

Background

- Transthyretin amyloidosis (ATTR) is a progressive, life-limiting disease, often causing cardiomyopathy (ATTR-CM) which leads to significant morbidity and mortality
- Prognosis and treatment of ATTR depend on wild-type (wtATTR) or hereditary (hATTR), determined by genetic testing
- Fewer than 10% of ATTR patients undergo genetic testing, often due to lack of access to genetics services
- Telehealth genetic counseling model to improve access to genetic counseling and testing across the U.S. and Canada

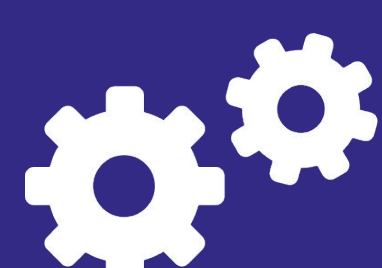
Methods



Retrospective chart review of patients with ATTR-CM seen by Genome Medical from August 2019 to May 2024

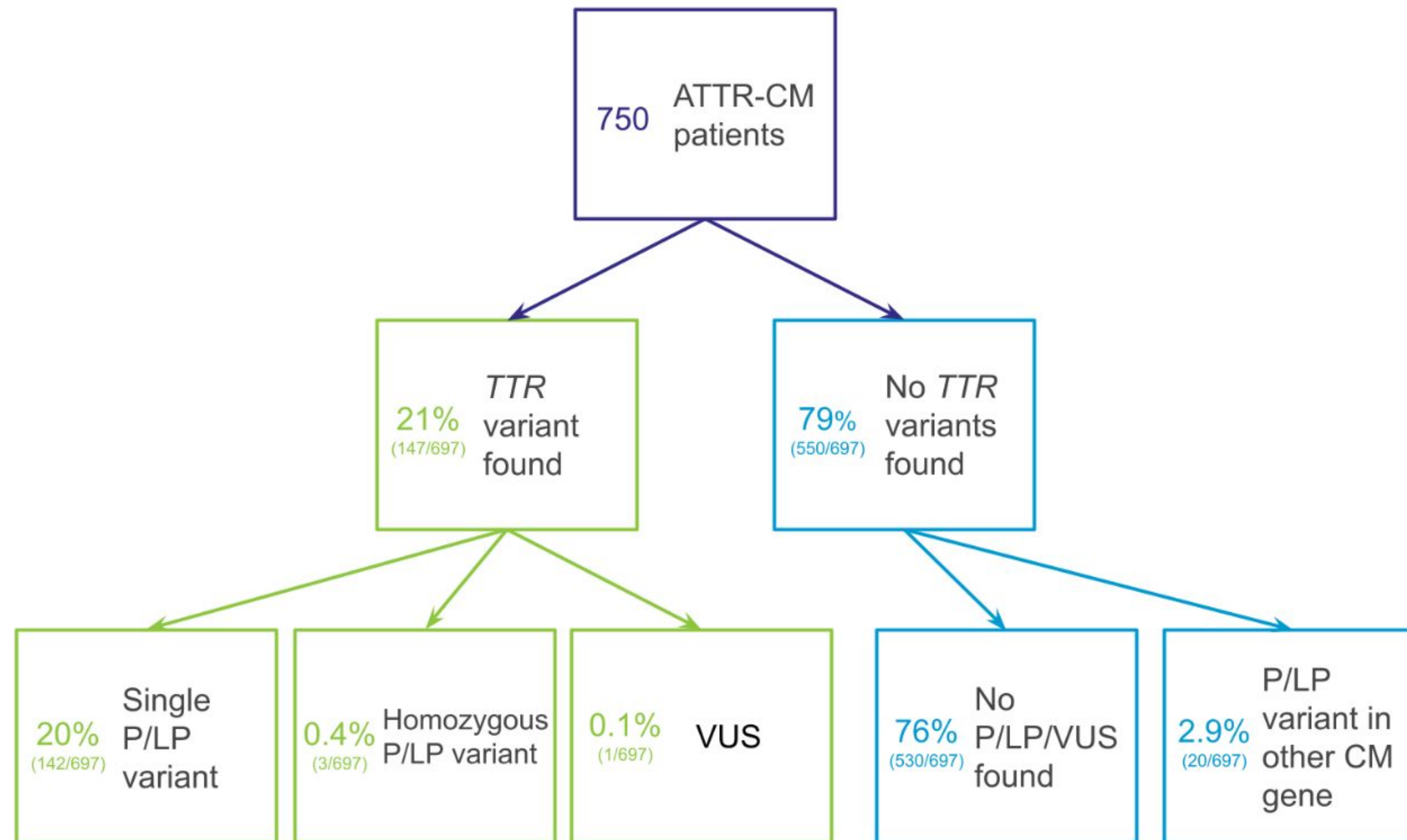


Patients included if there was reported evidence of a clinical diagnosis of ATTR-CM



Subgroups compared using t-tests or chi-squared tests

A nationwide-telehealth program enabled genetic counseling and genetic testing for 750 ATTR-CM patients over five years



Results

N	Mean age	Sex
750 Patients	75.3y SD 11.2	75% Male



41 states & 5 provinces
Top locations: Georgia (13.1%), Ontario (9.5%), Florida (8.3%), Tennessee (6.8%).

	wtATTR	hATTR
Mean age p<0.001	75.9y	72.8y

Sex p=0.003	21.5% Female	32.3% Female
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91% (132/145) Of hATTR patients had p.Val142Ile

2.1% (3/145) Were homozygous p.Val142Ile



P/LP variants also found in: *TTN, DSP, TNNT2, SDHA, SCN5A, PLN, MYH7, LMNA, GLA, FKTN, FKRP, and CACNA1C*

Conclusion

- A nationwide telehealth genetic counseling program enabled the diagnosis of hATTR in one-fifth of patients
- Variants found in other cardiomyopathy genes underscore importance of comprehensive genetic testing in this population

Nationwide Telehealth Genetic Counseling To Increase Access In Transthyretin Amyloidosis: A 5-Year Retrospective Review

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