**Supplementary Table.** Naxos disease and related cardio-cutaneous syndromes. Publication, clinical and genetic features of all published cases up to 07/2024.

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| ID | Author,  Year | Co\*\* | Age  year | S | Gene | Variant | Z | CP | Symptoms, events | ECG | ECHO | CMR | FU, yrs | Outcome | CH | Cutaneous manifestation,  Onset | |
| 1 | Protonotarios, 20011 | GR | 7 | M | *JUP* | c.2157del2 | Ho | ARVC | No | TWI V1-V3 | RV, progression° | NR | 16 | No  Events | NR | Infancy, PPK, WH | |
| 2 | Protonotarios, 20011 | GR | 17 | M | *JUP* | c.2157del2 | Ho | ARVC | Syncope | TWI V1-V6 | RV progression° | NR | 11 | rVT,  HFD | NR | Infancy, PPK, WH | |
| 3 | Protonotarios, 20011 | GR | 32 | F | *JUP* | c.2157del2 | Ho | ARVC | Syncope | TWI V1-V4 | RV, LV progression° | NR | 16 | rVT, ICDs | NR | Infancy, PPK, WH | |
| 4 | Protonotarios, 20011 | GR | 29 | M | *JUP* | c.2157del2 | Ho | ARVC | Syncope | TWI V1-V4 | RV, LV progression° | NR | 15 | rVT, HF, ICDs | NR | Infancy, PPK, WH | |
| 5 | Protonotarios, 20011 | GR | 17 | M | *JUP* | c.2157del2 | Ho | ARVC | Syncope | TWI V1-V3 | RV progression° | NR | 11 | rVT, HF, SCD | RV + | Infancy, PPK, WH | |
| 6 | Protonotarios, 20011 | GR | 41 | F | *JUP* | c.2157del2 | Ho | ARVC | Syncope,  Dyspnoea | TWI V1-V5 | RV, LV progression° | NR | 12 | NSVT,  HF, SCD | RV+,  LV+ | Infancy, PPK, WH | |
| 7 | Protonotarios, 20011 | GR | 36 | F | *JUP* | c.2157del2 | Ho | ARVC | No | TWI V1 | RV | NR | 15 | No Events | NR | Infancy, PPK, WH | |
| 8 | Protonotarios, 20011 | GR | 43 | M | *JUP* | c.2157del2 | Ho | ARVC | Palp | NR | RV, LV progression° | NR | 13 | rVT, HF | NR | Infancy, PPK, WH | |
| 9 | Protonotarios, 20011 | GR | 35 | M | *JUP* | c.2157del2 | Ho | ARVC | Chest Pain | TWI V1-V4 | RV, LV progression° | NR | 15 | SCD | NR | Infancy, PPK, WH | |
| 10 | Protonotarios, 20011 | GR | 32 | F | *JUP* | c.2157del2 | Ho | ARVC | No | TWI V1-V3 | RV progression° | NR | 16 | No Events | NR | Infancy, PPK, WH | |
| 11 | Protonotarios, 20011 | GR | 74 | M | *JUP* | c.2157del2 | Ho | ARVC | Syncope | TWI V1-V5 | RV progression° | NR | 7 | No Events | NR | Infancy, PPK, WH | |
| 12 | Protonotarios, 20011 | GR | 61 | F | *JUP* | c.2157del2 | Ho | ARVC | Syncope | TWI V1-V4 | RV progression° | NR | 16 | NSTV | NR | Infancy, PPK, WH | |
| 13 | Protonotarios, 20011 | GR | 56 | F | *JUP* | c.2157del2 | Ho | ARVC | Syncope | TWI V1-V3 | RV progression° | NR | 15 | VES | NR | Infancy, PPK, WH | |
| 14 | Protonotarios, 20011 | GR | 49 | F | *JUP* | c.2157del2 | Ho | ARVC | Syncope | TWI V1-V6 | RV progression° | NR | 12 | Recurrent VT | NR | Infancy, PPK, WH | |
| 15 | Protonotarios, 20011 | GR | 40 | M | *JUP* | c.2157del2 | Ho | ARVC | Syncope | TWI V1-V5 | RV, LV progression° | NR | 15 | VES | NR | Infancy, PPK, WH | |
| 16 | Protonotarios, 20011 | GR | 60 | M | *JUP* | c.2157del2 | Ho | ARVC | No | TWI V1-V4 | RV progression° | NR | 12 | No Events | NR | NR Infancy, PPK, WH | |
| 17 | Protonotarios, 20011 | GR | 26 | M | *JUP* | c.2157del2 | Ho | ARVC | Syncope | TWI V1-V6 | RV, LV ° | NR | 1 | SCD | NR | Infancy, PPK, WH | |
| 18 | Protonotarios, 20011 | GR | 19 | M | *JUP* | c.2157del2 | Ho | ARVC | Palp | TWI V1-V6 | RV, LV progression° | NR | 14 | rVT, HFD | RV + LV+ | Infancy, PPK, WH | |
| 19 | Protonotarios, 20011 | GR | 16 | F | *JUP* | c.2157del2 | Ho | ARVC | Syncope | LQRSV  TWI inferior leads | RV, LV° | NR | 1 | HF, SCD | NR | Infancy, PPK, WH | |
| 20 | Protonotarios, 20011 | GR | 47 | M | *JUP* | c.2157del2 | Ho | No | No | TWI V1-V4 | RV alterations° | NR | 8 | No Events | NR | Infancy, PPK, WH | |
| 21 | Protonotarios, 20011 | GR | 45 | F | *JUP* | c.2157del2 | Ho | No | No | TWI V1-V3 | RV alterations° | NR | 4 | No Events | NR | Infancy, PPK, WH | |
| 22 | Protonotarios, 20011 | GR | 15 | F | *JUP* | c.2157del2 | Ho | ARVC | VES, Syncope | TWI V1-V2 | RV, LV° | NR | 5 | SCD | RV+  LV+ | Infancy, PPK, WH | |
| 23 | Protonotarios, 20011 | GR | 17 | F | *JUP* | c.2157del2 | Ho | No | Syncope | TWI V1-V3 | RV progression° | NR | 4 Years | ICD | NR | Infancy, PPK, WH | |
| 24 | Protonotarios, 20011 | GR | 58 | F | *JUP* | c.2157del2 | Ho | No | No | TWI V1-V6 | RV alterations° | NR | 2 Years | No Events | NR | Infancy, PPK, WH | |
| 25 | Protonotarios, 20011 | GR | 13 | M | *JUP* | c.2157del2 | Ho | No | Syncope | TWI V1-V4 | RV alterations° | NR | 2 Years | No Events | NR | Infancy, PPK, WH | |
| 26 | Protonotarios, 20011 | GR | 5 | F | *JUP* | c.2157del2 | Ho | No | No | TWI V1-V2 | RV alterations° | NR | 2 Years | No Events | NR | Infancy, PPK, WH | |
| 27 | Protonotarios, 20011 | GR | 39 | F | *JUP* | c.2157del2 | Ho | No | No | TWI V1-V2 | RV alterations° | NR | 2 Years | No Events | NR | Infancy, PPK, WH | |
| 28 | Protonotarios, 20011 | GR | 1 | M | *JUP* | c.2157del2 | Ho | No | No | TWI V1-V2 | RV alterations° | NR | 3 Years | No Events | NR | Infancy, PPK, WH | |
| 29 | Narin, 20032 | TR | 13 | M | *JUP* | c.2157del2 | Ho | ARVC | Syncope | TWI V1-V6 | RV | NR | NR | VT | NR | Infancy, PPK, WH | |
| 30 | Lazaros, 20093 | GR | 43 | M | *JUP* | c.2157del2 | Ho | ARVC | Syncope | TWI V1-V4 | RV, LV | RV, LV | NR | VT,  ICD | NR | Infancy, PPK, WH | |
| 31 | Erken, 20114 | TR | 34 | M | *JUP* | c.794G>A | Ho | ARVC | Syncope,  Dyspnoea | TWI precordial | RV, LV | RV, LV | 3 m | VT, | RV (+) | Infancy, PPK, WH, WH | |
| 32 | Erken, 20014 | TR | 46 | M | *JUP* | c.794G>A | Ho | ARVC | HF | TWI V1-V3 | RV | NR | NR | ICD | RV (+)  EMB | PPK, Alopecia |
| 33 | Baykan, 20155 | TR | 13 | M | *JUP* | c.2157del2 | Ho | ARVC | Chest Pain, Sync | TWI | RV, LV | NR | 5 | rVT, ICD | NR | Infancy, PPK, WH |
| 34 | Baykan, 20155 | TR | 6 | M | *JUP* | c.2157del2 | Ho | No | Chest Pain | NSVT | Normal | NR | 5 | No Events | NR | Infancy, PPK, WH |
| 35 | Baykan, 20155 | TR | 1.5 | F | *JUP* | NR | Ho | No | NR | NR | NR | NR | NR | NR | NR | Toddler, PPK, WH,  Dystrophic Nails |
| 36 | Baykan, 20155 | TR | 3 | M | *JUP* | c.2157del2 | Ho | DCM | Dyspnoea |  | LV, RV | NR | 0 | SCD | NR | Toddler, PPK, WH |
| 37 | Boente, 20166 | AR | 17 | M | *JUP* | c.71C>A | Ho | ARVC | NR | NR | RV, LV | NR | NR | NR | NR | PPK, WH,  Oligodontia  Nail Dystrophy |
| 38 | Sonsoz, 20217 | TR | 19 | F | *JUP* | c.901\_903delGAG | Ho | ARVC | NR | TWI V1-V6 | RV, LV | LV, RV (LVEF 21%). LV, RV LGE | NR | ICD | NR | PPK, WH,  Sparse Eyebrows |
| 39 | Sonsoz, 20217 | TR | 15 | M | *JUP* | c.901\_903delGAG | Ho | No | NR | NR | Normal | Normal | NR | NR | NR | PPK, WH,  Nail Dysplasia  Sparse Eyebrows |
| 40 | Te Riele ASJM, 20218 | GR | 1.6 | M | *JUP* | c.2157del2 | Ho | No | no | Normal | Normal | Not done | 5 years | 3 VES in Holter | No | Infancy, PPK, dorsal eczematous lesions, WH |
| 41 | Alcalai, 20039 | IL | 16 | F | *DSP* | c.7402G>C | Ho | ARVC | Syncope | TWI V1-V3, VT | RV | NR | NR | ICD | NR | PPK, WH |
| 42 | Uzumcu, 200610 | TR | 3 | M | *DSP* | c.3799C>T | Ho | DCM | NR | NSVT | LV, RV (LVEF <25%) | NR | NR | HFD | NR | Infancy, PPK, WH |
| 43 | Norgett, 200611 | UK | 3 | M | *DSP* | c.1817\_1846dup | He | DCM | NR | NSVT | LV | NR | 4 Years | ICD, SCD | NR | Toddler, PPK, WH,  Oligodontia |
| 44 | Prompona, 200712 | GER | 12 | M | *DSP* | Deletion in Exon 23 | Ho | DCM | NR | TWI V2-V6, | LV, RV (LVEF < 20%), | LV, RV (LVEF 24%).  LV LGE | NR | Heart Transplantation | RV, LV + | Infancy, PPK, WH |
| 45 | Alonso-Orgaz, 200713 | ES | 43 | M | *DSP* | c.2447A>C | He  2 | ARVC | Palp. Syncope | TWI  II,III,aVF  V1-V6 | NR | NR | NR | NR | NR | Infancy, PPK,WH |
| 46 | Tanaka, 200914 | BR | 5 | F | *DSP* | c.2516del; c.3971del | He  2 | DCM | Dyspnoea | Frequent PVCs | RV, LV | NR | NR | NR | NR | Infancy, PPK, WH  Thick nails, Alopecia |
| 47 | Mahoney, 201015 | FI | 14 | F | *DSP* | c.6310del; c.7964C>A | He2 | DCM | Asymptomatic | NR | NR | NR | NR | SCD (sleep) | LV, RV  + | Adolescence, PPK, WH,  Enamel Defects  Dystrophy and Thickening of the Toenails |
| 48 | Christensen, 201016 | DK | 35 | M | *DSP* | c.6166G>C | Ho | Biventricular ARVC | Palp. Dyspnoea | NR | NR | NR | NR | NR | NR | PPK, Curly hair |
| 49 | Chalabreysse, 201117 | FR | 17 | M | *DSP* | c.1790C>T | He | DCM | Syncope (Exercise) | IRBBB, NSVT | Dilated LV (LVEF 20%) | NR | 4 Years | LV progression (LVEF 20%), Heart Transplantation | LV, RV  + | PPK, WH,  Oligodontia |
| 50 | Chalabreysse, 201117 | FR | NR | M | *DSP* | c.1790C>T | He | No | Syncope (Exercise) | IVCD | LV Dilation (LVEF 55%) | NR | NR | NR | NR | PPK, WH, Oligodontia |
| 51 | Williams, 201118 | GER | 6 | M | *DSP* | c.5208del | Ho | DCM | NR | NR | LV (LVEF 11%) | NR | 2 Years | HFD | NR | Toddler, PPK, WH |
| 52 | Williams, 201118 | GER | 5 | M | *DSP* | c.5208delAG | Ho | DCM | NR | NR | LV  (LVEF 26%) | LV (LVEF 26%)  LV LGE | 5 Years | Heart Transplantation | RV+  LV+ | Toddler, PPK |
| 53 | Krishnamurthy, 201119 | IN | 11 | F | *DSP* | c.3901C>T | Ho | DCM | Palp,  Dyspnoea | Frequent PVCs | RV, LV  (LVEF 30%) | NR | NR | NR | NR | PPK, WH |
| 54 | Boulé, 201220 | FR | 29 | M | *DSP* | c.1691C>T | He | ARVC | Asymptomatic | TWI II,  III, aVF, V4-V6, Freq VES | LV (LVEF 35%). RV Aneurysm | RV, LV LGE +\_ | NR | ICD | NR | Infancy, PPK, WH,  Oligodontia  Leukonychia |
| 55 | Boulé, 201220 | FR | 10 | M | *DSP* | c.1691C>T | He | ARVC | Chest Pain | PVCs, Epsilon | RV | RV LGE | NR | NR | NR | PPK, WH, Oligodontia  Brittle Nails |
| 56 | Keller, 201221 | CH | 7 | M | *DSP* | c.1748T>C | He | DCM | NR | NR | RV, LV  (LVEF 19%). | RV, LVLGE | 7 | ICDs | NR | Toddler, PPK, WH  Dystrophic Nails  Sparse Eyebrows |
| 57 | Keller, 201221 | CH | 13 | F | *DSP* | c.1691C>T | He | DCM | Syncope | IVCD Epsilon PVCs | RV, LV | NR | 9 | ICD | NR | PPK, WH,  Oligodontia, Leukonychia |
| 58 | Rasmussen, 201322 | DK | 8 | M | *DSP* | c.7780del | Ho | DCM | HF | NR | LV, RV  (LVEF 15%) | NR | 5 | Heart Transplantation | RV+  LV+ | Childhood, PPK, WH |
| 59 | Yesudian, 201423 | UK | 2 | F | *DSP* | c.7567del; c.6577G>A | HE2 | DCM | NR | LV | NR | NR | NR | NR | NR | Infancy, PPK,  Alopecia |
| 60 | Baykan, 201524 | TR | 6 | M | *DSP* | NR | NR | DCM | Dyspnoea | 1st AV Block, TWI V1-V3 | RV, LV  (LVEF 27%) | NR | 2 | HFD | NR | Childhood, PPK, WH |
| 61 | Molho-Pessach, 201525 | IL | 4 | M | *DSP* | c.3924del | Ho | DCM | Dyspnoea | Frequent PVCs | LV | RV, LV, LVLGE | NR | ICD | NR | Infancy, PPK, WH |
| 62 | Molho-Pessach, 201525 | IL | 59 | F | *DSP* | c.7111C>A | Ho | ARVC | Chest Pain, Syncope | NR | RV, LV | NR | NR | ICD | NR | Infancy  PPK, WH,  Onychogryphosis, Sparse Eyebrows |
| 63 | Molho-Pessach, 201525 | IL | 21 | F | *DSP* | c.7111C>A | Ho | DCM | Chest Pain, Syncope | Frequent PVCs, Couples and Triplets | LV, RV | NR | NR | SCD | NR | Infancy, PPK, WH,  Sparse Eyebrows |
| 64 | Pigors, 201526 | RO | 5 | M | *DSP* | c.7566\_7567delAAinsC; c.7756C>T | He2 | No | No | No | No | NR | NR | NR | NR | Toddler, PPK,  Hypotrichosis |
| 65 | Pigors, 201526 | GER | 14 | M | *DSP* | c.1067C>A; c.2131\_2132del | He2 | DCM | HF | NR | NR | NR | NR | Heart Transplantation | NR | Infancy, PPK, WH |
| 66 | Pigors, 201525 | GER | 10 | F | *DSP* | c.1067C>A; c.2131\_2132del | He2 | DCM | NR | NR | NR | NR | NR | NR | NR | Infancy, PPK,  Alopecia |
| 67 | Bitar, 201628 | USA | 17 | M | *DSP* | c.1865T>C | He2 | DCM | No | NR | LV (LVEF 25%) | NR | NR | NR | NR | Infancy, PPK, WH  Oligodontia  Leukonychia |
| 68 | Finsterer, 201629 | AT | 43 | F | *DSP* | c.1678A>T | He | DCM | NR | NR | LV | NR | NR | NR | NR | Adolescence, PPK, WH,  Oligodontia |
| 69 | Finsterer, 201629 | AT | NR | M | *DSP* | c.1678A>T | He | DCM | Recurrent Syncope | NR | NR | NR | NR | Cardiac Death | RV+ | Dental abnormalities |
| 70 | Ramoglu, 201730 | TR | 5 | F | *DSP* | c.4650\_4651del | Ho | DCM | Dyspnoea | NR | LV, RV,  (LVEF 25%) | NR | 2 | Heart Transplantation | NR | PPK, WH |
| 71 | Erolu, 201831 | TR | 4 | M | *DSP* | c.3564T>A; c.4395T>A | He2 | DCM | Palp | IVCD Epsilon Waves | RV, LV (LVEF 45%) | RV, LVLGE | NR | VT, ICD | NR | PPK,WH,  Sparse Eyebrows |
| 72 | Akdogan, 201832 | TR | 8 | F | *DSP* | c.4650\_4651del | Ho | DCM | NR | Polymorphic PVC | LV (LVEF 25%) | NR | NR | NR | NR | Toddler, PPK, WH  Leukonychia |
| 73 | Akdogan, 201832 | TR | 5 | F | *DSP* | c.4650\_4651del | Ho | DCM | NR | NR | NR | NR | NR | NR | NR | Toddler, PPK,WH |
| 74 | Ou, 202233 | FR | 13 | F | *DSP* | c.1782C>G | He | ARVC | NR | Frequent PVCs | NR | RV (RVEF 32%) | NR | NR | NR | Infancy, PPK, WH  Oligodontia  Fragile Nails |
| 75 | Zhao, 202334 | CN | 7 | F | *DSP* | c.4597C>T | Ho | DCM | Dyspnoea | IVCD Frequent PVCs | RV, LV (LVEF 25%) | RV, LV | NR | NR | NR | Toddler, PPK, WH  Oligodontia  Leukonychia |
| 76 | Kincaid, 202335 | CA | 37 | M | *DSP* | c.2652\_2653del | He | NR | Chest Pain, Palp | TWI, NSTV (exercise) | LV | Fibrofatty Infiltration | NR | ICD | NR | PPK, WH |
| 77 | Kincaid, 202335 | CA | 34 | M | *DSP* | c.2652\_2653del | He | NR | NR | NR | NR | No | NR | NR | NR | PPK, WH |
| 78 | Kincaid, 202335 | CA | 44 | F | *DSP* | c.2652\_2653del | He | NR | NR | NR | NR | NR | NR | NR | NR | PPK, WH |
| 79 | Kincaid, 202335 | CA | 1 | F | *DSP* | c.2652\_2653del | He | NR | NR | NR | NR | NR | NR | NR | NR | curly hair |
| 80 | Kincaid, 202335 | CA | 3 | F | *DSP* | c.2652\_2653del | He | NR | NR | NR | NR | NR | NR | NR | NR | Focal Hyperkeratosis |
| 81 | Kincaid, 202335 | CA | 4 | M | *DSP* | c.2652\_2653del | He | NR | NR | NR | NR | NR | NR | NR | NR | Focal Hyperkeratosis |
| 82 | Kincaid, 202335 | CA | 2 | F | *DSP* | c.2652\_2653del | He | NR | NR | NR | NR | NR | NR | NR | NR | Focal Hyperkeratosis |
| 83 | Kincaid, 202335 | CA | Months | F | *DSP* | c.2652\_2653del | He | NR | NR | NR | NR | NR | NR | NR | NR | NR |
| 84 | Norget 200037  Carvajal-Huerta, 199836  Kaplan 2004 59 | EC | 10 | F | *DSP* | c.7901del | Ho | DCM | NR | LQRSV,  TWI V1-V4,  III, aVF,  PVCs | LV | NR | 1 | HFD | RV, LV, aneurysms, fibrosis | Infancy, PPK, WH |
| 85 | Norget 200037  Carvajal-Huerta, 199836 | EC | 12 | F | *DSP\** | c.7901del | Ho | DCM | NR | Abnormal | LV | NR | 5 Years | HFD | NR | Infancy, PPK, WH |
| 86 | Norget 200037  Carvajal-Huerta, 199836 | EC | 14 | F | *DSP\** | c.7901del | Ho | DCM | NR | Abnormal, Frequent PVCs and NSVT | LV | NR | NR | NR | NR | Infancy, PPK, WH |
| 87 | Norget 200037  Carvajal-Huerta, 199836 | EC | 8 | F | *DSP\** | c.7901del | Ho | DCM | NR | Abnormal, Frequent PVCs and Couplets | LV | NR | NR | NR | NR | Infancy, PPK, WH |
| 88 | Norget 200037  Carvajal-Huerta, 199836 | EC | 10 | M | *DSP\** | c.7901del | Ho | No | NR | Abnormal, Frequent PVCs | Normal | NR | NR | NR | NR | Infancy, PPK, WH |
| 89 | Carvajal-Huerta, 199836, 37 | EC | 13 | M | *DSP\** | c.7901del | Ho | DCM | NR | Abnormal | LV | NR | 4 Years | HFD | NR | Infancy, PPK, WH |
| 90 | Carvajal-Huerta, 199836 | EC | 32 | F | NR | NR | NR | No | NR | Abnormal | Normal | NR | NR | NR | NR | Infancy, PPK, WH |
| 91 | Carvajal-Huerta, 199836 | EC | 34 | F | *NR* | NR | NR | No | NR | Abnormal | Normal | NR | NR | NR | NR | Infancy, PPK, WH |
| 92 | Carvajal-Huerta, 199836 | EC | 11 | F | *NR* | NR | NR | DCM | NR | Abnormal | LV | NR | 1 Year | Advanced HF-Related Death | NR | Infancy, PPK, WH |
| 93 | Carvajal-Huerta, 199836 | EC | 12 | F | *NR* | NR | NR | DCM | NR | Abnormal | LV | NR | NR | Advanced HF-Related Death | NR | Infancy, PPK, WH |
| 94 | Carvajal-Huerta, 199836 | EC | 14 | F | *NR* | NR | NR | DCM | NR | Abnormal, Frequent PVCs and Couplets | LV | NR | NR | NR | NR | Infancy, PPK, WH |
| 95 | Carvajal-Huerta, 199836 | EC | 17 | F | *NR* | NR | NR | DCM | NR | Abnormal, Frequent PVCs and Couplets | LV | NR | NR | NR | NR | Infancy, PPK, WH |
| 96 | Bukhari, 200338 | SA | 2 | F | NR | NR | NR | No | NR | NR | Normal | NR | NR | NR | NR | Toddler, PPK, WH,  Thin Nails  Sparse Eyebrows |
| 97 | Sajeev, 200639 | IN | 55 | F | NR | NR | NR | ARVC | Palp, Syncope | TWI V1-V4 | RV Aneurysms | NR | NR | Sustained VT | NR | PPK, WH |
| 98 | Adhisivam, 200640 | IN | 2 | F | NR | NR | NR | DCM | HF | Normal | RV, LV Dysfunction (LVEF 40%) | NR | 0 | HFD | NR | Infancy, PPK, WH |
| 99 | Kilic, 200741 | TR | 17 | M | NR | NR | NR | ARVC | Dyspnoea, Palp | TWI V1-V3, Epsilon Waves | RV, LV | RV, LV | NR | ICD | NR | Adolescent, PPK, WH |
| 100 | Kilic, 200741 | TR | NR | M | NR | NR | NR | ARVC | NR | Epsilon Waves | NR | NR | NR | NR | NR | No |
| 101 | Kilic, 200741 | TR | NR | M | NR | NR | NR | ARVC | NR | NR | RV | RV | NR | NR | NR | PPK |
| 102 | Rai, 200842 | IN | 7 | M | NR | NR | NR | No | NR | TWI V1-V2 | RV and RA Dilation | NR | NR | NR | NR | Infancy.PPK,WH |
| 103 | Kolar, 200843 | UK | 16 | F | NR | NR | NR | DCM | NR | NR | NR | NR | NR | SCD | RV, LV Fibrosis and Myocyte Loss. | Adolescence, PPK, WH |
| 104 | Tanaka, 200914 | BR | 10 | M | *PKP1* | c.2014C>T | Ho | NR | NR | NR | NR | NR | NR | NR | NR | Toddler, PPK, WH  Thickening of the Fingernails and Toenails |
| 105 | Simpson, 200944 | UK | 29 | M | *DSC2* | c.1841del | Ho | ARVC | Syncope, Resuscitated SCD | IVCD  TWI V2, III, aVF, Frequent PVCs | LV (LVEF 45%), RV Dilation and Dysfunction | LV (LVEF 50%) RV (RVEF 32%) RV, LV LGE | NR | ICD | NR | PPK WH |
| 106 | Simpson, 200944 | UK | 25 | F | *DSC2* | c.1841del | Ho | ARVC | NR | IVCD Biphasic TW in V2, Frequent PVCs | Normal | RV+/- | NR | NR | NR | PPK, WH |
| 107 | Koumantaki, 201045 | GR | 54 | F | NR | NR | NR | NR | HF | NR | NR | NR | NR | ICD | NR | PPK, WH |
| 108 | Chalabreysse, 201117 | FR | 15 | M | NR | NR | NR | No | Chest Pain | ST Elevation in V3-V4, PVCs | No | NR | NR | NR | NR | PPK, WH  Oligodontia |
| 109 | Noin, 201246 | ES | 14 | M | NR | NR | NR | ARVC | AbortedSCD (Exercise) | IVCD | RV, Aneurysm | NR | 0 | ICD | NR | Infancy, PPK, WH  Yes |
| 110 | Saravanan, 201247 | IN | 21 | M | NR | NR | NR | ARVC | VT | Epsilon Wave | RV | RV, RVLGE, | NR | VT | NR | PPK, WH |
| 111 | Barber, 201248 | UK | 14 | F | NR | NR | NR | DCM | Asymptomatic | NR | NR | NR | NR | SCD (Exertion) | LV Dilation LV Patchy Fibrosis | Adolescence, PPK, WH,  White thickened nails  Sparse Eyebrows |
| 112 | Nehme, 201249 | FR | 57 | M | NR | NR | NR | DCM | Dyspnoea | 1st AV Block | LV (LVEF 30%) and LVNC | NR | NR | NR | NR | Childhood, PPK, WH |
| 113 | Gultekin, 201350 | TR | 54 | F | NR | NR | NR | ARVC | HF, Syncope | TWI V1-V3 Epsilon Waves. Polymorphic NSVT | RV (RVEF 14%). (LVEF 60%) | RV | NR | No Events | NR | PPK, WH  Alopecia |
| 114 | Salam, 201351 | IN | 11 | F | NR | NR | NR | DCM | Syncope | CRBBB, LPFB | LV (LVEF 45%) | No LGE | 2 | Refractory Sustained VT, Progressive HF | NR | PPK, WH |
| 115 | Kaya, 201352 | TR | 23 | F | NR | NR | NR | ARVC | Dyspnoea | LQRSV, TWI V2-V6 | RV, LV  (LVEF 23%) | RV, LV, LVLGE | NR | NR | NR | Childhood, PPK, WH |
| 116 | Yildiz, 201353 | TR | 9 | F | NR | NR | NR | DCM | Syncope, Palp | NR | LV | NR | NR | VT | NR | Infancy, PPK, WH |
| 117 | Baykan, 201554 | TR | 9 | F | NR | NR | NR | DCM | HF Palp, Syncope | NR | RV, LV | NR | 1 | SCD | NR | Childhood, PPK, WH |
| 118 | Islam, 201655 | BD | 10 | M | NR | NR | NR | ARVC | Palp, Chest Pain, Progressive Dyspnoea | IRBBB, Epsilon Waves | RV Aneurysms | NR | 9m | VT, SCD | NR | PPK, WH |
| 119 | Dutta, 201656 | IN | 4 | F | NR | NR | NR | ARVC | Recurrent Syncope, Dyspnoea | Frequent PVC, IVCD | RV, LV (LVEF 48%) | NR | NR | NR | NR | PPK, WH |
| 120 | Mandal, 202257 | NP | 9 | F | NR | NR | NR | DCM | Dyspnoea | LQRSV, TWI, Frequent PVCs | LV (LVEF 20%) and RV dysfunction | NR | NR | NR | NR | Childhood, PPK, WH |

*Abbreviations:* ARVC, arrhythmogenic right ventricular cardiomyopathy; AF, atrial fibrillation; AVB, atrioventricular block; CH, cardiac histology; Co: Country; CP, cardiac phenotype; CRBBB, complete right bundle branch block; CMR, cardiac magnetic resonance; DCM, dilated cardiomyopathy; ECHO, echocardiography; EMB, endomyocardial biopsy; F, female; FH, family history; FW, free wall; HF, heart failure; HFD, Heart failure related death; He, heterozygosity; He2, compound heterozygosity; Ho, homozygosity; GER, Germany; GR, Greece; ICD, implantable cardioverter-defibrillator; ICDs, appropriate ICD shock; IRBBB, incomplete right bundle branch block; IV, interventricular; IVCD, Intraventricular conduction delay; LGE, late gadolinium enhancement; LPFB, left posterior fascicular block; LQRSV, low QRS voltages; LV, left ventricle; LVEF, left ventricular ejection fraction; m, months; M, male; NDLVC, non-dilated left ventricular cardiomyopathy; NR, not reported; NSVT, non-sustained ventricular tachycardia; Palp, palpitations; PPK, Palmoplantar keratoderma; PVC, premature ventricular complex; RA, right atrium; RV, right ventricle; rVT, recurrent VT; S, Sex; SCD, sudden cardiac death; Symptoms; TWI, T-wave inversion; VES, frequent ventricular extrasystoles; VT, ventricular tachycardia; WH, wolly hair; Z, zygosity.

°RV abnormalities including hypokinetic, akinetic or dyskinetic areas, diastolic bulging and trabecular disarrangement. Structural progression was defined as progressive alteration in ventricular dimensions and/or wall motion abnormalities during serial echocardiographic evaluation.

\*\*: Countries are reported by international codes

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