| **Table S8: Genic regions recurrently affected by SNVs and indels** | | | | |
|---|---|---|---|---|
| **Gene** | **Patients** | **Reason for gene selection** | **Inheritance** | **Variant** |
| *ACOX2* | HK180C | Seed -CH- | | NC_000003.11:g.[58510169C>T];[58512313C>T] |
| | VH106C | | Maternal | NC_000003.11:g.58520147C>T |
| *CDC14A* | HK164C | Seed -CH- | | NG_051602.1:g.[159238G>A];[84272C>A] |
| | HD09C | | Paternal | NG_051602.1:g.159358C>G |
| *CUL7* | HK180C | Seed -CH- | | NG_016205.1:g.[21037C>G];[10418C>T] |
| | VH105C | Seed -CH- | | NG_016205.1:g.[12938C>G];[20980G>A] |
| | HD09C | | Paternal | NG_016205.1:g.18757A>G |
| *FGFRL1* | HD09C | Seed -CH- | | NC_000004.11:g.[1018102G>A];[1018242G>A] |
| | VH106C | | Paternal | NC_000004.11:g.1016203G>C |
| *FRAS1* | HD09C | | Maternal | NG_015812.1:g.228865_228866insT |
| | HK164C | Seed -CH- | | NG_015812.1:g.[454891A>G];[388680A>C] |
| *GLI2* | HK164C | ENS gene -Table S3- | Paternal | NG_009030.1:g.195951G>A |
| | VH105C | ENS gene -Table S3- | Maternal | NG_009030.1:g.5166G>A |
| *LAMA5* | HK9C | Seed -CH- | | NG_050626.1:g.[53760G>A];[57239G>A] |
| | VH106C | | Paternal | NG_050626.1:g.51487C>T |

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|---|---|---|---|---|
| | VH108C | | Maternal | NG_050626.1:g.57903T>G |
| | HK96C | | Maternal | NG_050626.1:g.44336C>T |
| *NEK1* | HK164C | Seed -Homo- | | NG_027982.1:g.[109891G>A];[109891G>A] |
| | HK97C | | Paternal | NG_027982.1:g.55474A>G |
| | VH106C | | Maternal | NG_027982.1:g.55474A>G |
| *NRG1* | VH106C | Seed -De novo-; ENS gene -Table S3- | *NCDS de novo* | NG_012005.2:g.667454G>C |
| | VH108C | Seed -De novo-; ENS gene -Table S3- | *NCDS de novo* | NG_012005.2:g.146124A>G |
| | HK164C | Seed -De novo-; ENS gene -Table S3- | *NCDS de novo* | NG_012005.2:g.92222G>T |
| *PLXNB1* | HK180C | ENS gene -Table S3- | Paternal | NC_000003.11:g.48453691G>T |
| | VH106C | ENS gene -Table S3- | Maternal | NC_000003.11:g.48453691G>T |
| *RET* | HD09C | ENS gene -Table S3- | Paternal | NG_007489.1:g.39340G>A |
| | HK180C | ENS gene -Table S3- | Paternal | NG_007489.1:g.30277G>A |
| | HK9C | ENS gene -Table S3- | Maternal | NG_007489.1:g.52819C>T |
| *SLC24A1* | HK164C | Seed -CH- | | NG_031968.2:g.[39211A>G];[19238G>A] |
| | HK97C | | Paternal | NG_031968.2:g.19346C>G |
| *VRK2* | HK164C | Seed -Homo- | | NG_029717.2:g.[256898G>A];[256898G>A] |
| | HD09C | | Maternal | NG_029717.2:g.186911A>G |

| | | | | |
|---|---|---|---|---|
| | HK97C | | Maternal | NG_029717.2:g.237075A>C |
| *ZEB2* | HK164C | Seed -de novo-; ENS gene -Table S3- | *NCDS de novo* | NC_000002.11:g.145137510C>T |
| | HK97C | ENS gene -Table S3- | Paternal | NG_016431.1:g.125370A>C |

CH: compound heterozygous; *RET* mutations have been described previously and are known to be present in healthy individuals.