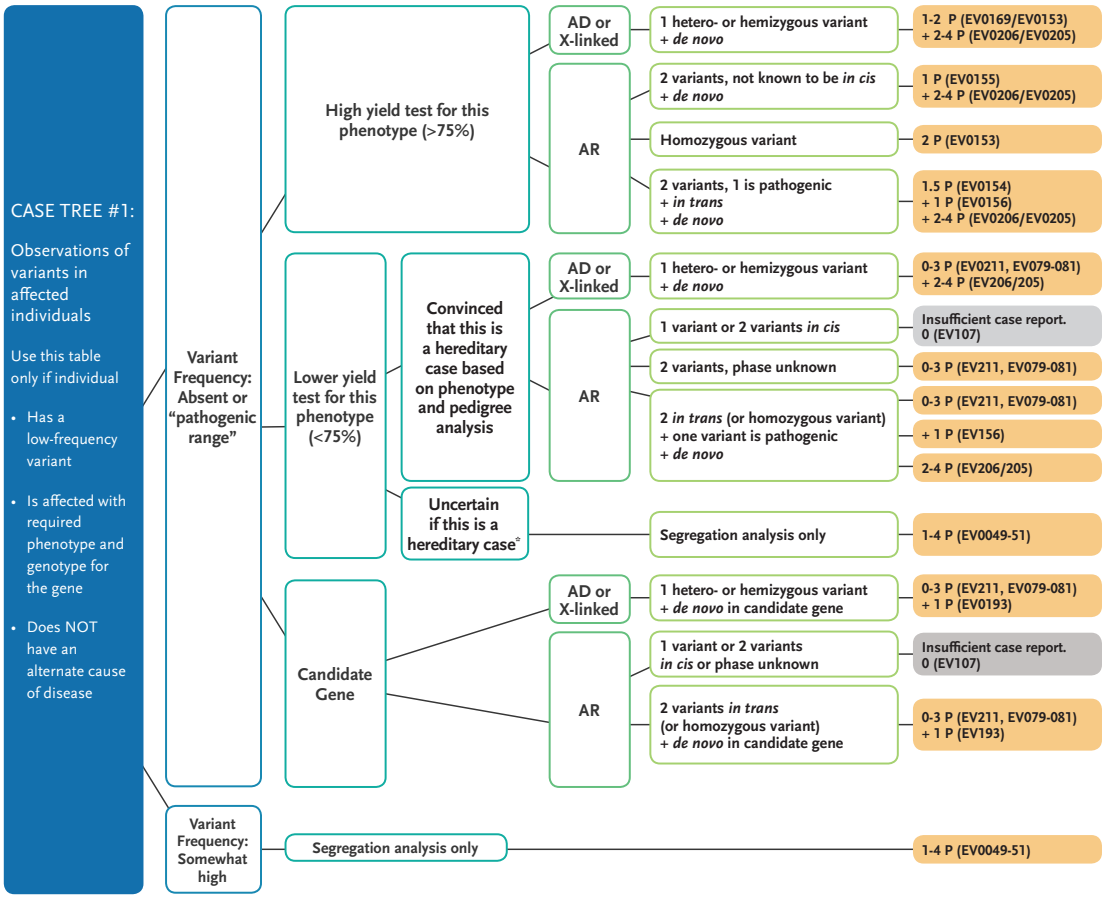
CASE TREE #1:
Observations of variants in affected individuals
Use this table only if individual
• Has a low-frequency variant
• Is affected with required phenotype and genotype for the gene
• Does NOT have an alternate cause of disease
Variant Frequency: Absent or "pathogenic range"
High yield test for this phenotype (>75%)
AD or X-linked
1 hetero- or hemizygous variant + de novo
1-2 P (EV0169/EV0153) + 2-4 P (EV0206/EV0205)
AR
2 variants, not known to be in cis + de novo
1 P (EV0155) + 2-4 P (EV0206/EV0205)
Homozygous variant
2 P (EV0153)
2 variants, 1 is pathogenic + in trans + de novo
1.5 P (EV0154) + 1 P (EV0156) + 2-4 P (EV0206/EV0205)
Lower yield test for this phenotype (<75%)
Convinced that this is a hereditary case based on phenotype and pedigree analysis
AD or X-linked
1 hetero- or hemizygous variant + de novo
0-3 P (EV0211, EV079-081) + 2-4 P (EV206/205)
AR
1 variant or 2 variants in cis
Insufficient case report. 0 (EV107)
2 variants, phase unknown
0-3 P (EV211, EV079-081)
2 in trans (or homozygous variant) + one variant is pathogenic + de novo
0-3 P (EV211, EV079-081)
+ 1 P (EV156)
2-4 P (EV206/205)
Uncertain if this is a hereditary case°
Segregation analysis only
1-4 P (EV0049-51)
Candidate Gene
AD or X-linked
1 hetero- or hemizygous variant + de novo in candidate gene
0-3 P (EV211, EV079-081) + 1 P (EV0193)
AR
1 variant or 2 variants in cis or phase unknown
Insufficient case report. 0 (EV107)
2 variants in trans (or homozygous variant) + de novo in candidate gene
0-3 P (EV211, EV079-081) + 1 P (EV193)
Variant Frequency: Somewhat high
Segregation analysis only
1-4 P (EV0049-51)