

Video-Based Germline Genetic Testing for Individuals with Pancreatic Ductal Adenocarcinoma: Before and After the COVID-19 Pandemic

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Introduction

National guidelines recommend universal germline genetic testing (GT) for patients with Pancreatic Ductal Adenocarcinoma (PDAC)¹, but rates of testing remain low². Given the aggressiveness of PDAC, the window of opportunity for GT is short and often overshadowed by treatment initiation. Thus, there is a lack of a model that streamlines GT and makes it available to a wider audience in a rapid fashion. Moreover, in pandemic times, video-based alternatives for medical care are increasingly relevant.

Methodology

We implemented a novel care delivery model in which a 5-minute educational video describing the benefits and risks of GT was shown to PDAC patients. The video was shown in lieu of an initial consult with a genetic counselor. After watching the video, patients could elect to pursue GT and get tested on-site or remotely (at home). Genetic counselors provided post-test counseling by phone. Clinical data were collected through medical records on a cohort of PDAC patients seen at the Gastrointestinal Center-MD Anderson during a 2-year enrollment period, which included the COVID-19 pandemic period.

Relation to Cancer Prevention

Through identification and GT of at-risk family members, these patients can receive PDAC screening earlier and more frequently. Early detection of pancreatic cancer has been linked with better prognosis³.

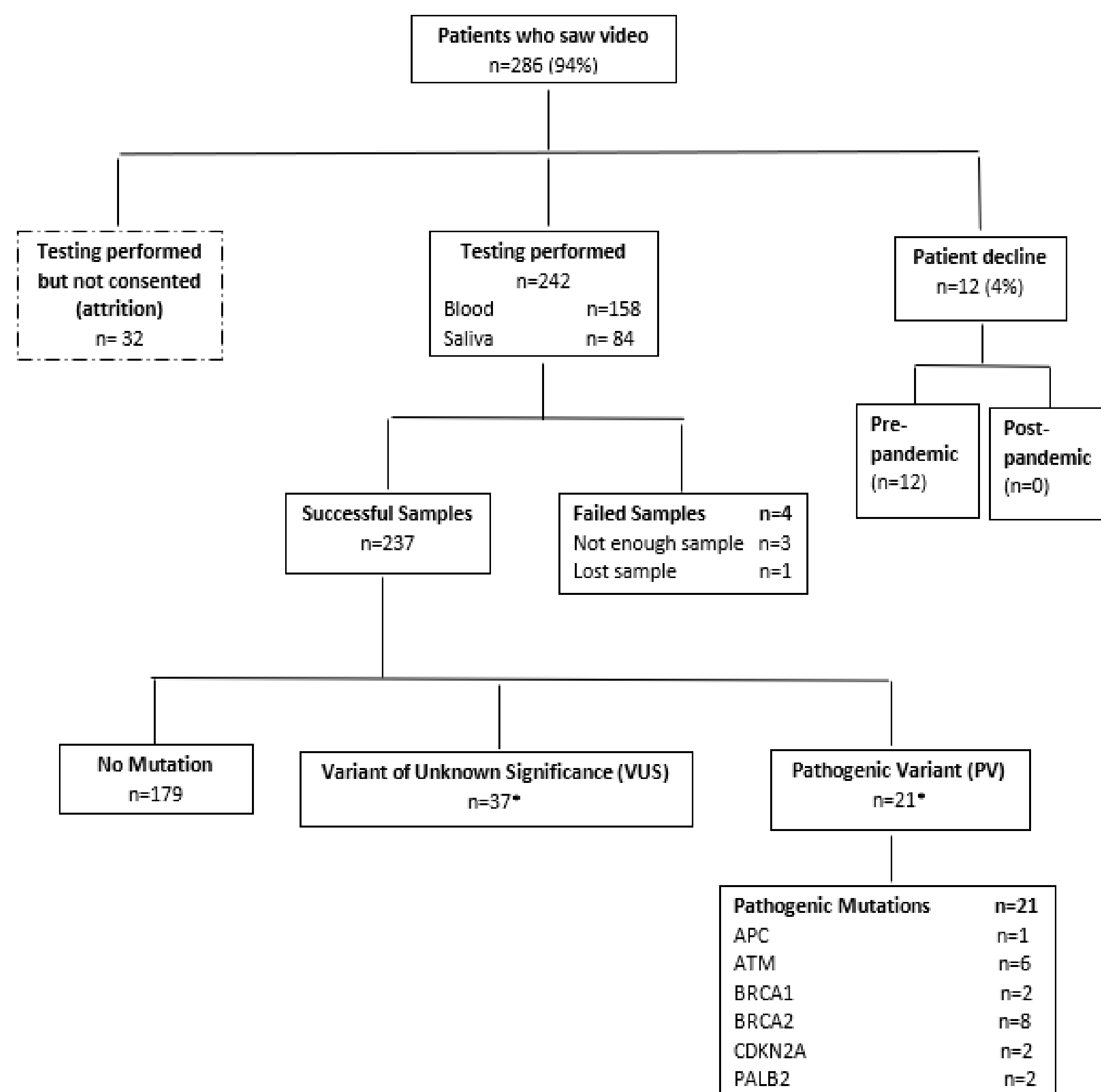


Figure 1: Study Enrollment and Workflow Overview (*1 patient had both a + VUS, 1 patient had 2 VUS)

Variables	PV (n=21)	VUS (n=37)	Negative (n=179)	Total (n=237)	P Value*
Age at Diagnosis					
Median (min-max)	63 (40-86)	65(47-81)	66(38-88)	65(38-88)	0.35/0.70
Gender					
Female	7 (33.3%)	14 (37.8%)	75 (41.9%)	96 (40.5%)	0.45/0.64
Male	14 (66.7%)	23 (62.2%)	104 (58.1%)	141 (59.5%)	
Race					
Caucasian	17(81%)	25(67.5)	147 (82.2%)	189(79.7%)	0.08/0.19
Hispanic	0	2(5.4%)	4 (2.2%)	6 (2.5%)	
African	1(4.75%)	7(19%)	13(7.3%)	21 (8.8%)	
Asian	2(9.5%)	1(2.7%)	9 (5%)	12 (5.1%)	
Other or unknown	1(4.75%)	2(5.4%)	6 (3.3%)	9 (3.9%)	
Personal History of non-PDAC Cancer					
Yes	5(23.8%)	4 (10.8%)	35 (19.5%)	44 (18.5%)	0.67/0.14
No	16(76.2%)	33(89.2%)	144 (80.5%)	193(81.5%)	
Family History of PDAC in FDR					
Yes	7 (33.3%)	2 (5.4%)	7 (4%)	16(6.8%)	0.01/0.7
No	14 (66.7%)	35(94.6%)	172(96%)	221	

* First p-value compares the PV cases to negative cases, and the second p-value compares the VUS cases to negative cases.
PV: Pathogenic Variant; VUS: Variant of Uncertain Significance; PDAC: Pancreatic Ductal Adenocarcinoma; FDR: First Degree Relative

Table 1: Patient Clinical Demographics

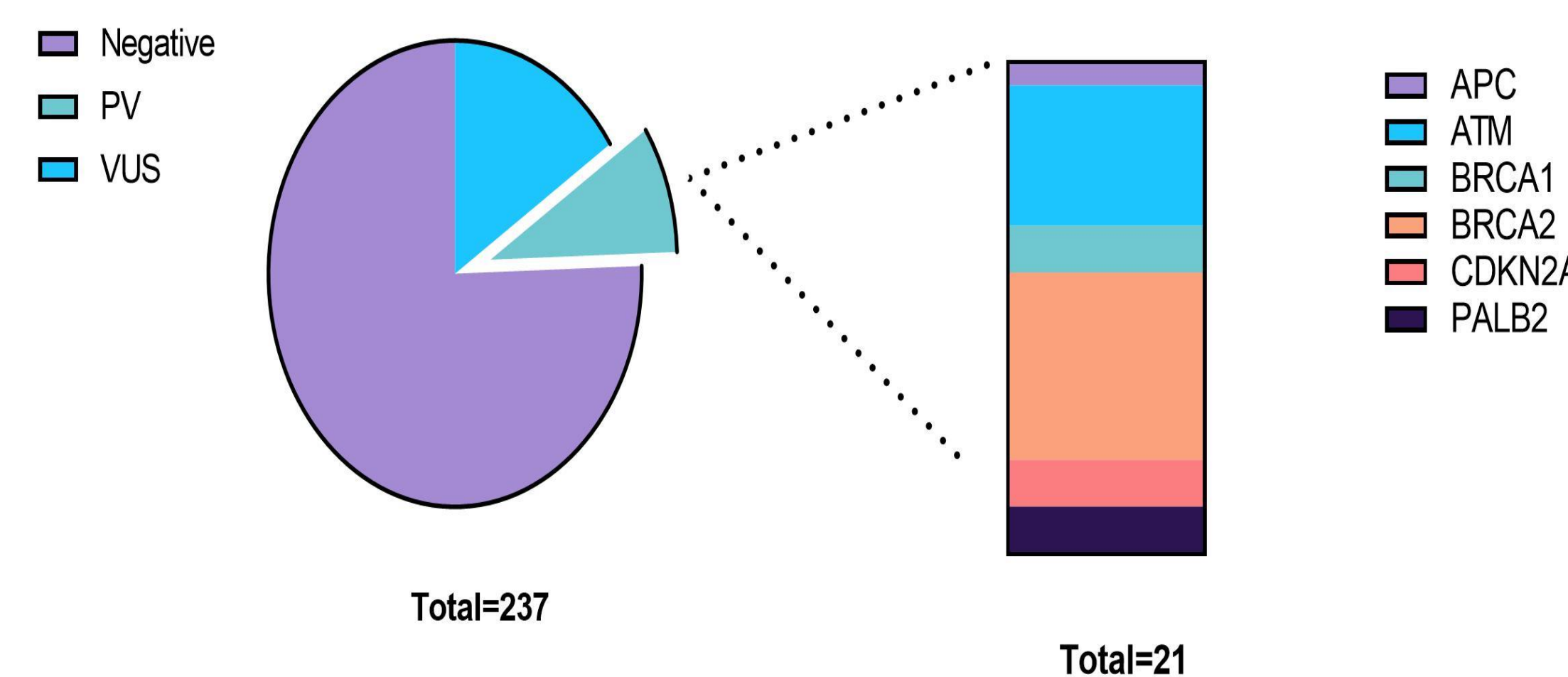


Figure 2: Overview of Positive Results (n=237, 21 total positive patients: 1 APC; 6 ATM; 2 BRCA1; 8 BRCA2; 2 CDKN2A; 2 PALB2)

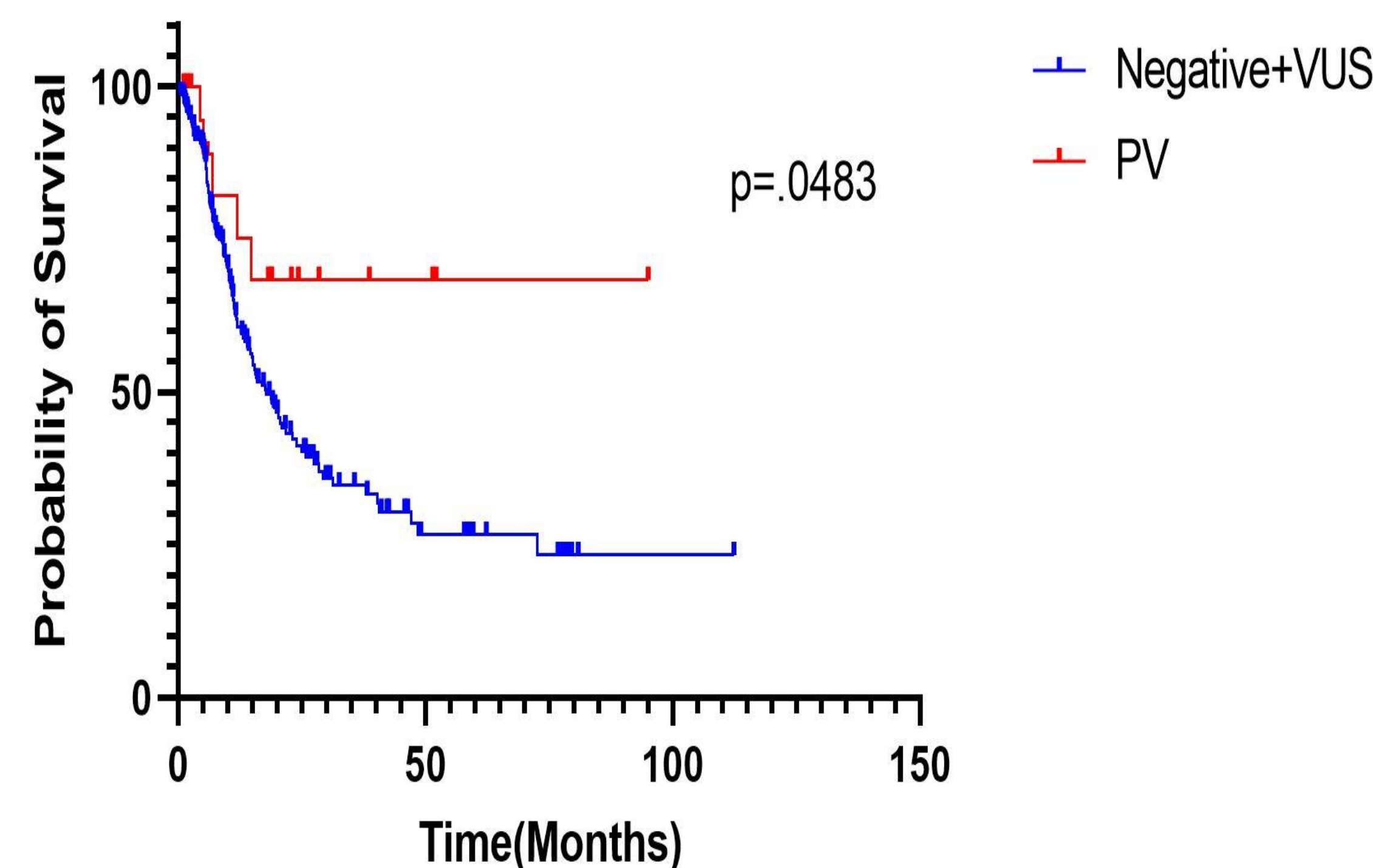


Figure 3: Kaplan-Meier Survival Curve (Overall survival in months among PDAC patients in cohort, stratified by genetic testing result of negative+VUS versus PV; p=.0483)

Results

A total of 285 PDAC patients saw the educational video, after which 12 patients declined GT. These 12 patients were enrolled pre-pandemic, and no patients declined GT post-pandemic. Of the 242 successfully collected samples, 179 patients tested negative, while 37 had a Variant of Uncertain Significance (VUS) and 21 patients had a Pathogenic variant (PV). There was no association between PV and age at diagnosis, gender, race, or personal history of cancer. Family history of PDAC in a first degree relative was associated with having a PV (p=.01). Survival analysis demonstrated that patients with PV tended to live longer (p=.048).

Conclusions

GT can have tremendously beneficial effects, such as qualifying for targeted treatment options and facilitating cancer prevention in probands' at-risk family members. Comparing uptake of GT pre and post-pandemic suggests that patients were more willing to trust information from a video platform, likely due to living in a "virtual" society as a result of the pandemic. We suggest an approach in which every PDAC patient is shown a genetics educational video and given the option of GT and post-result counseling, greatly reducing the burden on genetic counselors. Further investigation is needed to explore the feasibility of a fully remote GT model (in which the video and saliva collection kit are sent directly to the patient's residence)

Responsible Conduct of Research

This study was approved by the University of Texas MD Anderson Cancer Center institutional review board. The MD Anderson PI (Dr. McAllister) was responsible for maintaining documents and approvals for all modifications in the protocol. We worked to maximize security of patient data.

Acknowledgements

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