Table 6. Association between ultrasound findings and WES-detected variants and diagnostic performance of TVNS and TAS (n = 128)

Genetic outcome	Modality	Sensitivity (%)	Specificity (%)	PPV (%)	NPV (%)	Accuracy (%)	Cohen's kappa
P/LP variants	TVNS	80.0	70.4	33.3	95.0	71.9	0.321
	TAS	45.0	84.3	34.6	89.2	78.1	0.261
VM-related P/LP variants	TVNS	75.0	66.4	18.8	96.2	67.2	0.176
	TAS	50.0	82.8	23.1	94.1	79.7	0.215
VUS	TVNS	57.1	64.9	16.7	92.5	64.1	0.107
	TAS	21.4	79.8	11.5	89.2	73.4	0.009

Abbreviations: P/LP, pathogenic/likely pathogenic; VUS, variant of uncertain significance; PPV, positive predictive value; NPV, negative predictive value.