Indian Pacing and Electrophysiology Journal xxx (xxxx) xxx

Contents lists available at ScienceDirect

Indian Pacing and Electrophysiology Journal

journal homepage: www.elsevier.com/locate/IPEJ

Review Article

India-specific cardiogenetic aspects: Focus on cardiomyopathies and inherited arrhythmia syndromes

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A B S T R A C T

India is a land of diversity with its deep evolutionary history, demographic shifts, archaic and recent gene flow events and a high level of endogamy resulting in a unique genetic structure and variation. Yet, very little knowledge exists about population-specific and disease susceptibility variants in the country as Indian populations remain underrepresented in genomic studies. This review article, the final in the Cardiogenetic series, aims to highlight the India-specific knowledge on cardiomyopathies and inherited arrhythmia syndromes, enumerate the best practices and future directions, and emphasize the need for a nationwide database for cardiogenetic diseases. The genotype-phenotype correlations for HCM, DCM, ACM, LQTS, CPVT, sodium channelopathies and sudden cardiac death are outlined while touching upon the growing need for incorporating phenotype-guided genetic testing modalities in the management protocol of affected individuals and their families. The already functioning multidisciplinary cardiogenetic centres with dedicated healthcare teams comprised of cardiologists, electrophysiologists, geneticists, genetic counsellors and specialized nurses could be used as a model to scale-up and establish further facilities across the country and fill the existing gap in meting out comprehensive care to patients and their families.

1. Introduction

Genetics, said to be the defining field of the 21st century, is revolutionizing healthcare in general and the practice of cardiology in particular across the globe. The recent international consensus document on the state of genetic testing in cardiac diseases is a culmination of three decades of dedicated work and provides comprehensive recommendations to practitioners managing individuals with genetic diseases of the heart [1]. Envisioned to fulfil an unmet need in India, the current special series on Cardiogenetics has delved upon the foundational aspects of genetics and its application in cardiology, the genetics of inherited arrhythmia syndromes, cardiomyopathies and sudden cardiac death (SCD) and the cardiovascular screening of athletes [2–6]. This review article, the final in the series, aims to highlight the India-specific knowledge on cardiomyopathies and inherited arrhythmia syndromes, enumerate the best practices and future directions, and emphasize the need for a nationwide database for cardiogenetic diseases.

1.1. Unique genetic tapestry

Known as the land of diversity for its rich cultural and geographic variety, India is also a one-of-a-kind genetic tapestry with its 1.4 billion population belonging to approximately 5000 anthropologically well-defined ethno-linguistic and religious communities. Deep evolutionary history, demographic shifts and archaic and recent gene flow events together with a high level of endogamy have resulted in a unique genetic structure and variation in the Indian subcontinent. Yet, very little knowledge exists about population-specific and disease susceptibility variants in the country as Indian populations remain underrepresented in genomic studies [7,8].

1.2. Population-specific datasets and multidisciplinary clinics

The ongoing GenomeAsia100K and the Genome India Projects are efforts in the right direction to create our own reference databases [9,10]. In order to better understand cardiogenetic diseases of our population and their genotype-phenotype correlations, we need a nationwide registry as these conditions are not only rare but are also shaped by the genetic landscape of the different ethnicities within India. A few academic institutions and tertiary cardiac care hospitals across the country have succeeded in establishing multidisciplinary cardiogenetic units with dedicated healthcare teams comprised of cardiologists, electrophysiologists, geneticists, genetic counsellors and specialized nurses to manage patients and families with inherited heart diseases [11]. Additionally, there are physicians and allied professionals with expertise in this domain contributing to the knowledge pool. Recently, a consortium

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https://doi.org/10.1016/j.ipej.2026.02.001
Received 14 December 2025; Received in revised form 24 December 2025; Accepted 1 February 2026
Available online 2 February 2026

Please cite this article as: Priya Chockalingam, Rajaram Anantharaman, *Indian Pacing and Electrophysiology Journal*, https://doi.org/10.1016/j.ipej.2026.02.001

of experts have published a position statement on the genetic testing of cardiomyopathies, elaborating on various aspects pertinent to patients and practitioners in India [12]. However, the lack of multicentre collaboration and a nationwide registry is limiting the growth of the field in the country [13].

2. Inherited cardiomyopathies

Being a country heavily burdened with coronary artery- and cardiometabolic disease-related cardiomyopathies, there has been a paucity of focus on primary cardiomyopathies and more specifically on inherited or genetic cardiomyopathies in India. A lack of awareness about the clinical applications of genetic testing, limited access to testing laboratories and genetic counsellors and a deficit of physicians trained in interpreting genetic test reports have all contributed to the widening gap between the scientific developments and the actual day-to-day practice. Nevertheless, just as cutting-edge heart failure management including heart transplantation has become mainstream in India [14,15], we strongly believe that the field of cardiogenetics is set to advance rapidly in the next decade.

2.1. Hypertrophic cardiomyopathy (HCM)

HCM, the commonest inherited heart disease with a prevalence of approximately 1 in 500, is a leading cause of SCD in apparently healthy, young and active individuals. The earliest published report on the genetics of cardiomyopathy in India details the detection of a then novel variant R712L in the *MYH7* gene in a HCM family [16]. This variant is currently known to cause a severe form of HCM in which the actin-gliding motility of R712L-myosin is inhibited and this inhibition seems to be rescued by the heart failure drug omecamtiv mecarbil [17].

MYBPC3$^{\Delta 25}$, the intronic deletion common in the South Asian population was initially reported in two Indian HCM families in 2003 and even back then was considered to be a "modifying gene", which per se did not or only rarely caused HCM but which enhanced the phenotype of a pathogenic variant responsible for disease [18]. Subsequent studies identified the variant's propensity to cause heart failure in individuals of Indian subcontinental ancestry [19]. A small subset of individuals were found to bear both the deletion and a rare pathogenic variant c.1224-52G > A in the *MYBPC3* gene, the latter variant explaining the disease risk [20]. Further research into the deletion variant revealed that it is associated with left ventricular hypercontraction under stress conditions with evidence of diastolic impairment [21]. In our own experience, we have found this variant to be aggressive, initially manifesting as biventricular HCM in adolescence in a South Indian female patient and later progressing to severe heart failure warranting heart transplantation by her 30 years of age [22].

Bashyam et al. conducted one of the earliest genotype-phenotype analysis by performing polymerase chain reaction (PCR) in 55 HCM patients detecting *MYBPC3* variants in 12, *MYH7* variants in 5 and double heterozygosity in 2 patients. Four novel variants in *MYBPC3* namely c.456delC, c.2128G > A (p.E710K), c.3641G > A (p.W1214X), and c.3656T > C (p.L1219P) and one in *MYH7* namely c.965C > T (p.S322F) were identified [23]. A more recent PCR study in 115 HCM patients had detected 34 single nucleotide variations in *MYBPC3*, of which 19 were novel. The key findings of this study were (1) late onset of symptoms in the third decade of life (2) milder forms of disease progression (3) lower penetrance and (4) dose-dependent effects in patients with compound variations in *MYBPC3* [24].

Three recent studies have reported their whole exome sequencing (WES) results in 80 (Delhi study), 200 (Kerala study) and 335 (multicentre South India study) HCM patients respectively [25–27]. They have provided the following invaluable insights on HCM in Indian patients: (1) *MYBPC3* and *MYH7* are the predominant genes with a high frequency of missense variants (2) the specific variants identified reflect both established causal mutations and novel or population-specific variants requiring further validation (3) gene-positivity was associated with younger age-at-diagnosis, family history of HCM, asymmetric hypertrophic pattern, severe mitral regurgitation and late gadolinium enhancement detected fibrosis (4) ventricular tachycardia and premature cardiovascular death were predictive of *MYBPC3* or loss-of-function variants whereas left atrial and interventricular septal dimensions were associated with *MYH7* variants (5) the yield of genetic testing was 36% in South Asian Indian patients with primary HCM, compound heterozygosity being present in 11% (6) *ACTC1*, *MYL3*, and *TNNC1* genes are underrepresented and *MYH6* gene variants are higher in our population compared to global HCM cohorts.

A 2023 scoping review of sarcomeric gene variants among Indian HCM patients assessed 54 P/LP (pathogenic/likely pathogenic) variants from *MYBPC3*, *MYH7*, *TNNT2*, *TNNI3* and *TPM1* to conclude that carriers of single homozygous, de novo and digenic variants were observed to be associated with severe phenotypes compared to carriers of single heterozygous variant [28]. While Rao et al. [27] have analysed their results based on the recently reappraised HCM genes [29], further large studies on Indian HCM patients are required to develop a population-specific reappraisal.

2.2. Dilated cardiomyopathy (DCM)

DCM is a highly heterogeneous condition characterized by left ventricular or biventricular dilatation, systolic dysfunction and heart failure in the absence of abnormal loading conditions and is associated with a complex and diverse genetic architecture currently spanning 19 genes with high-level evidence of causation [30]. The most notable is *TTN*, an enormous scaffolding protein of the sarcomere, which contributes the most cases of DCM followed by *LMNA*, encoding a protein of the inner nuclear membrane that exhibits striking pleiotropic effects in skeletal muscle, adipose, and other tissues.

Whilst the literature on the genetic underpinnings of Indian DCM patients is limited, Rani et al. first identified that the R144W variant found to be within the tropomyosin-binding domain of the *TNNT2* gene was causative of DCM and SCD [31] and later went on to detect numerous novel, unique, and rare variants in the *MYH7* gene that were exclusive in our DCM population [32]. A large multicentre study involving 513 DCM cases from various ethnically distinct cohorts identified functional *RAF1* variants in young patients of South Indian, North Indian and Japanese origin, documenting its prevalence as ~9% in childhood-onset DCM cases in these three cohorts [33].

A recent study on 20 DCM patients from Delhi with positive family history, defined as ≥2 first degree relatives with DCM, or SCD in them before 35 years of age, reported a high yield (70%) of gene positivity. *TTN* gene variants were the most common, manifesting in younger (<50 years) population with a male predilection and with a more severely depressed left ventricular function. *LAMA* and *FLNC* variants were associated with younger population, female predilection, severely depressed left ventricular function with more dilatation of left ventricle and a family history of SCD suggesting increased susceptibility to arrhythmias [34].

We now know that arrhythmias in the form of atrial fibrillation, conduction system disease or ventricular arrhythmias may precede the clinical manifestation of genetic cardiomyopathies and should by themselves raise the suspicion of an underlying genetic aberration [35]. A case of short-coupled premature ventricular complex-triggered ventricular fibrillation in a young man with a previous aborted cardiac arrest and implantable cardioverter defibrillator (ICD) insertion has been reported [36]. The patient was subsequently found to harbour a likely pathogenic de novo splice site variant in the *TTN* gene (c.56647+1G > A), implicated in DCM.

Our own cardiogenetic clinic experience on six South Indian paediatric patients with symptomatic DCM and strong family history (n = 6) revealed variants in *NEXN* (n = 2) and *TNNT2* (n = 1) genes and 1 VUS each in *MYH7*, *DES*, *PRDM16* and *RAF1* genes, with one patient

presenting with severe heart failure at 1 year of age and showing compound heterozygosity (*NEXN* and *MYH7* variants) [37]. Our three adult patients with DCM were found to have variants in *NEXN*, *MYH7*, *LMNA* and *TNNT2* genes, two of them being compound heterozygous, all variants being reported as conflicted classification of VUS/LP and the *NEXN* variant p. Glu421Ter being reclassified as LP during re-analysis [22].

2.3. Arrhythmogenic cardiomyopathy (ACM)

ACM, earlier known as arrhythmogenic right ventricular cardiomyopathy (ARVC), is a progressive condition characterized by fibro or fibrofatty myocardial replacement associated with a spectrum of symptoms from benign palpitations to severe arrhythmias and SCD [1]. With a low estimated prevalence of about 1 in 5000, ACM has barely been systematically studied in India. Pamuru et al. have reported their findings on 34 patients from the state of Andhra Pradesh [38]. Conducted In the pre-NGS era, they used PCR-based single-strand conformation polymorphism analysis, and samples with abnormal band pattern were commercially sequenced to detect novel variants in the *RYR2* and *PKP2* genes in affected individuals.

In our experience with South Indian ACM patients (n = 3), we have reported a pathogenic *PKP2* variant presenting as a sports-related SCD in a 17-year-old male, a VUS each in the *DSP* and *DOLK* genes in a 42-year-old male with cardioembolic stroke and recurrent ventricular tachycardia, and a VUS each in genes *KCNH2* and *MYLK2* in a 59-year-old female with ACM morphology and recurrent ventricular tachycardia [22,39]. While both *KCNH2* and *MYLK2* genes are not associated with ACM, variants in the *DOLK* gene are associated with congenital disorders of glycosylation which may present as DCM or ACM. A young girl from South India presenting with congestive heart failure due to ACM, curly, woolly hair since birth and palmoplantar keratoderma was diagnosed with Carvajal disease and confirmed with a genetic test that revealed her *DSP* gene to be homozygous for the c.3901C > T (p.Gln1301X) change in exon 23 [40].

3. Inherited arrhythmia syndromes

Inherited arrhythmia syndromes or cardiac channelopathies, known to cause life-threatening arrhythmias and SCD in young individuals with structurally normal hearts, are best diagnosed by a systematic investigation protocol that includes a thorough symptom and family history, an electrocardiogram (resting/exercise/ambulatory/drug challenge), imaging modalities as warranted to rule out structural abnormalities, and phenotype-guided genetic testing [41].

3.1. Congenital long QT syndrome (LQTS)

LQTS is the prototype of the inherited arrhythmias as it has had the longest runway for unravelling genotype-phenotype correlations since its first gene *KCNQ1* was discovered in the 1990s. However, it was not until two decades later that the first study on 46 South Indian LQTS probands and their first degree relatives was published [42]. Novel variations and/or compound heterozygosity were identified in 18 probands, resulting in a 39% yield. Subsequently, novel biallelic *KCNQ1* variants were reported in 10 patients with Jervell and Lange-Nielsen syndrome (JLNS) and autosomal recessive LQTS [43].

LQTS masquerading as seizures and delaying diagnosis is a well-recognized phenomenon. This has been reported in two gene-positive South Indian patients presenting with seizures in childhood [44] and in a few other case reports where genetic confirmation was not sought. Also, the global prevalence of LQTS is around 1 in 2000 to 1 in 2500 but this likely is an under estimation due to silent mutations and variable penetrance.

In our own experience, we have genotyped 6 paediatric and 1 adult proband with clinical suspicion of LQTS and detected LP/P variants in 4 (57%), 2 loss-of-function variants in *KCNQ1* and 2 gain-of-function variants in *SCN5A* [22,37]. Similar to Qureshi et al., [42] we observed that genotyping patients with a QTc <500 ms resulted in a poor yield. Interestingly, both the LQT3 probands reported from South India were middle-aged female patients presenting with minimal or no symptoms, a QTc >500 ms and history of SCD of a young male child [22,45].

Kakarla et al. have recently reported a case of neonatal calmodulinopathy with a novel *CALM3* variant (c.287A > G in exon 5), posing a management challenge but responding well to propranolol, mexiletine and left cardiac sympathetic denervation [46].

3.2. Catecholaminergic polymorphic ventricular tachycardia (CPVT)

CPVT, an inherited arrhythmogenic disorder characterized by malignant arrhythmias during physical activity and emotional bursts, is rare and sparingly reported in our population. A likely pathogenic heterozygous variant Ser72Tyr in exon 3 and a pathogenic homozygous missense variant in exon 65 (g.237700452G > A) of the *RYR2* gene have been independently reported in two CPVT probands, respectively [37,47]. Running-induced syncope was present in both cases; history of SCD of a sibling and several members in the maternal lineage triggered an early genetic testing in one while absence of such history lead to a delay of about a year before arriving at the diagnosis in the other. A case of heterozygous *CASQ2* variant-related CPVT has been reported in a young male with exertional syncope and a strong family history of SCD [48].

3.3. Brugada syndrome (BrS)

BrS is a sodium channelopathy characterized by ST-segment elevation in the right precordial leads and malignant ventricular arrhythmias with a prevalence of about 1 in 2000 worldwide and a higher prevalence in Asia. While there are a few reports of fever-triggered Brugada ECG pattern and a study documenting the efficacy of flecainide challenge test in unmasking type 1 Brugada ECG pattern in suspicious cases [49], there are no reports of gene-positive BrS in our population. Nevertheless, genetic testing with sequencing of *SCN5A* is recommended for an index case diagnosed with BrS with a type I ECG in standard or high precordial leads occurring either spontaneously, or induced by sodium-channel blockade in the presence of supporting clinical features or family history [1].

3.4. Multifocal ectopic Purkinje-related premature contractions (MEPPC)

MEPPC is a new kid on the block and a rare cardiac disorder characterized by frequent multifocal ectopic ventricular beats with narrow QRS complexes, originating from various ectopic foci along the fascicular-Purkinje system. Affected individuals have a high daily burden of multifocal premature ventricular contractions which can potentially induce a reversible form of left ventricular dilation with systolic dysfunction, known as premature ventricular contraction-induced cardiomyopathy. We have reported a case of MEPPC with an *SCN5A* R222Q variant and a family history of SCD in an asymptomatic 13-year old female patient [50] while a symptomatic adult female with MEPPC and DCM harbouring a different *SCN5A* variant (p. Arg814Trp; R814W) has also been reported [51].

3.5. Sudden cardiac death (SCD)

Postmortem genetic testing or molecular autopsy is indicated in SCD victims in addition to proper examination of the heart during autopsy by an expert cardiac pathologist [5]. Arava et al. have elegantly documented the molecular autopsy findings in 59 young SCD (18–45 years of age) cases from North India identifying P/LP variants in 12% cases [52]. It is imperative for tertiary care hospitals in India to counsel the family of the deceased on the need for a thorough medical autopsy which includes genetic testing.

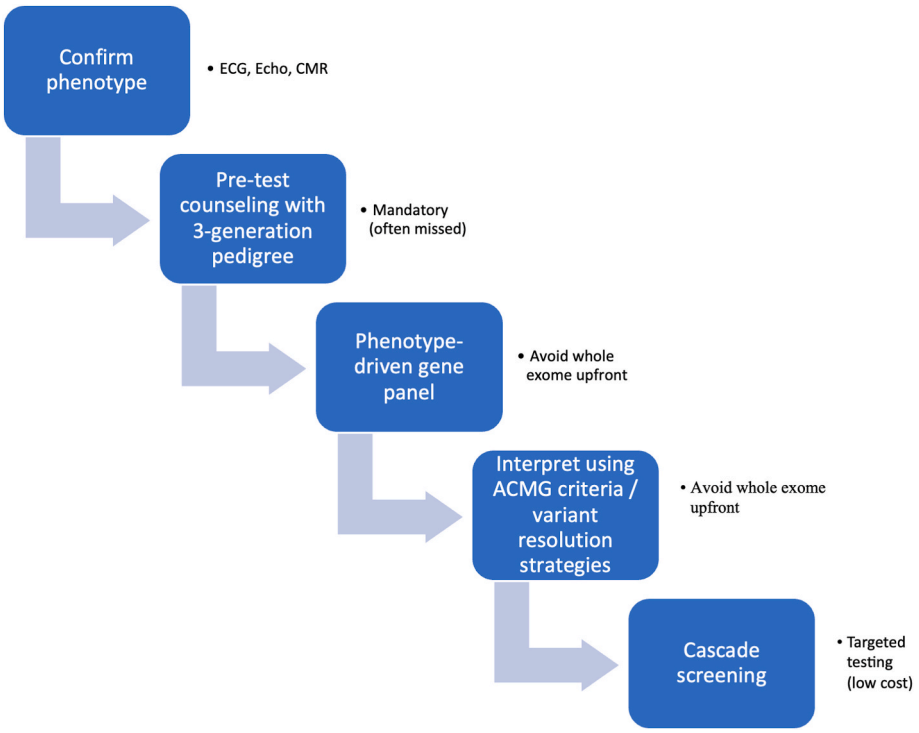

Fig. 1. A stepwise genetic testing pathway for India.

4. Next generation sequencing (NGS)

NGS which refers to an advanced gene sequencing technology allows for the rapid sequencing of millions of DNA fragments simultaneously, providing comprehensive insights into genome structure, genetic variations, gene expression profiles and epigenetic modifications [53]. NGS is currently available in India at an affordable cost. A recent study on 22 primary cardiomyopathy probands performed using targeted capture and sequencing resulted in identification of candidate variants in about 70% enabling appropriate management of disease in affected individuals as well as in screening and early diagnosis in asymptomatic family members [54]. Amongst the analysed cases, 22% were inconclusive without any significant variant identified. This is an expected outcome of NGS studies and should be borne in mind while counseling the patient prior to the test and also during interpretation of the report. Established variant resolution strategies have to be followed to interpret the findings and counsel the carriers effectively. As an increasing number of physicians start practicing phenotype-directed genetic testing such as specific gene panels, and as more and more genetic information pertaining to our population with cardiac diseases becomes available, it is expected that variants of uncertain significance (VUS) will reduce significantly. Fig. 1 outlines the steps to be followed by physicians when submitting an individual to genetic testing.

4.1. The role of genetic counseling

Genetic counseling is an invaluable but often overlooked component of genetic testing. It is ideally provided pre-test and post-test; the first, to gather a three-generational history and draft a pedigree chart, and inform the patient about the nuances and the possible outcomes of the test and the second, to help the patient understand the report with the relevance and implications of the various aspects like inheritance and classification of variant [55]. With various commercial laboratories offering genetic testing services, it is the responsibility of referring physicians to verify the credibility of the laboratory and specifically request for genetic counseling by a certified/trained counsellor with expertise in this domain. The role of cascade screening, which refers to clinical evaluation and when indicated genetic testing of relatives, should be explained to the affected individuals and their family members as it is the best available tool to identify and treat asymptomatic carriers and prevent SCD. When a LP/P variant has been identified in a proband, targeted variant testing which is low cost and looks for the particular variant only, is performed in the family members.

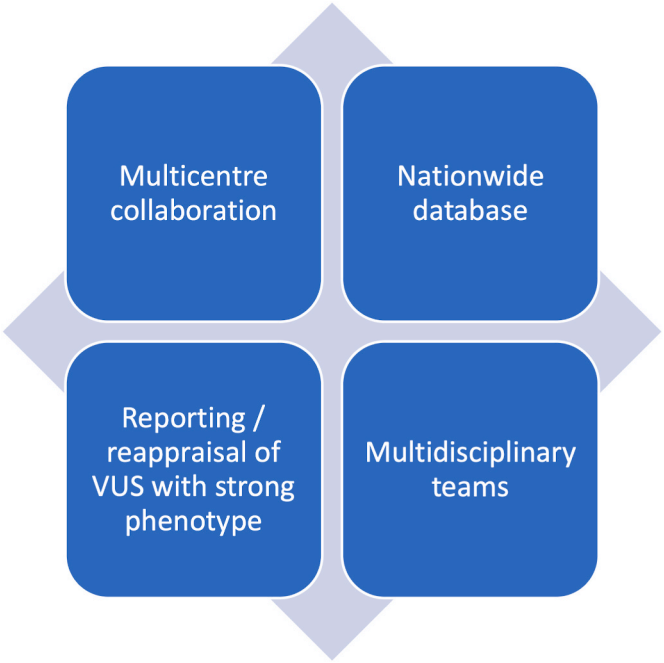

Fig. 2. Future directions to grow the field of Cardiogenetics in India.

4.2. Future directions

Multicentre collaboration, nationwide database, reporting and reappraisal of all VUS with strong phenotype and multidisciplinary cardiogenetic teams (Fig. 2) are needed to move the field forward in our country. Parallelly, we need to recognize that there is a pressing need for physicians trained in the diagnosis and management of persons with complex genetic diagnoses and work towards the promotion of enhanced undergraduate medical education in genetics and increased recruitment of medical graduates into genetics residency training programs.

5. Conclusions

India is a treasure trove of cultural and genetic variation and we are just beginning to understand the genetic underpinnings of cardiomyopathies and channelopathies in our population. With heightened awareness and knowledge, cardiologists, paediatricians and other physicians will be enabled to identify suspicious cases, perform thorough phenotyping and proceed with judicious genotyping or refer them to a multidisciplinary expert cardiogenetic clinic for further management.

Declaration of competing interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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