

Supplementary Material

Population-Based Characterization of PTEN Hamartoma Tumor Syndrome

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Supplementary Table 1: Comparison of Demographic and Cancer-Related Characteristics by Pathogenic Significance

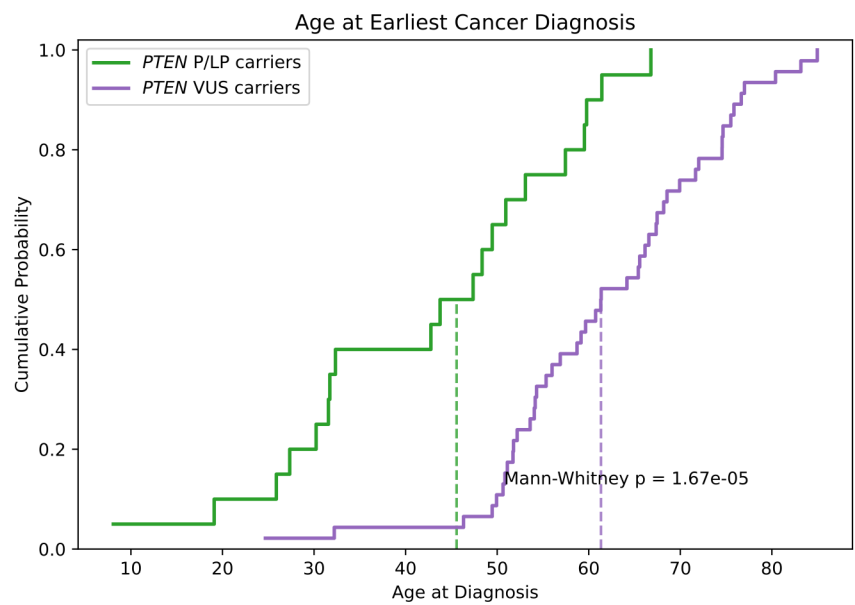
Variable	Carriers of P/LP <i>PTEN</i> Variant N = 55 ¹	Carriers of <i>PTEN</i> Variant of Uncertain Significance N = 342 ²	p-value ³
Sex			>0.9
Female	31 (59.6%)	205 (60.5%)	
Male	21 (40.4%)	134 (39.5%)	
Race			<0.001
American Indian or Alaska Native	0 (0%)	9 (2.6%)	
Asian	*	8 (2.3%)	
Black or African American	*	84 (24.6%)	
Middle Eastern or North African	0 (0%)	2 (0.6%)	
More than one population	*	11 (3.2%)	
Native Hawaiian or Other Pacific Islander	0 (0%)	0 (0%)	
Other/Unspecified	*	61 (17.8%)	
White	36 (65%)	167 (48.8%)	
Cancer Diagnosis			<0.001
No	26 (56.5%)	191 (80.6%)	
Yes	20 (43.5%)	46 (19.4%)	
Age at First Cancer Diagnosis	46 (8 – 67)	61 (25 – 85)	<0.001
¹ Counts of 20 or fewer are suppressed as '*' in compliance with AOU guidelines			
² n (%); Median (Min – Max)			
³ p-values calculated using Kruskal-Wallis test for age and Chi-Squared test for others.			

List of *PTEN* Variants of Unspecified Significance

Variant ID	Consequence	n (% of Cohort)
10-87966902-GTT-G	3 Prime UTR	261 (76.316%)
Other (n<20)	Other (n<20)	259 (75.731%)
10-87966902-GT-G	3 Prime UTR	119 (34.795%)
10-87960984-C-G	Missense	42 (12.281%)
10-87952087-A-G	Intron	41 (11.988%)
10-87966902-G-GT	3 Prime UTR	35 (10.234%)
10-87952066-A-G	Intron	30 (8.772%)

Supplementary Figure 1: Characterization of Participants with *PTEN* Variants of Uncertain Significance (VUS)

a. Age at First Cancer Diagnosis by Variant Group



b. Phenotypic Data of Carriers of *PTEN* VUS

