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UNSEEN AND UNHEARD Addressing Barriers in Diagnostic and Treatment Inequalities for p.V142I hATTR-CM in Black Communities

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Contents

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Authors

Christine Chiti, TTR Specialist Nurse, **National Amyloidosis Centre, Royal Free London NHS Foundation Trust, Pond Street, London, NW3 2QG**
Kate Taylor, Managing Director, **Amyloidosis UK, Hackamor House, Grove Lane, Redlynch, Salisbury, SP5 2NR**
Seema Patel, Specialty Care Medical Director, **Pfizer Ltd UK, Walton Oaks, Dorking Road, Tadworth, Surrey, KT20 7NS**
Ben Laryea, Patient author, **London, UK**; Trustee, **Amyloidosis UK, Hackamor House, Grove Lane, Redlynch, Salisbury, SP5 2NR**
Vincent Nicholas, Trustee, Treasurer & Founding Member, **Amyloidosis UK, Hackamor House, Grove Lane, Redlynch, Salisbury, SP5 2NR**
Dr Jason N Dungu, Consultant Cardiologist, **Essex Cardiothoracic Centre, Nether Mayne, Basildon, SS16 5NL**; Honorary Senior Clinical Lecturer, **Anglia Ruskin University**

Unseen and Unheard: Addressing Barriers in Diagnostic and Treatment Inequalities for p.V142I hATTR-CM in Black Communities

Christine Chiti, Kate Taylor, Seema Patel, Ben Laryea, Vincent Nicholas, Dr Jason N Dungu

Key words

cardiomyopathy, p.V142I, Transthyretin amyloidosis (ATTR)

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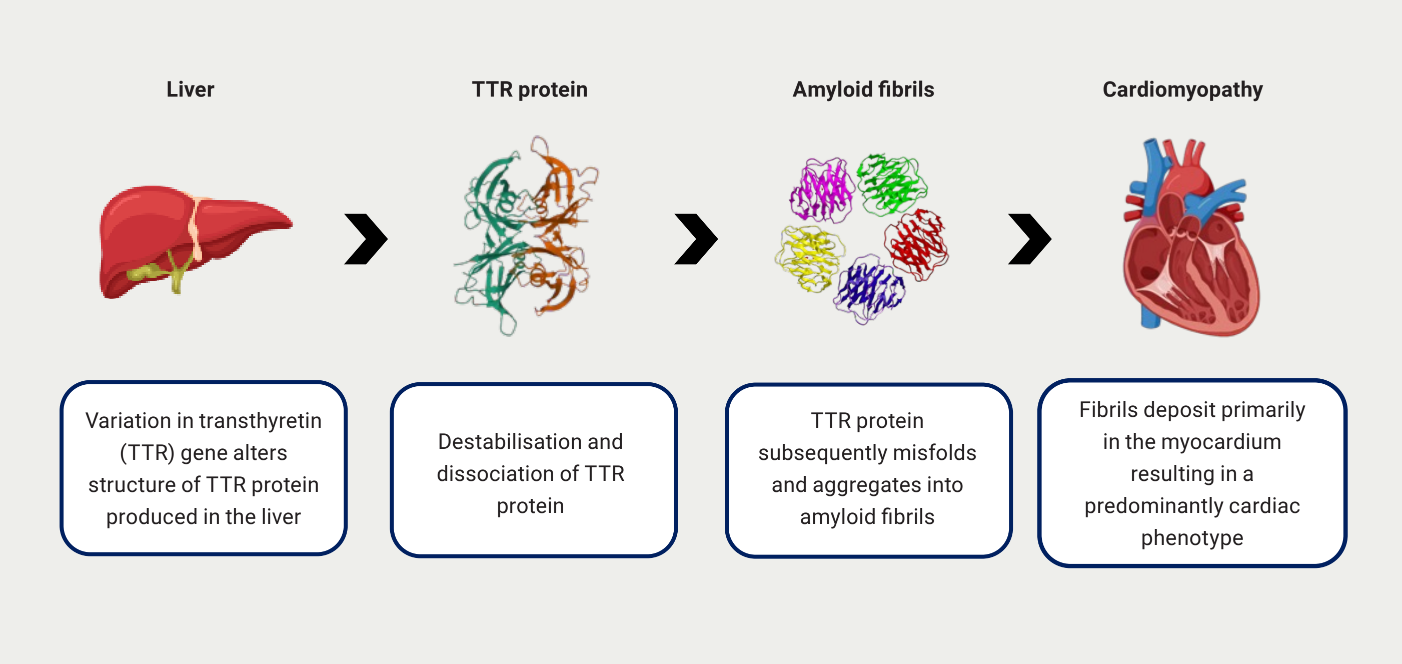
Transthyretin Amyloidosis (ATTR) is a progressive, systemic and life-shortening condition that remains widely under-recognised and frequently misdiagnosed.[1] It occurs in two distinct forms: **wild-type (wtATTR), which is typically age-related**, and **hereditary (hATTR), which is caused by pathogenic variants in the transthyretin (TTR) gene**.[1,2] The p.V142I (formerly V122I) variant of hATTR primarily presents as cardiomyopathy (CM), but can also result in other clinical manifestations such as polyneuropathy.[3] The p.V142I variant is most commonly found in individuals with West African and African-Caribbean ancestry.[4–7]

Diagnosis of ATTR-CM can take years - by which time irreversible heart failure may have already occurred.[2,8] hATTR-CM severely impairs quality of life, and median survival in advanced stages is typically only two to three years without timely intervention.[2]

The Genetic Link to Heart Failure

Figure 1. Underlying Pathology of hATTR Caused by the p.V142I Variant[9,10]



Variation in transthyretin (TTR) gene alters structure of TTR protein produced in the liver

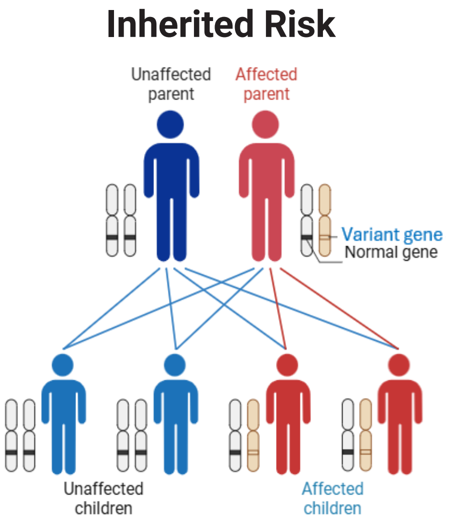
Destabilisation and dissociation of TTR protein

TTR protein subsequently misfolds and aggregates into amyloid fibrils

Fibrils deposit primarily in the myocardium resulting in a predominantly cardiac phenotype

Individuals with p.V142I typically present with heart failure (HF), restrictive cardiomyopathy, and arrhythmias which can ultimately lead to death.[5,11,12]

The hereditary form follows an autosomal dominant inheritance pattern. Only one parent needs to pass on the variant for a child to carry it.[13–15] Genetic testing is critical - not only to confirm diagnosis and identify family members who carry the variant, but also to enable early detection and timely intervention for improved outcomes.[16,17]



Prevalence of the p.V142I Variant

The p.V142I variant is much more prevalent in populations of African descent than globally and exposes a significant health disparity.[4,7,18]

Population genetics studies supports the origin of p.V142I in West African cohorts and its subsequent introduction into populations across the Americas, the United Kingdom, and Caribbean during periods of migration.[6]

Figure 2. Prevalence of p.V142I Among Subjects of African Descent[6]

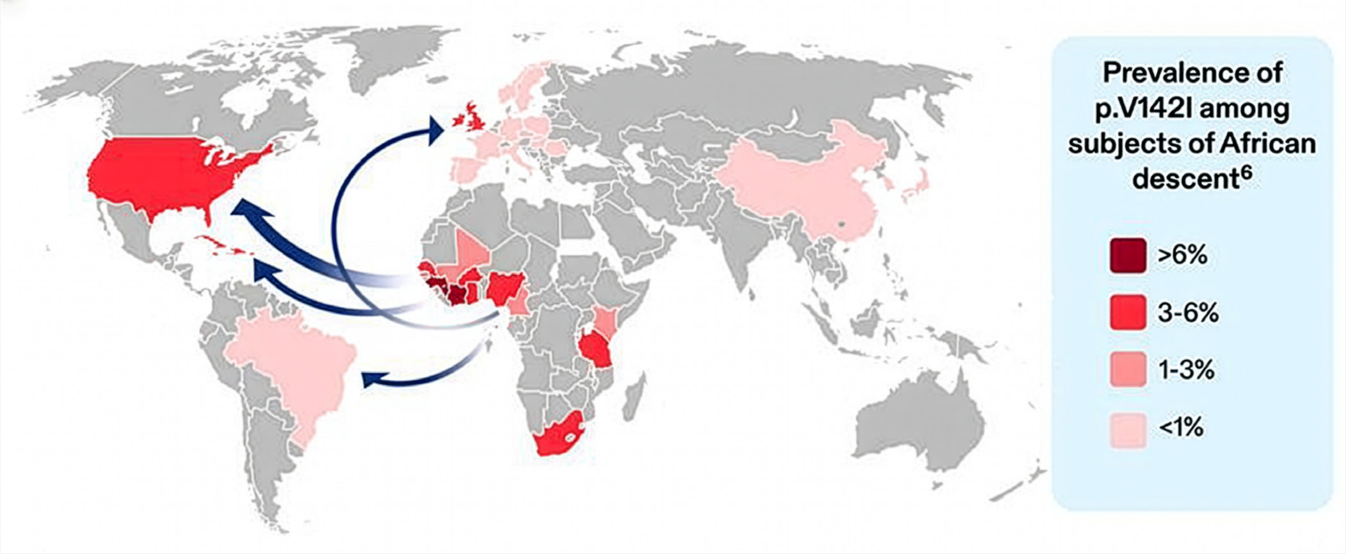


Figure adapted from: Chandrashekar, P., *et al*. Prevalence and Outcomes of p.Val142Ile TTR Amyloidosis Cardiomyopathy: A Systematic Review. *Circ Genom Precis Med*, 2021. 14(5): p. e003356.[6]

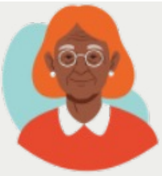
Under-recognition and Systemic Barriers

Underdiagnosis in p.V142I Carriers[3]

A study conducted using the UK Biobank analysed genetic and clinical data from 243 individuals carrying the p.V142I variant.

This study provides stark evidence of underdiagnosis in this patient population.

243 p.V142I carriers had an enrichment of common hATTR amyloidosis manifestations.



2 out of **243** were formally diagnosed with amyloidosis, highlighting the extent of under-recognition.

A UK study showed at the time of diagnosis, the p.V142I group of patients had significantly worse LVEF and functional status than the rest of the cohort. Survival was also reduced in the p.V142I group compared to the rest of the subjects.[19]

This could indicate that the p.V142I variant may present a more aggressive phenotype.[19]

A US study also showed that black patients, the majority of whom had hATTR-CM caused by p.V142I (102/129, 79%), often presented with more severe disease at onset and poorer survival than their white counterparts who primarily had wild type ATTR-CM (96/135 71.1%).[20]

Access Challenges and Diagnostic Delays

Varied access to diagnostics, the lack of formal amyloidosis care pathways, and limited specialist resources create significant inequities in diagnosis and care across the UK.

Historically, there was only one specialised centre - the National Amyloidosis Centre (NAC) in London - although the network is in the process of being expanded with two additional centres in Liverpool and Birmingham.[21] Despite this progress, many regions remain underserved.

Delayed Diagnosis in hATTR-CM

A prospective protocolised clinical follow-up programme at the NAC of 1034 patients with ATTR-CM included 323 patients with hATTR, 205 of whom carried the p.V142I variant. In a subset of patients with 3 years of hospital usage records prior to diagnosis of ATTR-CM, time from first presentation with cardiac symptoms to diagnosis was determined.[19]

Average diagnostic delay

25 Months

Median time to diagnosis after first presentation at hospital (inpatient, outpatient or emergency department) with cardiac symptoms.

Maximum diagnostic delay

5 Years

For some patients.

These delays mean that many patients are only diagnosed when their disease is advanced, with irreversible and extensive cardiac damage, severely limiting their survival prospects.[3,8,22,23] Diagnostic delays are also seen for wild-type patients.[19] Another UK study reported that healthcare costs for ATTR-CM patients are high from symptom onset, rising with disease progression and linked to delayed diagnosis.[24]

Patient Advisory Board

Objectives

To understand the relevance of these systemic challenges in the UK, an advisory board was convened with four advisors from the UK, three with hATTR-CM with the p.V142I variant and one caregiver of a relative living with hATTR-CM with the p.V142I variant. The voices of these individuals are central to this white paper, which calls for urgent, collaborative action. Our aim is to improve long-term outcomes for affected individuals and families.

The Patient Voices: Summary of Diagnostic Journeys

Participant 1

Participant 1 experienced an unexplained decline in their physical capacity, with ankle swelling and unexplained weight loss, describing themselves as looking “quite skeletal”. After multiple investigations and differential diagnoses, NHS referral delays of 3–4 months led them to seek a private consultation, reducing diagnosis time to 4–5 weeks.

“I feel quite lucky to have received a diagnosis within a relatively short timeframe. I attribute this to seeing a cardiologist with experience in the condition. For some individuals, diagnosis can take 5 years.”

Participants 2 and 4

Participants 2 and 4 were diagnosed following an acute cerebrovascular event (stroke), that prompted cardiac tests. One of them went privately to receive their diagnosis.

Participant 3 (Caregiver)

Participant 3 became aware of hATTR-CM due to their mother’s recent diagnosis which took 3 to 4 months from symptom onset.

Patient Experience: Pre-diagnosis

Closing the Awareness Gap: hATTR-CM in Communities and Clinical Practice

Before diagnosis, none of the participants had heard of hATTR, and described low awareness in primary care. They felt the speed of diagnosis depended heavily on whether they reached a clinician familiar with the condition.

> “I was amazed at how little HCPs know about the condition. I remember a time when I had to explain my diagnosis to the GP, who admitted they would need to Google it”

For two participants, the diagnosis uncovered a hidden legacy of illness within their families, revealing that relatives who had died from heart conditions were now suspected to have had hATTR.

Participants unanimously called for hATTR-CM (p.V142I) to be treated with the same urgency and visibility as sickle cell disease and prostate cancer, both of which have benefitted from prominent, targeted public awareness campaigns in the black community. **They stressed that “awareness efforts must begin within communities.”**

Building Trust Through Representation: Overcoming Barriers in Health Communication

Participants expressed frustration with existing educational materials, which lacked cultural representation and were difficult for laypeople to understand. They emphasised that mistrust of healthcare professionals (HCPs) within the African and Afro-Caribbean communities was rooted in a “painful history of unethical medical practices” including people being “used as guinea pigs” in clinical trials.

Equity Gaps in ATTR-CM Clinical Trials

Although several randomised trials have focused on ATTR, they have rarely included African countries as recruitment centres or set explicit goals for enrolling Black patients or those with the p.V142I variant despite data suggesting these cohorts may have worse outcomes, therefore proactive strategies are needed to address these disparities.[20,26]

Because of this mistrust, they noted that people within the community are more likely to respond to health information delivered by individuals of the same ethnicity. Such concerns align with evidence showing that worse cardiovascular outcomes in Black patients are likely multifactorial, driven in part by mistrust in healthcare providers.[26]

Patient Experiences: During Diagnosis

The Emotional Impact of Diagnosis and Hereditary Guilt

Participants described diagnosis as emotionally challenging, with limited guidance at a life-changing moment.

Having a loved one present at the time of diagnosis helped with remembering details and asking key questions.

Learning hATTR is hereditary was described as particularly difficult, raising concerns about the implications for their children and other relatives. Some participants reported not receiving genetic counselling, and described feeling isolated when deciding how to start family conversations.

Patient Experiences: Post Diagnosis

Patient Advocacy Groups (PAGs) Awareness and Impact

One patient shared how joining a PAG (Amyloidosis UK) provided practical guidance and emotional support, helping them feel "less alone." They valued hearing from others with lived experience. The rest of the participants, however, were unaware of PAGs. It was suggested that **information about PAGs should be included with appointment letters** so patients and families can access support early. **Healthcare practitioners who implement patient advocacy can achieve greater patient satisfaction and reduce health disparities across all communities they serve.**[27]

Caregiver testimonial

"During the numerous consultations my mother had attended, PAGs had not been actively promoted. I felt surprised, disappointed, and angry about it. Awareness about PAGs via posters are not enough and people walk past posters. PAGs should be discussed more proactively during appointments."

Barriers to Information and genetic testing pathways

Participants described confusion about the genetic testing pathway, including uncertainty about who to contact and how relatives can access testing. While asymptomatic carriers are increasingly identified via genotyping, participants noted the need for clearer guidance on screening and management. Evidence suggests systematic screening can improve early identification and outcomes, reinforcing the need for formalised pathways and guidance.[28]

Recommendations and Clear Call to Action: A Mandate for Systemic Reform

This White Paper calls for urgent, collaborative action to embed equity and efficiency into the care pathway through three strategic pillars.

Pillar 1. Educate Public and HCPs to Improve Recognition and Empower Patients

Launch targeted community campaigns
Use proven channels - radio, community events - to reach at-risk populations, inspired by successful sickle cell and prostate cancer initiatives.

Strengthen HCP education and referral
Equip GPs, cardiologists and multi-disciplinary teams with the knowledge to recognise hATTR and refer patients swiftly to specialist care.

Guide reliable information access
Clinicians should proactively guide patients to reliable hATTR resources, while PAGs and trusted organisations improve their online visibility.

Pillar 2. Streamline Genetic Testing and Expand Support

Expand specialist centres
The NHS should consider establishing wider networks of regional amyloidosis centres to reduce travel barriers, under-diagnosis and increase equity of care.

Clarify genetic testing pathways
The UK Amyloidosis Network should consider providing clear, simple guidance on genetic testing and screening, including referral routes and eligibility.

Increase psychological and genetic counselling
The NHS and clinicians should consider offering immediate access to specialised support following diagnosis to address emotional impact and hereditary risk.

Pillar 3. Build Trust Through Diversity and Patient Leadership

Mandate culturally tailored education
Develop accessible, visually engaging materials that reflect African and Afro-Caribbean communities in the UK.

Empower patient advocates and diversify HCPs
Involve patients with lived experience and increase diversity among healthcare professionals to build trust and address ethical concerns.

Conclusion

Hereditary ATTR (hATTR) caused by the p.V142I variant is a significant under-recognised health inequity, especially for Black African and Afro-Caribbean communities. Systemic barriers and diagnostic delays compromise patient outcomes and quality of life. This white paper amplifies patient and caregiver voices, calling for urgent, collaborative action from the NHS, policymakers, advocacy groups, and the pharmaceutical industry. Implementing the recommended strategic pillars, including raising awareness, de-centralising care, streamlining genetic testing pathways, and integrating holistic support, will help drive meaningful reform, embed equity, and improve long-term outcomes for individuals and families affected by hATTR

Conflict of interest

SP is an employee of Pfizer Ltd (UK). JND has received speaker fees and/or consultancy fees from Pfizer, Bristol Myers Squibb, Alnylam and Bayer. CC reports relationships with Alnylam, AstraZeneca, Bayer, Springer Healthcare and CVRM Connect; CC is employed as a TTR Specialist Nurse at the National Amyloidosis Centre, Royal Free Hospital, London. BL is a patient and a Trustee of Amyloidosis UK. VN declares no competing interests; VN is a Trustee of Amyloidosis UK. KT: Managing Director at Amyloidosis UK. The non-governmental organisation receives support from competing companies of the pharmaceutical industry (BridgeBio, AstraZeneca, Alnylam, Intellia) without proximity, dominance or dependence on individual companies.

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Christine Chiti
TTR Specialist Nurse
National Amyloidosis Centre, Royal Free London NHS Foundation Trust, Pond Street, London, NW3 2QG

Kate Taylor
Managing Director
Amyloidosis UK, Hackamor House, Grove Lane, Redlynch, Salisbury, SP5 2NR

Seema Patel
Specialty Care Medical Director
Pfizer Ltd UK, Walton Oaks, Dorking Road, Tadworth, Surrey, KT20 7NS

Ben Laryea
Patient author
London, UK
Trustee
Amyloidosis UK, Hackamor House, Grove Lane, Redlynch, Salisbury, SP5 2NR

Vincent Nicholas
Trustee, Treasurer & Founding Member
Amyloidosis UK, Hackamor House, Grove Lane, Redlynch, Salisbury, SP5 2NR

Dr Jason N Dungu
Consultant Cardiologist
Essex Cardiothoracic Centre, Nether Mayne, Basildon SS16 5NL
Honorary Senior Clinical Lecturer
Anglia Ruskin University

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