**Supplement Table 2. Clinical and genetic information in the patients with *PRRT2* mutations**

|  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Year | Reference | Mode | Mutation | Code | Exon | Family | Patient | AAO | Age/Gender | Phenotype | Other |
| 2009 | Lipton J | S | Microdeletion | deletion | gene | 1 | 1 | Child/infancy | 17y/M | PKD/IC | Dopa-responsive parkinsonism, learning disability |
| 2011 | Dale RC | S | Microdeletion | deletion | gene | 1 | 1 | 7y/11m | NA/M | PKD/IC |  |
| 2011 | Najmabadi H | AR | 649dupC | R217PfsX8 | 2 | 1 | 1 | NA | NA/NA | Intellectual disability |  |
| 2011 | Chen W | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 5 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 6 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 7 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 8 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 9 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 10 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 11 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 12 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 13 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 14 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 15 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 16 | 6y | 14y/M | PKD |  |
|  |  |  |  |  |  |  | 17 | NA | NA/F | PKD |  |
| 2011 | Chen W | AD | 649dupC | R217PfsX8 | 2 | 2 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 5 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 6 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 7 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 8 | NA | NA/M | PKD |  |
| 2011 | Chen W | AD | 514\_517delTCTG | S172RfsX3 | 2 | 3 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
| 2011 | Chen W | AD | 649dupC | R217PfsX8 | 2 | 4 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
| 2011 | Chen W | AD | 972delA | V325SfsX12 | 3 | 5 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
| 2011 | Chen W | AD | 649dupC | R217PfsX8 | 2 | 6 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
| 2011 | Chen W | AD | 649dupC | R217PfsX8 | 2 | 7 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
| 2011 | Chen W | AD | 649dupC | R217PfsX8 | 2 | 8 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 5 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 6 | NA | NA/M | PKD |  |
| 2011 | Wang J | AD | 649dupC | R217PfsX8 | 2 | A | 1 | 18y | 65y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 10y | 41y/F | PKD |  |
|  |  |  |  |  |  |  | 3 | 6y | 39y/M | PKD |  |
|  |  |  |  |  |  |  | 4 | 10y/6m | 19y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 5 | 10y/12m | 18y/F | PKD/IC |  |
| 2011 | Wang J | AD | C487T | Q163X | 2 | B | 1 | 13y | 63y/F | PKD |  |
|  |  |  |  |  |  |  | 2 | 10y | 41y/M | PKD |  |
|  |  |  |  |  |  |  | 3 | 8y | 38y/M | PKD |  |
|  |  |  |  |  |  |  | 4 | 8y | 36y/F | PKD |  |
|  |  |  |  |  |  |  | 5 | 16y | 19y/F | PKD |  |
|  |  |  |  |  |  |  | 6 | 4y/6m | 6y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 7 | 5y/6m | 7y/M | PKD/IC |  |
| 2011 | Wang J | AD | 649dupC | R217PfsX8 | 2 | C | 1 | 10y | 59y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 9y | 52y/F | PKD |  |
|  |  |  |  |  |  |  | 3 | 9y | 50y/F | PKD |  |
|  |  |  |  |  |  |  | 4 | 10y | 47y/M | PKD |  |
|  |  |  |  |  |  |  | 5 | 8y | 19y/F | PKD |  |
|  |  |  |  |  |  |  | 6 | 9y | 16y/F | PKD |  |
| 2011 | Wang J | AD | 649dupC | R217PfsX8 | 2 | D | 1 | 8y | 48y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 10y | 43y/F | PKD |  |
|  |  |  |  |  |  |  | 3 | 9y | 26y/M | PKD |  |
|  |  |  |  |  |  |  | 4 | 13y | 21y/M | PKD |  |
| 2011 | Wang J | AD | C796T | R266W | 2 | E | 1 | 8y | 48y/F | PKD |  |
|  |  |  |  |  |  |  | 2 | 10y | 22y/F | PKD |  |
| 2012 | Li J | S | 649dupC | R217PfsX8 | 2 | 6 | 1 | 10y | 22y/M | PKD |  |
| 2012 | Li J | S | 649dupC | R217PfsX8 | 2 | 11 | 1 | 7y | 24y/M | PKD |  |
| 2012 | Li J | S | 649dupC | R217PfsX8 | 2 | 16 | 1 | 15y | 32y/M | PKD |  |
| 2012 | Li J | S | 649dupC | R217PfsX8 | 2 | 17 | 1 | 5y | 20y/M | PKD |  |
| 2012 | Li J | S | 649dupC | R217PfsX8 | 2 | 18 | 1 | 13y | 24y/M | PKD |  |
| 2012 | Li J | S | 649dupC | R217PfsX8 | 2 | 28 | 1 | 10y | 32y/M | PKD |  |
| 2012 | Li J | S | 1011\_1012delCG+1\_9delGTGAGTGGG | Splice site | 3 | 22 | 1 | 8y | 15y/M | PKD |  |
| 2012 | Li J | S | 964delG | V322WfsX15 | 3 | 26 | 1 | 10y | 23y/M | PKD |  |
| 2012 | Li J | S | T841C | W281R | 2 | 27 | 1 | 13y | 23y/M | PKD |  |
| 2012 | Li J | S | C922T | R308C | 3 | 29 | 1 | 7y | 24y/M | PKD |  |
| 2012 | Li J | AD | G859A | A287T | 2 | F1 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 16y | 88y/M | PKD |  |
|  |  |  |  |  |  |  | 3 | 7y/12m | 63y/F | PKD/IC |  |
|  |  |  |  |  |  |  | 4 | 10y | 60y/F | PKD |  |
|  |  |  |  |  |  |  | 5 | 10y | 54y/M | PKD |  |
|  |  |  |  |  |  |  | 6 | 8y | 53y/F | PKD |  |
|  |  |  |  |  |  |  | 7 | 10y | 53y/F | PKD |  |
|  |  |  |  |  |  |  | 8 | 13y/12m | 45y/M | PKD/FC |  |
|  |  |  |  |  |  |  | 9 | 1y | 58y/M | FC |  |
|  |  |  |  |  |  |  | 10 | 13y/5m | 37y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 11 | 10y/7m | 34y/F | PKD/IC |  |
|  |  |  |  |  |  |  | 12 | 9y/7m | 32y/F | PKD/IC |  |
|  |  |  |  |  |  |  | 13 | 7y/5m | 26y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 14 | 8y | 28y/M | PKD |  |
|  |  |  |  |  |  |  | 15 | 3m | 19y/F | FC |  |
|  |  |  |  |  |  |  | 16 | 9m | 7y/F | IC |  |
| 2012 | Li J | AD | 649dupC | R217PfsX8 | 2 | F2 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | 11y | 90y/M | PKD |  |
|  |  |  |  |  |  |  | 5 | 1y | 82y/M | PKD |  |
|  |  |  |  |  |  |  | 6 | 12y | 43y/M | PKD |  |
|  |  |  |  |  |  |  | 7 | 11y | 38y/F | PKD |  |
|  |  |  |  |  |  |  | 8 | 1y | 45y/M | PKD |  |
|  |  |  |  |  |  |  | 9 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 10 | - | NA/F | - | Incomplete |
| 2012 | Li J | AD | 369dupG | S124VfsX10 | 2 | F4 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 3 | 7y | 46y/F | PKD |  |
|  |  |  |  |  |  |  | 4 | 6y | 23y/M | PKD |  |
| 2012 | Liu Q | AD | 649dupC | R217PfsX8 | 2 | A | 1 | 6m | 61y/M | IC |  |
|  |  |  |  |  |  |  | 2 | <1m | 40y/F | IC |  |
|  |  |  |  |  |  |  | 3 | <1m | 38y/M | IC |  |
|  |  |  |  |  |  |  | 4 | 5m | 22y/M | IC |  |
|  |  |  |  |  |  |  | 5 | 3m | 2y/F | IC |  |
|  |  |  |  |  |  |  | 6 | 5m | 3y/M | IC |  |
|  |  |  |  |  |  |  | 7 | 8y | 29y/M | PKD |  |
|  |  |  |  |  |  |  | 8 | 10y | 18y/M | PKD |  |
|  |  |  |  |  |  |  | 9 | 8y | 9y/M | PKD |  |
| 2012 | Liu Q | AD | 904dupG | D302GfsX39 | 3 | B | 1 | 18y/<1m | 36y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | 10y/5m | 11y/M | PKD/IC |  |
| 2012 | Liu Q | AD | 649dupC | R217PfsX8 | 2 | C | 1 | NA | NA/M | PNKD-like |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PNKD-like |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | PNKD-like |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | PNKD-like |  |
|  |  |  |  |  |  |  | 5 | NA | NA/F | PNKD-like |  |
|  |  |  |  |  |  |  | 6 | 8y | NA/F | PNKD-like |  |
|  |  |  |  |  |  |  | 7 | 7y | 40y/M | PNKD-like |  |
|  |  |  |  |  |  |  | 8 | 2y | 18y/M | PNKD-like | Migraine |
| 2012 | Liu Q | AD | 649dupC | R217PfsX8 | 2 | E | 1 | 5y | 38y/F | PKD | Migraine |
|  |  |  |  |  |  |  | 2 | 5y | 35y/F | PKD |  |
|  |  |  |  |  |  |  | 3 | 7y/NA | 18y/F | PKD/FC | Migraine |
|  |  |  |  |  |  |  | 4 | 7y | 14y/M | PKD |  |
| 2012 | Liu Q | AD | 649dupC | R217PfsX8 | 2 | F | 1 | 8y | 60y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | 10y | 21y/M | PKD |  |
| 2012 | Liu Q | S | G913A | G305R | 3 | Case13 | 1 | 15y | 21y/M | PKD, Epilepsy | Migraine |
| 2012 | Liu Q | S | 649dupC | R217PfsX8 | 2 | Case14 | 1 | 11y | 12y/M | PKD |  |
| 2012 | Liu Q | S | 649dupC | R217PfsX8 | 2 | Case15 | 1 | 12y | 14y/F | PKD |  |
| 2012 | Heron SE | AD | 879+5G>A | Splice site | Intron2 | F1 | n=9 | 9m† | NA/4M5F | BFIE |  |
|  |  |  |  |  |  |  | n=4 | - | NA/3M1F | - | Incomplete |
| 2012 | Heron SE | AD | 629dupC | A211SfsX14 | 2 | F2 | n=11 | 6.7m† | NA/5M6F | PKD(1M3F)BFIE(2M2F)PKD/IC(2M1F) |  |
| 2012 | Heron SE | AD | 879+1G>T | Splice site | Intron2 | F3 | n=3 | 6m† | NA/2M1F | BFIE |  |
|  |  |  |  |  |  |  | n=1 | - | NA/F | - | Incomplete |
| 2012 | Heron SE | AD | G950A | S317N | 3 | F4 | n=10 | 4.4m† | NA/5M5F | BFIE(2M5F)PKD(2M) Epilepsy(1M) |  |
|  |  |  |  |  |  |  | n=10 | - | NA/4M6F | - | Incomplete |
| 2012 | Heron SE | AD | 649dupC | R217PfsX8 | 2 | F5 | n=9 | 5.2m† | NA/7M2F | PKD/IC(3M)BFIE(3M2F)Epilepsy(1M) |  |
| 2012 | Heron SE | AD | 649dupC | R217PfsX8 | 2 | F6 | n=7 | 6.4m† | NA/5M2F | BFIE |  |
| 2012 | Heron SE | AD | 649dupC | R217PfsX8 | 2 | F7 | n=6 | 8.2m† | NA/2M4F | BFIE |  |
| 2012 | Heron SE | AD | 649dupC | R217PfsX8 | 2 | F8 | n=5 | 9.5m† | NA/1M4F | BFIE |  |
| 2012 | Heron SE | AD | 649dupC | R217PfsX8 | 2 | F9 | n=7 | 6.5m† | NA/3M4F | BFIE |  |
|  |  |  |  |  |  |  | n=3 | - | NA/1M2F | - | Incomplete |
| 2012 | Heron SE | AD | 649dupC | R217PfsX8 | 2 | F10 | n=2 | 6.8m† | NA/2M | BFIE |  |
|  |  |  |  |  |  |  | n=1 | - | NA/F | - | Incomplete |
| 2012 | Heron SE | AD | 649dupC | R217PfsX8 | 2 | F11 | n=3 | 4.3m† | NA/2M1F | BFIE |  |
|  |  |  |  |  |  |  | n=2 | - | NA/2F | - | Incomplete |
| 2012 | Heron SE | AD | 649dupC | R217PfsX8 | 2 | F12 | n=4 | 4.5m† | NA/2M2F | BFIE |  |
| 2012 | Heron SE | AD | 649dupC | R217PfsX8 | 2 | F13 | n=3 | 4m† | NA/1M2F | BFIE |  |
|  |  |  |  |  |  |  | n=1 | - | NA/M | - | Incomplete |
| 2012 | Heron SE | AD | 649dupC | R217PfsX8 | 2 | F14 | n=3 | 4.3m† | NA/2M1F | BFIE |  |
|  |  |  |  |  |  |  | n=1 | - | NA/F | - | Incomplete |
| 2012 | Heron SE | AD | 649dupC | R217PfsX8 | 2 | F15 | n=2 | 3.3m† | NA/2F | BFIE |  |
| 2012 | Heron SE | AD | 649dupC | R217PfsX8 | 2 | F16 | n=3 | 10.7m† | NA/1M2F | BFIE |  |
| 2012 | Heron SE | AD | 649dupC | R217PfsX8 | 2 | F17 | n=5 | 6.3m† | NA/4M1F | BFIE |  |
| 2012 | Heron SE | AD | 649dupC | R217PfsX8 | 2 | F18 | n=2 | 6m† | NA/1M1F | PKD(1M)BFIE(1F) |  |
| 2012 | Heron SE | AD | 649dupC | R217PfsX8 | 2 | F19 | n=2 | 5.5m† | NA/2M | PKD/IC(1M)BFIE(1M) |  |
| 2012 | Cao L | AD | 573dupT | G192WfsX7 | 2 | M6 | 1 | 10y/NA | 21y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | - | NA/F | - |  |
| 2012 | Cao L | AD | 649dupC | R217PfsX8 | 2 | M3 | 1 | 22y | 26y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | - | NA/F | - | Incomplete |
| 2012 | Cao L | AD | 649dupC | R217PfsX8 | 2 | M11 | 1 | 7y | 22y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | PKD |  |
| 2012 | Van Strien TW | AD | NA | R217X | 2 | 1 | 1 | 1.5y | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | 5m | NA/M | IC |  |
|  |  |  |  |  |  |  | 3 | 6m | NA/F | IC |  |
|  |  |  |  |  |  |  | 4 | 15y/4m | 16y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 5 | 15y | NA/M | PKD |  |
| 2012 | Dale RC | S | Microdeletion | Gene deletion | Gene | 1 | 1 | 6y | NA/NA | PKD |  |
| 2012 | Dale RC | S | Microdeletion | Gene deletion | Gene | 1 | 1 | 10y | NA/NA | PKD |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS1 | 1 | 6m | 68y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6m | 65y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 6m | 47y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 6m | 47y/M | BFIE |  |
|  |  |  |  |  |  |  | 5 | 6m | 12y/M | BFIE |  |
|  |  |  |  |  |  |  | 6 | 6m | 37y/F | BFIE |  |
|  |  |  |  |  |  |  | 7 | 6m | 15y/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS5 | 1 | 6m | 46y/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6m | 15y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 4m | 13y/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | 5m | 5y/M | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS9 | 1 | 3m | 26y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 5m | 3y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 5m | 0.5y/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS10 | 1 | 6m | 63y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6m | 58y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 6m | 70y/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | 4m | 33y/M | BFIE |  |
|  |  |  |  |  |  |  | 5 | 4m | 34y/F | BFIE |  |
|  |  |  |  |  |  |  | 6 | 4m | 24y/F | BFIE |  |
|  |  |  |  |  |  |  | 7 | 5m | 3y/F | BFIE |  |
|  |  |  |  |  |  |  | 8 | 5m | 5m/M | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS11 | 1 | NA | 27y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | NA | 39y/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | 5m | 9y/M | BFIE |  |
|  |  |  |  |  |  |  | 5 | 6m | 5y/M | BFIE |  |
|  |  |  |  |  |  |  | 6 | 6m | 3y/M | BFIE |  |
|  |  |  |  |  |  |  | 7 | 3m | 30y/F | BFIE |  |
|  |  |  |  |  |  |  | 8 | NA | 18y/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS12 | 1 | infancy | 60y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 3m | 27y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 6m | 11y/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | 2m | 8y/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | 6m | 35y/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS13 | 1 | 8m | 39y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 8m | 34y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 8m | 17y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 5 | - | NA/F | - |  |
|  |  |  |  |  |  |  | 6 | - | NA/F | - |  |
|  |  |  |  |  |  |  | 7 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 8 | - | NA/M | - |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS14 | 1 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 2 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | NA | 65y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 6m | 6y/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | 5m | 40y/M | BFIE |  |
|  |  |  |  |  |  |  | 6 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 7 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 8 | 6m | 35y/M | BFIE |  |
|  |  |  |  |  |  |  | 9 | 6m | 4y/F | BFIE |  |
|  |  |  |  |  |  |  | 10 | 6m | 6y/M | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS15 | 1 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | infancy | 65y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 4 | - | NA/F | - |  |
|  |  |  |  |  |  |  | 5 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 6 | 6m | 3y/F | BFIE |  |
|  |  |  |  |  |  |  | 7 | 4m | 5y/M | BFIE |  |
| 2012 | Schubert J | S | 649dupC | R217PfsX8 | 2 | BFIS17 | 1 | 8m | 8m/M | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS18 | 1 | 6m | 72y/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | 3m | 38y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 6m | 34y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 3m | 36y/M | BFIE |  |
|  |  |  |  |  |  |  | 5 | 6m | 8y/M | BFIE |  |
|  |  |  |  |  |  |  | 6 | 4m | 9y/M | BFIE |  |
| 2012 | Schubert J | AD | 629delC | A211SfsX14 | 2 | BFIS19 | 1 | 6m | 42y/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6m | 4y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 5m | 2y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 4m | 8y/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | 6m | 15y/F | BFIE |  |
|  |  |  |  |  |  |  | 6 | 4m | 12y/M | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIE20 | 1 | infancy | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 7m | 7m/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS21 | 1 | 3.5m | 9y/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS22 | 1 | 8m | 38y/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | 8m | 12y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 6m | 7y/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS24 | 1 | NA | 73y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | 16y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 5m | 5m/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS25 | 1 | 7m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | 7m | 7m/M | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS26 | 1 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | 5m | 7y/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS28 | 1 | 7y | 54y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | infancy | 56y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 9m | 31y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | infancy | 40y/M | BFIE |  |
|  |  |  |  |  |  |  | 5 | infancy | 30y/M | BFIE |  |
|  |  |  |  |  |  |  | 6 | 2m | 21y/M | BFIE |  |
|  |  |  |  |  |  |  | 7 | 8m | 2y/F | BFIE |  |
|  |  |  |  |  |  |  | 8 | 6m | 9m/F | BFIE |  |
| 2012 | Schubert J | NA | G968A | G323E | 3 | BFIS30 | 1 | 7m | 1.5y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS34 | 1 | infancy | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 3m | 22y/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS35 | 1 | infancy | 75y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 4m | 40y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 4m | 50y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 4m | 37y/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | 4m | 42y/F | BFIE |  |
|  |  |  |  |  |  |  | 6 | 6m | 39y/F | BFIE |  |
|  |  |  |  |  |  |  | 7 | 6m | 39y/F | BFIE |  |
|  |  |  |  |  |  |  | 8 | 5m | 23y/M | BFIE |  |
|  |  |  |  |  |  |  | 9 | 5m | 28y/F | BFIE |  |
|  |  |  |  |  |  |  | 10 | 5m | 9y/F | BFIE |  |
|  |  |  |  |  |  |  | 11 | 3m | 30y/F | BFIE |  |
|  |  |  |  |  |  |  | 12 | 4m | 9y/M | BFIE |  |
|  |  |  |  |  |  |  | 13 | 3y | 6y/M | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS36 | 1 | 6m | 32y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 11m | 11y/M | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS37 | 1 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | NA | 7y/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS38 | 1 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | 3m | 41y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 5m | 5m/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 9m | 4y/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS39 | 1 | 7m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 7m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 7m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 7m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | 7m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 6 | 7m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 7 | 7m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 8 | 7m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 9 | 7m | NA/ F | BFIE |  |
|  |  |  |  |  |  |  | 10 | 7m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 11 | 7m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 12 | 4.5m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 13 | 7m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 14 | 7m | NA/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS42 | 1 | 6m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 6m | 12y/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS43 | 1 | infancy | 82y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6m | 38y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 9m | 2y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 5m | 4y/F | BFIE |  |
| 2012 | Schubert J | AD | 291delC | N98TfsX17 | 2 | BFIS44 | 1 | NA | 44y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6m | 14y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 6m | 24y/M | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS45 | 1 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | 71y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 6m | 33y/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | 5m | 7y/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | 6m | 3y/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS46 | 1 | 4m | 78y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 5m | 46y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 4m | 10y/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS47 | 1 | NA | 38y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 3 | 6m | 34y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 5m | 20y/M | BFIE |  |
|  |  |  |  |  |  |  | 5 | 5m | 10y/F | BFIE |  |
|  |  |  |  |  |  |  | 6 | 5m | 6y/M | BFIE |  |
|  |  |  |  |  |  |  | 7 | - | NA/F | - |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS48 | 1 | 5m | 11y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 5m | 7y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 4m | 7y/M | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS49 | 1 | 6m | 15y/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | 7m | 11y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 8m | 11y/M | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS50 | 1 | 5m | 6y/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | 5m | 5y/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS51 | 1 | 5m | 7y/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6m | 10y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 4m | 8y/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | 6m | 46y/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS53 | 1 | 7m | 8y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | 43y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | NA | 69y/M | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS54 | 1 | 7m | 4y/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6m | 35y/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS55 | 1 | 6m | 5y/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6m | 8y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 5m | 10y/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | 6m | 43y/M | BFIE |  |
|  |  |  |  |  |  |  | 5 | NA | 41y/F | BFIE |  |
|  |  |  |  |  |  |  | 6 | 5m | 60y/M | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS57 | 1 | 8m | 13y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6m | 34y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | NA | 59y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | NA | 91y/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | NA | 41y/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS59 | 1 | 6m | 8y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 5m | 32y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 8m | 24y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 6m | 36y/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | 6m | 53y/M | BFIE |  |
|  |  |  |  |  |  |  | 6 | NA | 92y/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS61 | 1 | 8m | 12y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 7m | 7y/F | BFIE |  |
| 2012 | Schubert J | AD | 649dupC | R217PfsX8 | 2 | BFIS62 | 1 | NA | 4m/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | 3y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | NA | 7y/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | NA | 32y/F | BFIE |  |
| 2012 | Ono S | AD | 649dupC | R217PfsX8 | 2 | F1 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 5 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 6 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 7 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 8 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 9 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 10 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 11 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 12 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 13 | NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 14 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 15 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 16 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 17 | NA | NA/M | PKD |  |
| 2012 | Ono S | AD | 649dupC | R217PfsX8 | 2 | F2 | 1 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 4 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 5 | NA/NA | NA/M | PKD/IC |  |
| 2012 | Ono S | AD | 649dupC | R217PfsX8 | 2 | F3 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
| 2012 | Ono S | AD | 649dupC | R217PfsX8 | 2 | F4 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 5 | NA/NA | NA/M | PKD/IC |  |
| 2012 | Ono S | AD | 649dupC | R217PfsX8 | 2 | F5 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | PKD |  |
| 2012 | Ono S | AD | 649dupC | R217PfsX8 | 2 | F6 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | NA/NA | NA/M | PKD/IC |  |
| 2012 | Ono S | AD | 649dupC | R217PfsX8 | 2 | F7 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 5 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 6 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 7 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 8 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 9 | NA | NA/F | PKD |  |
| 2012 | Ono S | AD | 649dupC | R217PfsX8 | 2 | F8 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | PKD |  |
| 2012 | Ono S | AD | 649dupC | R217PfsX8 | 2 | F9 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 5 | NA | NA/M | PKD |  |
| 2012 | Ono S | AD | 649dupC | R217PfsX8 | 2 | F10 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | IC |  |
| 2012 | Ono S | AD | 649dupC | R217PfsX8 | 2 | F11 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 5 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 6 | NA | NA/F | PKD |  |
| 2012 | Ono S | AD | 649dupC | R217PfsX8 | 2 | F12 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
| 2012 | Ono S | AD | 649dupC | R217PfsX8 | 2 | F13 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 5 | NA | NA/M | PKD |  |
| 2012 | Ono S | AD | 649dupC | R217PfsX8 | 2 | F14 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | IC |  |
| 2012 | Ono S | AD | C748T | Q250X | 2 | F15 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | IC |  |
| 2012 | Ono S | AD | 649dupC | R217PfsX8 | 2 | F16 | 1 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 5 | NA | NA/M | IC |  |
| 2012 | Ono S | AD | 649dupC | R217PfsX8 | 2 | F17 | 1 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | IC |  |
| 2012 | Meneret A | AD | C562T | Q188X | 2 | 942-001 | 1 | 6y/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | PKD |  |
| 2012 | Meneret A | AD | 649dupC | R217PfsX8 | 2 | 783-003 | 1 | 12y | NA/F | PKD |  |
| 2012 | Meneret A | AD | 649dupC | R217PfsX8 | 2 | 855-001 | 1 | 10y | NA/M | PKD |  |
| 2012 | Meneret A | AD | 649dupC | R217PfsX8 | 2 | 022-007 | 1 | 7y | NA/M | PKD |  |
| 2012 | Meneret A | AD | 649dupC | R217PfsX8 | 2 | 448-016 | 1 | 7y | NA/M | PKD |  |
| 2012 | Meneret A | AD | 649dupC | R217PfsX8 | 2 | 208-010 | 1 | 6y | NA/M | PKD |  |
| 2012 | Meneret A | AD | 649dupC | R217PfsX8 | 2 | 946-006 | 1 | 10y | NA/M | PKD |  |
| 2012 | Meneret A | AD | 649dupC | R217PfsX8 | 2 | 943-008 | 1 | 14y | NA/F | PKD |  |
| 2012 | Meneret A | AD | 649dupC | R217PfsX8 | 2 | 939-001 | 1 | 6y/NA | NA/M | PKD/IC |  |
| 2012 | Meneret A | AD | 649dupC | R217PfsX8 | 2 | 937-001 | 1 | 10y | NA/M | PKD |  |
| 2012 | Meneret A | AD | 649dupC | R217PfsX8 | 2 | 378-003 | 1 | 8y | NA/M | PKD |  |
| 2012 | Meneret A | AD | 649dupC | R217PfsX8 | 2 | 450-054 | 1 | 6y | NA/M | PKD |  |
| 2012 | Meneret A | AD | 649dupC | R217PfsX8 | 2 | 936-003 | 1 | 6y | NA/M | PKD |  |
| 2012 | Meneret A | S | 629dupC | A211SfsX14 | 2 | 1250-002 | 1 | 6.5y | NA/M | PKD |  |
| 2012 | Meneret A | S | C649T | R217X | 2 | 945-011 | 1 | 12y | NA/M | PKD |  |
| 2012 | Meneret A | S | 649dupC | R217PfsX8 | 2 | 621-001 | 1 | 9y | NA/M | PKD |  |
| 2012 | Meneret A | S | 649dupC | R217PfsX8 | 2 | 926-003 | 1 | 12y | NA/M | PKD |  |
| 2012 | Meneret A | S | 649dupC | R217PfsX8 | 2 | 428-001 | 1 | 9y | NA/M | PKD |  |
| 2012 | Meneret A | S | 649dupC | R217PfsX8 | 2 | 753-001 | 1 | 11y | NA/F | PKD |  |
| 2012 | Meneret A | S | 649dupC | R217PfsX8 | 2 | 163-001 | 1 | 7y | NA/M | PKD |  |
| 2012 | Meneret A | S | 649delC | R217EfsX12 | 2 | 944-010 | 1 | 11y | NA/F | PKD |  |
| 2012 | Meneret A | S | 649dupC | R217PfsX8 | 2 | 930-001 | 1 | 13y | NA/F | PKD |  |
| 2012 | Groffen AJ | AD | 649dupC | R217PfsX8 | 2 | F1 | 1 | 12y | 37y/F | PKD |  |
|  |  |  |  |  |  |  | 2 | 15y | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | Migraine |  |
|  |  |  |  |  |  |  | 4 | 6m | NA/F | IC |  |
| 2012 | Groffen AJ | AD | C649T | R217X | 2 | F3 | 1 | 15y | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 15y | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 5 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 6 | NA | NA/F | IC |  |
| 2012 | Groffen AJ | S | 649delC | R217EfsX12 | 2 | 4 | 1 | 13y | NA/M | PKD |  |
| 2012 | Groffen AJ | S | 649dupC | R217PfsX8 | 2 | 6 | 1 | 11y | NA/M | PKD |  |
| 2012 | Schmidt A | AD | 649dupC | R217PfsX8 | 2 | A | 1 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 5 | NA/NA | NA/M | PKD/IC |  |
| 2012 | Schmidt A | AD | 649dupC | R217PfsX8 | 2 | B | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA/NA | NA/M | PKD/IC |  |
| 2012 | Schmidt A | AD | 649dupC | R217PfsX8 | 2 | C | 1 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 3 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | NA | NA/M | PKD |  |
| 2012 | Schmidt A | AD | 649dupC | R217PfsX8 | 2 | D | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
| 2012 | Lee HY | AD | C718T | R240X | 2 | K3323 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 5 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 6 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 7 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 8 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 9 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 10 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 11 | NA | NA/F | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K4962 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | GS |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 5 | NA | NA/M | PKD |  |
| 2012 | Lee HY | AD | 517insT | E173X | 2 | K5471 | 1 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 5 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 6 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 7 | NA | NA/F | IC |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K4998 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K3538 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K5770 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K4874 | 1 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 5 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 6 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 7 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 8 | NA | NA/M | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K2916 | 1 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K5212 | 1 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | NA/NA | NA/F | PKD/IC |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K3534 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 4 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 5 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 6 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 7 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 8 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 9 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 10 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 11 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 12 | NA | NA/F | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K3446 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 5 | NA/NA | NA/M | PKD/IC |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K7717 | 1 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 3 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 5 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 6 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 7 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 8 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 9 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 10 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 11 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 12 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 13 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 14 | NA/NA | NA/M | PKD/IC |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K7720 | 1 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 5 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 6 | NA/NA | NA/M | PKD/IC |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K7721 | 1 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 5 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 6 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 7 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 8 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 9 | NA | NA/F | IC |  |
| 2012 | Lee HY | AD | C718T | R240X | 2 | K7722 | 1 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 2 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | NA/NA | NA/F | PKD/IC |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K7719 | 1 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 5 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 6 | NA/NA | NA/M | PKD/IC |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K7718 | 1 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 4 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 5 | NA | NA/F | IC |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K7716 | 1 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 5 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 6 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 7 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 8 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 9 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 10 | NA | NA/F | IC |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | Wash U | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 5 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 6 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 7 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 8 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 9 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 10 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 11 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 12 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 13 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 14 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 15 | NA | NA/F | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K5118 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 5 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 6 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 7 | NA | NA/M | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K19599 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 5 | NA | NA/F | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K821 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K30085 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K16719 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 5 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 6 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 7 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 8 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 9 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 10 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 11 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 12 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 13 | NA | NA/M | PKD |  |
| 2012 | Lee HY | AD | 980\_981insT | I327IfsX14 | 3 | K3391 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K10615 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K50049 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K8664 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K12206 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K50112 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K6661 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | IC |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K7920 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 5 | NA/NA | NA/F | PKD/IC |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K9278 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K50078 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 4 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 5 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 6 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 7 | NA | NA/F | PKD |  |
| 2012 | Lee HY | AD | 649dupC | R217PfsX8 | 2 | K7253 | 1 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 5 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 6 | NA/NA | NA/M | PKD/IC |  |
| 2012 | Lee HY | S | 649dupC | R217PfsX8 | 2 | S1 | 1 | NA | NA/NA | PKD |  |
| 2012 | Lee HY | S | 649dupC | R217PfsX8 | 2 | S2 | 1 | NA | NA/NA | PKD |  |
| 2012 | Lee HY | S | 649dupC | R217PfsX8 | 2 | S3 | 1 | NA | NA/NA | PKD |  |
| 2012 | Lee HY | S | 649dupC | R217PfsX8 | 2 | S4 | 1 | NA | NA/NA | PKD |  |
| 2012 | Lee HY | S | 649dupC | R217PfsX8 | 2 | S5 | 1 | NA | NA/NA | PKD |  |
| 2012 | Lee HY | S | 649dupC | R217PfsX8 | 2 | S6 | 1 | NA | NA/NA | PKD |  |
| 2012 | Lee HY | S | 649dupC | R217PfsX8 | 2 | S7 | 1 | NA | NA/NA | PKD |  |
| 2012 | Lee HY | S | 649dupC | R217PfsX8 | 2 | S8 | 1 | NA | NA/NA | PKD |  |
| 2012 | Lee HY | S | 649dupC | R217PfsX8 | 2 | S9 | 1 | NA | NA/NA | PKD |  |
| 2012 | Lee HY | S | 649dupC | R217PfsX8 | 2 | S10 | 1 | NA | NA/NA | PKD |  |
| 2012 | Lee HY | S | 649dupC | R217PfsX8 | 2 | S11 | 1 | NA | NA/NA | PKD |  |
| 2012 | Lee HY | S | 649dupC | R217PfsX8 | 2 | S12 | 1 | NA | NA/NA | PKD |  |
| 2012 | Lee HY | S | 649dupC | R217PfsX8 | 2 | S13 | 1 | NA | NA/NA | PKD |  |
| 2012 | Lee HY | S | 649dupC | R217PfsX8 | 2 | S14 | 1 | NA | NA/NA | PKD |  |
| 2012 | Lee HY | S | 649dupC | R217PfsX8 | 2 | S15 | 1 | NA | NA/NA | PKD |  |
| 2012 | Lee HY | S | 649dupC | R217PfsX8 | 2 | S16 | 1 | NA | NA/NA | PKD |  |
| 2012 | Lee HY | S | 649dupC | R217PfsX8 | 2 | S17 | 1 | NA | NA/NA | PKD |  |
| 2012 | Dale RC | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | 3m/4m | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | 7y | NA/M | PKD | Migraine |
|  |  |  |  |  |  |  | 3 | 13y | NA/M | Migraine |  |
|  |  |  |  |  |  |  | 4 | 11y | NA/M | Migraine |  |
|  |  |  |  |  |  |  | 5 | NA | NA/F | Migraine |  |
| 2012 | Lee YC | AD | 272delC | P91QfsX24 | 2 | 1 | 1 | 11y | 24y/M | PKD |  |
| 2012 | Lee YC | AD | G595T | E199X | 2 | 2 | 1 | 10y | 44y/F | PKD |  |
| 2012 | Lee YC | S | 604\_607delTCAC | S202HfsX26 | 2 | 3 | 1 | 11y | 22y/M | PKD |  |
| 2012 | Lee YC | AD | 649dupC | R217PfsX8 | 2 | 4 | 1 | 13y | 21y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 5 | NA | NA/F | PKD |  |
| 2012 | Lee YC | AD | 649dupC | R217PfsX8 | 2 | 5 | 1 | 7y | 15y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
| 2012 | Lee YC | AD | 649dupC | R217PfsX8 | 2 | 6 | 1 | 8y/<4y | 21y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 4 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 5 | NA | NA/M | PKD |  |
| 2012 | Lee YC | AD | 649dupC | R217PfsX8 | 2 | 7 | 1 | 8y/<2y | 18y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
| 2012 | Lee YC | S | 649dupC | R217PfsX8 | 2 | 8 | 1 | 20y/<4y | 22y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | - | NA/M | - | Incomplete |
| 2012 | Lee YC | S | 649dupC | R217PfsX8 | 2 | 9 | 1 | 11y | 27y/M | PKD |  |
| 2012 | Lee YC | S | 649dupC | R217PfsX8 | 2 | 10 | 1 | 10y | 21y/M | PKD |  |
| 2012 | Lee YC | S | 649delC | R217EfsX12 | 2 | 11 | 1 | 18y/<4y | 19y/M | PKD/IC |  |
| 2012 | Lee YC | AD | C718T | R240X | 2 | 12 | 1 | 15y/<2y | 21y/M | PKD/IC |  |
| 2012 | Lee YC | AD | C922G | R308C | 3 | 13 | 1 | 10y | 19y/M | PKD |  |
| 2012 | van Vliet R | AD | 649delC | R217EfsX12 | 2 | A | 1 | NA | NA/NA | IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/NA | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/NA | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/NA | PKD |  |
|  |  |  |  |  |  |  | 5 | NA/NA | NA/NA | PKD/IC |  |
|  |  |  |  |  |  |  | 6 | NA | NA/NA | IC | MWA |
|  |  |  |  |  |  |  | 7 | NA | NA/NA | BFIE | MWA |
|  |  |  |  |  |  |  | 8 | NA | NA/NA | IC | MWOA |
|  |  |  |  |  |  |  | 9 | NA | NA/NA | MWOA |  |
|  |  |  |  |  |  |  | 10 | NA | NA/NA | BFIE |  |
|  |  |  |  |  |  |  | 11 | NA | NA/NA | PKD |  |
|  |  |  |  |  |  |  | 12 | NA | NA/NA | PKD | Writer’s cramp |
|  |  |  |  |  |  |  | 13 | NA/NA | NA/NA | PKD/IC |  |
|  |  |  |  |  |  |  | 14 | NA | NA/NA | PKD |  |
|  |  |  |  |  |  |  | 15 | NA | NA/NA | BFIE | MWA |
|  |  |  |  |  |  |  | 16 | NA | NA/NA | IC | MWA |
|  |  |  |  |  |  |  | 17 | NA/NA | NA/NA | PKD/IC | Migraine |
|  |  |  |  |  |  |  | 18 | NA | NA/NA | BFIE |  |
|  |  |  |  |  |  |  | 19 | NA/NA | NA/NA | PKD/IC |  |
| 2012 | van Vliet R | AD | 649delC | R217EfsX12 | 2 | B | 1 | NA | NA/NA | PKD | MWA |
|  |  |  |  |  |  |  | 2 | NA | NA/NA | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/NA | IC |  |
|  |  |  |  |  |  |  | 4 | NA | NA/NA | IC |  |
|  |  |  |  |  |  |  | 5 | NA | NA/NA | PKD |  |
|  |  |  |  |  |  |  | 6 | NA | NA/NA | BFIE |  |
| 2012 | van Vliet R | AD | C824T | S275F | 2 | C | 1 | NA | NA/NA | BFIE | MWA |
|  |  |  |  |  |  |  | 2 | NA | NA/NA | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/NA | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/NA | PKD | MWA |
|  |  |  |  |  |  |  | 5 | NA | NA/NA | PKD |  |
| 2012 | van Vliet R | AD | 649dupC | R217PfsX8 | 2 | D | 1 | NA | NA/NA | PKD |  |
| 2012 | van Vliet R | S | 649dupC | R217PfsX8 | 2 | G | 1 | NA | NA/NA | PKD |  |
| 2012 | van Vliet R | S | C649T | R217X | 2 | H | 1 | NA | NA/NA | PKD |  |
| 2012 | van Vliet R | AD | 649dupC | R217PfsX8 | 2 | I | 1 | NA | NA/NA | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/NA | PKD |  |
| 2012 | Steinlen OK | AD | 649dupC | R217PfsX8 | 2 | D12 | 1 | 5m | 49y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 3 | 8m | 24y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 5m | 18y/M | BFIE |  |
| 2012 | Steinlen OK | AD | 649dupC | R217PfsX8 | 2 | D34 | 1 | NA | 72/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6m | 43y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 5m | 5y/F | BFIE |  |
| 2012 | Steinlen OK | AD | 649dupC | R217PfsX8 | 2 | D75 | 1 | 3m | 60y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 3 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 4 | 5m | 37y/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | 6m | 24y/M | BFIE |  |
|  |  |  |  |  |  |  | 6 | 6m | 12y/F | BFIE |  |
|  |  |  |  |  |  |  | 7 | 6m | 10y/F | BFIE |  |
| 2012 | Steinlen OK | AD | 649dupC | R217PfsX8 | 2 | D96 | 1 | 12m | 70y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6m | 42/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 9m | 45y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 10m | 14y/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | 6m | 12y/F | BFIE | Migraine |
| 2012 | Steinlen OK | AD | 649dupC | R217PfsX8 | 2 | D124 | 1 | 6m | 39y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6m | 1y/M | BFIE |  |
| 2012 | Friedman J | AD | G913A | G305R | 3 | 1 | 1 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 16y | 24y/M | PKD |  |
|  |  |  |  |  |  |  | 3 | 12y | 19y/F | PKD |  |
|  |  |  |  |  |  |  | 4 | 12y | 19y/F | PKD |  |
|  |  |  |  |  |  |  | 5 | 14y | 19y/M | PKD |  |
| 2012 | Specchio N | AD | 649dupC | R217PfsX8 | 2 | F1 | 1 | <1y | 58y/M | IC |  |
|  |  |  |  |  |  |  | 2 | 5m | 33y/F | IC |  |
|  |  |  |  |  |  |  | 3 | 5m | 30y/M | IC |  |
|  |  |  |  |  |  |  | 4 | 5m | 17m/F | IC |  |
| 2012 | Specchio N | AD | 649dupC | R217PfsX8 | 2 | F2 | 1 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 4m | 46y/M | IC |  |
|  |  |  |  |  |  |  | 3 | 4m | 13y/F | IC |  |
|  |  |  |  |  |  |  | 4 | 5m | 15y/M | IC |  |
|  |  |  |  |  |  |  | 5 | 4m | 3.75y/F | IC |  |
| 2012 | Specchio N | AD | 649dupC | R217PfsX8 | 2 | F3 | 1 | <1y | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | 7m | 31y/M | IC |  |
|  |  |  |  |  |  |  | 3 | 3m | 3y/M | IC |  |
| 2012 | Specchio N | AD | 649dupC | R217PfsX8 | 2 | F4 | 1 | 6m | 48y/M | IC |  |
|  |  |  |  |  |  |  | 2 | 3m | 22y/F | IC |  |
| 2012 | Specchio N | AD | 649dupC | R217PfsX8 | 2 | F5 | 1 | 10m | 29y/M | IC |  |
|  |  |  |  |  |  |  | 2 | 5m | 6y/M | IC |  |
| 2012 | Specchio N | S | 649dupC | R217PfsX8 | 2 | Case 1 | 1 | 5m | 4y/F | IC |  |
| 2012 | Specchio N | S | C718T | R240X | 2 | Case 2 | 1 | 5m | 7y/M | IC |  |
| 2012 | Riant F | S | 649dupC | R217PfsX8 | 2 | Patient13 | 1 | 9y | NA/F | HM |  |
| 2012 | Riant F | AD | 649dupC | R217PfsX8 | 2 | Patient29 | 1 | 13y | NA/M | PKD | PED, HM, GS |
|  |  |  |  |  |  |  | 2 | 12y | NA/M | HM |  |
| 2012 | Riant F | AD | 649delC | R217EfsX12 | 2 | Patient21 | 1 | 15y | NA/F | HM |  |
|  |  |  |  |  |  |  | 2 | 14y | NA/M | HM |  |
| 2012 | Riant F | AD | 649delC | R217EfsX12 | 2 | Patient23 | 1 | 6y | NA/M | HM |  |
| 2012 | Cloarec R | AD | 649dupC | R217PfsX8 | 2 | P7-0101 | 1 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 8y/3.5m | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | 7y/4m | NA/F | PKD/IC |  |
| 2012 | Cloarec R | AD | 649dupC | R217PfsX8 | 2 | P1-0101 | 1 | 2-3y | NA/F | IC |  |
|  |  |  |  |  |  |  | 2 | 10y | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | 8y/18m | NA/F | PKD/IC | MWA |
| 2012 | Cloarec R | AD | 649dupC | R217PfsX8 | 2 | P7-0112 | 1 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 12y/6m | NA/F | PKD/IC |  |
| 2012 | Cloarec R | AD | 649dupC | R217PfsX8 | 2 | P0116-15 | 1 | 7y | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 6y/NA | NA/M | PKD/IC |  |
| 2012 | Cloarec R | AD | 434delC | R145GfsX31 | 2 | P0114 | 1 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 6m | NA/M | IC |  |
|  |  |  |  |  |  |  | 3 | 5m | NA/F | IC |  |
| 2012 | Cloarec R | AD | 649dupC | R217PfsX8 | 2 | P011-12 | 1 | NANA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 5 | 5m | NA/F | IC |  |
| 2012 | Cloarec R | AD | 649dupC | R217PfsX8 | 2 | P0131 | 1 | 11y/9m | NA/M | PKD/IC |  |
| 2012 | Cloarec R | AD | 649dupC | R217PfsX8 | 2 | P11-23 | 1 | 7y/Infancy | NA/M | PKD/IC |  |
| 2012 | Cloarec R | AD | 649dupC | R217PfsX8 | 2 | P011-13 | 1 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 5 | 5m | NA/F | IC |  |
| 2012 | Cloarec R | AD | 649dupC | R217PfsX8 | 2 | P6-010 | 1 | 3y | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | 6y/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | 8y/4m | NA/M | PKD/IC |  |
| 2012 | Cloarec R | AD | 649dupC | R217PfsX8 | 2 | P011-14 | 1 | Infancy | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | 2m | NA/M | IC |  |
| 2012 | Cloarec R | AD | 649dupC | R217PfsX8 | 2 | P0116-1 | 1 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | IC |  |
| 2012 | Cloarec R | AD | 649dupC | R217PfsX8 | 2 | P4604 | 1 | NA | NA/M | PKD | MWOA |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD | MWOA |
|  |  |  |  |  |  |  | 3 | NA | NA/M | MWOA |  |
|  |  |  |  |  |  |  | 4 | 9y | NA/M | PKD | MWOA |
| 2012 | Cloarec R | AD | 649dupC | R217PfsX8 | 2 | P2315 | 1 | NA | NA/M | PKD | HM |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD | HM |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD | HM |
| 2012 | Cloarec R | AD | 742delC | S248AfsX65 | 2 | P0116-12 | 1 | NA | NA/F | MWA |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | MWA |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | IC | MWA |
|  |  |  |  |  |  |  | 4 | 12y/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 5 | 3m | NA/F | IC |  |
| 2012 | Cloarec R | AD | 649dupC | R217PfsX8 | 2 | P0111 | 1 | 9m | NA/F | IC | MWA |
|  |  |  |  |  |  |  | 2 | NA/NA | NA/M | PKD/IC |  |
| 2012 | Cloarec R | AD | 649dupC | R217PfsX8 | 2 | P0115 | 1 | 4y | NA/M | PKD | MWOA |
|  |  |  |  |  |  |  | 2 | 4y | NA/F | PKD |  |
| 2012 | Cloarec R | AD | 649dupC | R217PfsX8 | 2 | P0113 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | PKD | MWOA |
|  |  |  |  |  |  |  | 4 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 5 | 5y/3m | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 6 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 7 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 8 | NA/NA | NA/F | PKD/IC |  |
| 2012 | Schetter IE | AD | 649dupC | R217PfsX8 | 2 | FA | 1 | NA | NA/F | Seizure |  |
|  |  |  |  |  |  |  | 2 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 3 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | NA | NA/F | BFIE |  |
| 2012 | Schetter IE | AD | 649dupC | R217PfsX8 | 2 | FB | 1 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 3 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | BFIE |  |
| 2012 | Schetter IE | AD | 649dupC | R217PfsX8 | 2 | FC | 1 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | BFIE |  |
| 2012 | Schetter IE | AD | 649dupC | R217PfsX8 | 2 | FD | 1 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | BFIE |  |
| 2012 | Schetter IE | AD | 649dupC | R217PfsX8 | 2 | FE | 1 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 4 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 5 | NA | NA/M | FS |  |
|  |  |  |  |  |  |  | 6 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 7 | NA | NA/F | BFIE |  |
| 2012 | Schetter IE | AD | 649dupC | R217PfsX8 | 2 | FF | 1 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 5 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 6 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 7 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 8 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 9 | NA | NA/F | BFIE |  |
| 2012 | Schetter IE | AD | 649dupC | R217PfsX8 | 2 | FG | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | BFIE |  |
| 2012 | Schetter IE | AD | 649dupC | R217PfsX8 | 2 | FH | 1 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | BFIE |  |
| 2012 | Schetter IE | AD | 649dupC | R217PfsX8 | 2 | FI | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA/NA | NA/M | PKD/IC |  |
| 2012 | Schetter IE | S | 649dupC | R217PfsX8 | 2 | FJ | 1 | NA | NA/M | BFIE |  |
| 2012 | Schetter IE | S | 649dupC | R217PfsX8 | 2 | FK | 1 | NA | NA/M | BFIE |  |
| 2012 | De Vries B | AD | 649dupC | R217PfsX8 | 2 | F1 | 1 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 2 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 5 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 6 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 7 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 8 | - | NA/F | - |  |
|  |  |  |  |  |  |  | 9 | - | NA/F | - |  |
|  |  |  |  |  |  |  | 10 | - | NA/F | - |  |
|  |  |  |  |  |  |  | 11 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 12 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 13 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 14 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 15 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 16 | - | NA/F | - |  |
|  |  |  |  |  |  |  | 17 | - | NA/F | - |  |
|  |  |  |  |  |  |  | 18 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 19 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 20 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 21 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 22 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 23 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 24 | NA | NA/M | BFIE |  |
| 2012 | De Vries B | AD | 649dupC | R217PfsX8 | 2 | F2 | 1 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 5 | NA | NA/F | BFIE |  |
| 2012 | De Vries B | AD | 649dupC | R217PfsX8 | 2 | F3 | 1 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 5 | NA | NA/M | BFIE |  |
| 2012 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | F1 | 1 | 12y | 27y/M | PKD |  |
| 2012 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | F2 | 1 | 14y | 36y/M | PKD |  |
| 2012 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | F3 | 1 | child | 47y/M | PKD |  |
| 2012 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | F4 | 1 | 11y | 47y/M | PKD |  |
| 2012 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | F5 | 1 | 27y | 48y/M | PKD |  |
| 2012 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F7 | n=3 | 17y | 42y/2M1F | PKD, Migraine(1) | Seizure(1) |
| 2012 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F8 | n=7 | 8-12y | 25-73y/3M4F | PKD, Migraine (3(HM2)) | Seiure4 |
| 2012 | Gardiner AR | AD | NA | 332insGAC | 3 | F9 | 1 | 5y | 15y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 5y/NA | 44y/M | PKD/IC |  |
| 2012 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F10 | 1 | 10y/NA | 22y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | 10y | 40y/M | PKD | Migraine |
| 2012 | Gardiner AR | S | C644G | P215R | 2 | F12 | 1 | 22y | 31/F | PKD | MWA |
| 2012 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | F13 | 1 | 13y | 24/M | PKD | GTCS |
| 2012 | Gardiner AR | AD | NA | L171LfsX3 | 2 | F14 | n=2 | 14y | 33/1M1F | PKD |  |
| 2012 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F15 | n=2 | 8y | 40/2M | PKD | Seizure |
| 2012 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | F16 | 1 | 11y | 44y/F | PKD | Migraine, Seizure |
| 2012 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F17 | n=3 | 6y | 49y/2M1F | PKD |  |
| 2012 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F18 | n=2 | 12y | 56y/2F | HM | Episode ataxia |
| 2012 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F19 | 1 | 8y | 48y/M | PKD | Migraine |
|  |  |  |  |  |  |  | 2 | 14y | 48y/F | PKD |  |
| 2012 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F21 | n=2 | 8-11y | 45-51y/1M1F | PKD |  |
| 2012 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F22 | 1 | 6y | 35y/M | PKD, Migraine | Seizure |
| 2012 | Gardiner AR | S | C647A | P216H | 2 | F23 | 1 | 4 | 9y/F | PKD |  |
| 2012 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F24 | n=3 | 6-14 | 12-42/3M | PKD | Migraine |
| 2012 | Gardiner AR | NA | 649dupC | R217PfsX8 | 2 | F25 | 1 | child | child/F | PKD | Attention deficit |
| 2012 | Gardiner AR | AD | C1011T | Splice site | 2 | F26 | 1 | 8y | 18/F | PKD |  |
|  |  |  |  |  |  |  | 2 | 12y | 18/F | PKD |  |
| 2012 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | F27 | 1 | child | teens/F | PKD |  |
| 2012 | Gardiner AR | S | G913T | G305W | 3 | F28 | 1 | child | teens/F | PKD |  |
| 2012 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F29 | n=2 | 6y | 34y/2M | PKD |  |
| 2012 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F30 | n=7 | 12y | 20y/(4M3F) | PKD | Early seizure and HM |
| 2012 | Marini C | AD | 649dupC | R217PfsX8 | 2 | F1 | 1 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 2 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 3 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 4 | 6-8m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 5 | 6-8m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 6 | 6-8m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 7 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 8 | 6-8m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 9 | - | NA/F | - |  |
|  |  |  |  |  |  |  | 10 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 11 | - | NA/F | - |  |
|  |  |  |  |  |  |  | 12 | 6-8m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 13 | 6-8m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 14 | 6-8m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 15 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 16 | 6-8m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 17 | 6-8m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 18 | - | NA/M | - |  |
| 2012 | Marini C | AD | G970A | G324R | 3 | F2 | 1 | 4m | 2-36y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA/4m | 2-36y/F | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | 4m | 2-36y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 4m | 2-36y/M | BFIE |  |
| 2012 | Marini C | AD | 649dupC | R217PfsX8 | 2 | F3 | 1 | 8m | 3y/F | BFIE | Childhood absence epilesy |
|  |  |  |  |  |  |  | 2 | 8m | 42y/F | BFIE |  |
| 2012 | Marini C | AD | 649dupC | R217PfsX8 | 2 | F4 | 1 | 7m | 2-32y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 7m | 2-32y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 7m | 2-32y/F | BFIE |  |
| 2012 | Marini C | S | 649dupC | R217PfsX8 | 2 | F5 | 1 | 6-8m | NA/F | BFIE |  |
| 2012 | Marini C | AD | G971A | G324E | 3 | F6 | 1 | 3.5-6m | 3-39y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 3.5-6m | 3-39y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 3.5-6m | 3-39y/F | BFIE |  |
| 2012 | Marini C | AD | 649dupC | R217PfsX8 | 2 | F8 | 1 | NA/6m | NA/F | BFIE | Migraine |
|  |  |  |  |  |  |  | 2 | NA/6m | NA/F | BFIE | Migraine |
|  |  |  |  |  |  |  | 3 | NA/6m | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 4 | NA/6m | NA/F | PKD/IC | Migraine |
|  |  |  |  |  |  |  | 5 | NA/6m | NA/F | PKD/IC | Migraine |
|  |  |  |  |  |  |  | 6 | NA/6m | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 7 | NA/6m | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 8 | NA/6m | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 9 | NA/6m | NA/M | PKD/IC | Migraine |
|  |  |  |  |  |  |  | 10 | NA/6m | NA/M | PKD/IC |  |
| 2012 | Marini C | AD | G916A | A306T | 3 | F9 | 1 | 7-8m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 7-8m | NA/F | BFIE |  |
| 2012 | Marini C | AD | 880-2A>T | Splice site | 3 | F10 | 1 | 7m | NA/F | BFIE | Migraine |
|  |  |  |  |  |  |  | 2 | 7m | NA/F | BFIE |  |
| 2012 | Marini C | AD | 649dupC | R217PfsX8 | 2 | F11 | 1 | 7m | 7-65y/F | BFIE | HM |
|  |  |  |  |  |  |  | 2 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 3 | 7m | 7-65y/M | BFIE | HM |
|  |  |  |  |  |  |  | 4 | 7m | 7-65y/F | BFIE | HM |
|  |  |  |  |  |  |  | 5 | 7m | 7-65y/M | BFIE | HM |
|  |  |  |  |  |  |  | 6 | 7m | 7-65y/F | BFIE | HM |
|  |  |  |  |  |  |  | 7 | 7m | 7-65y/M | BFIE |  |
|  |  |  |  |  |  |  | 8 | 7m | 7-65y/M | BFIE |  |
| 2012 | Lobate A | AD | 649dupC(homozygous) | R217PfsX8 | 2 | 1 | 1 | 4y/3m | 16y/M | PKD/IC | MR, absence epilepsy, ataxia |
|  |  |  |  |  |  |  | 2 | 2y/3m | 7y/M | PKD/IC | MR, absence epilepsy, ataxia |
|  |  |  |  |  |  |  | 3 | 4m | 13y/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | 7m | 44y/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | 6m | 47y/M | BFIE |  |
|  |  |  |  |  |  |  | 6 | - | 44y/F | - | Incomplete |
|  |  |  |  |  |  |  | 7 | 5m | 35yy/F | BFIE |  |
|  |  |  |  |  |  |  | 8 | - | 74/M | - |  |
| 2013 | Hedera P | AD | 776dupG | P260X | 2 | FA | 1 | 10y | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 10y | 22y/F | PKD |  |
|  |  |  |  |  |  |  | 3 | 12y | 28y/F | PKD |  |
|  |  |  |  |  |  |  | 4 | 10y | 25y/F | PKD |  |
|  |  |  |  |  |  |  | 5 | 13y | 18y/F | PKD |  |
| 2013 | Hedera P | AD | 649dupC | R217PfsX8 | 2 | FB | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | 12y | 30y/M | PKD |  |
| 2013 | Hedera P | AD | 649dupC | R217PfsX8 | 2 | Case 7 | 1 | 21y | 27y/M | PKD |  |
| 2013 | Wang K | AD | C649T | R217X | 2 | 1 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | puberty | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | - | 35y/M | - | Incomplete |
|  |  |  |  |  |  |  | 4 | 12y | 13y/M | PKD |  |
| 2013 | He YF | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | 7y | 26y/M | PKD | In Chinese |
|  |  |  |  |  |  |  | 2 | 7y | 53y/M | PKD | FS |
|  |  |  |  |  |  |  | 3 | 14y | 46y/M | PKD | FS |
|  |  |  |  |  |  |  | 4 | 14y | 17y/M | PKD |  |
| 2013 | Cai C | AD | C412G/C917A | P138A/A306D | 2/3 | 1 | 1 | 20y | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | 20y | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | 7y | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | 4y/4y | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 5 | <10m/<10m | 10m/F | PKD/IC |  |
|  |  |  |  |  |  |  | 6 | 10m/NA | NA/M | PKD/IC |  |
| 2013 | Ishii A | AD | 649dupC | R217PfsX8 | 2 | F1 | 1 | 5m | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | 16m | NA/F | IC |  |
| 2013 | Ishii A | AD | 649dupC | R217PfsX8 | 2 | F2 | 1 | 5-28m | NA/F | IC |  |
|  |  |  |  |  |  |  | 2 | 5-28m | NA/M | IC |  |
|  |  |  |  |  |  |  | 3 | 5-28m | NA/F | IC |  |
| 2013 | Ishii A | AD | 649dupC | R217PfsX8 | 2 | F3 | 1 | 4m | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | 4m | NA/M | IC |  |
| 2013 | Ishii A | AD | 649dupC | R217PfsX8 | 2 | F4 | 1 | 4m | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | 4m | NA/M | IC |  |
|  |  |  |  |  |  |  | 3 | 4m | NA/M | FS |  |
|  |  |  |  |  |  |  | 4 | 4m | NA/M | IC |  |
| 2013 | Ishii A | AD | 649dupC | R217PfsX8 | 2 | F5 | 1 | 5m | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | 5m | NA/M | IC |  |
|  |  |  |  |  |  |  | 3 | 5m | NA/M | IC |  |
| 2013 | Ishii A | AD | 649dupC | R217PfsX8 | 2 | F6 | 1 | 4-6m | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | 4-6m | NA/F | IC |  |
|  |  |  |  |  |  |  | 3 | 4-6m | NA/M | IC |  |
| 2013 | Ishii A | AD | 649dupC | R217PfsX8 | 2 | F7 | 1 | 5m | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | 5m | NA/M | IC |  |
|  |  |  |  |  |  |  | 3 | 5m | NA/F | IC |  |
|  |  |  |  |  |  |  | 4 | 5m | NA/M | IC |  |
| 2013 | Ishii A | AD | 649dupC | R217PfsX8 | 2 | F8 | 1 | 3-3.5m | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | 3-3.5m | NA/M | IC |  |
|  |  |  |  |  |  |  | 3 | 3-3.5m | NA/F | IC |  |
| 2013 | Ishii A | AD | 649dupC | R217PfsX8 | 2 | F9 | 1 | 3-4m | NA/F | IC |  |
|  |  |  |  |  |  |  | 2 | 3-4m | NA/F | IC |  |
|  |  |  |  |  |  |  | 3 | 3-4m | NA/M | IC |  |
|  |  |  |  |  |  |  | 4 | 3-4m | NA/M | IC |  |
|  |  |  |  |  |  |  | 5 | 3-4m | NA/M | IC |  |
|  |  |  |  |  |  |  | 6 | 3-4m | NA/M | FS |  |
|  |  |  |  |  |  |  | 7 | 3-4m | NA/M | IC |  |
| 2013 | Ishii A | AD | 649dupC | R217PfsX8 | 2 | F10 | 1 | 4m | NA/F | IC |  |
|  |  |  |  |  |  |  | 2 | 4m | NA/M | IC |  |
| 2013 | Ishii A | AD | 649dupC | R217PfsX8 | 2 | F11 | 1 | 4-9m | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | 4-9m | NA/M | IC |  |
|  |  |  |  |  |  |  | 3 | 4-9m | NA/F | IC |  |
| 2013 | Ishii A | AD | 649dupC | R217PfsX8 | 2 | F12 | 1 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 6.5m | NA/F | IC |  |
| 2013 | Ishii A | AD | 649dupC | R217PfsX8 | 2 | F13 | 1 | NA/4-5m | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | 4-5m | NA/F | IC |  |
| 2013 | Ishii A | AD | 649dupC | R217PfsX8 | 2 | F14 | 1 | NA/5m | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | 5m | NA/F | IC |  |
|  |  |  |  |  |  |  | 3 | 5m | NA/F | FS |  |
|  |  |  |  |  |  |  | 4 | 5m | NA/M | IC |  |
|  |  |  |  |  |  |  | 5 | 5m | NA/F | IC |  |
| 2013 | Ishii A | AD | 649dupC | R217PfsX8 | 2 | F15 | 1 | 3-4m | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | 3-4m | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | 3-4m | NA/F | IC |  |
| 2013 | Ishii A | AD | c.1012+2dupT | Splice site | 2 | F16 | 1 | 4m | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | 4m | NA/F | IC |  |
| 2013 | Okamura A | AD | C981G | I327M | 3 | F 1 | 1 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 10m | NA/F | BIE |  |
| 2013 | Okamura A | AD | 649dupC | R217PfsX8 | 2 | F 2 | 1 | NA/6m | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | 4m | NA/M | BIE |  |
|  |  |  |  |  |  |  | 3 | 3m | NA/F | BIE |  |
| 2013 | Okamura A | AD | 649dupC | R217PfsX8 | 2 | F 3 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | 8m | NA/M | BIE |  |
|  |  |  |  |  |  |  | 4 | 5m | NA/F | BIE |  |
| 2013 | Okamura A | AD | 649dupC | R217PfsX8 | 2 | F 4 | 1 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | NA/7m | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | NA/4m | NA/M | PKD/IC |  |
| 2013 | Okamura A | AD | 649dupC | R217PfsX8 | 2 | F 5 | 1 | NA | NA/F | FS |  |
|  |  |  |  |  |  |  | 2 | 3m | NA/F | BIE |  |
| 2013 | Okamura A | S | 649dupC | R217PfsX8 | 2 | F 6 | 1 | 5m | NA/M | BIE |  |
| 2013 | Li H | S | 649dupC | R217PfsX8 | 2 | Case 4 | 1 | 12y | 27y/M | PKD |  |
| 2013 | Li H | AD | 649dupC | R217PfsX8 | 2 | Case 7 | 1 | 18y | 20y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | - | NA/M | - | Incomplete |
| 2013 | Li H | AD | 133\_136delCCAG | P45RfsX44 | 2 | Case 8 | 1 | 3y | 17y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | - | NA/F | - | Incomplete |
| 2013 | Sheerin UM | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | NA | 85y/F | Migraine |  |
|  |  |  |  |  |  |  | 2 | 5y/6m | 52y/F | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | 31y | 61y/F | PKD | MWA |
|  |  |  |  |  |  |  | 4 | 5m | 58y/F | IC |  |
|  |  |  |  |  |  |  | 5 | 9y | 61y/F | PKD | MWA |
|  |  |  |  |  |  |  | 6 | 3y/6m | 15y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 7 | 10y/12m | 25y/F | PKD/IC | MWA |
|  |  |  |  |  |  |  | 8 | adolescence | 39y/M | MWA |  |
|  |  |  |  |  |  |  | 9 | 6m | 30y/F | IC | MWA |
|  |  |  |  |  |  |  | 10 | 13y | 28y/F | GTCS |  |
|  |  |  |  |  |  |  | 11 | 9y | 37y/M | PKD | MWA |
|  |  |  |  |  |  |  | 12 | NA | 36y/F | MWA |  |
|  |  |  |  |  |  |  | 13 | 9y | 27y/M | GTCS |  |
| 2013 | Castiglioni C | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | 20y’s | NA/F | MWA |  |
|  |  |  |  |  |  |  | 2 | 10y | 22y/M | PKD | MWA/MWOA |
|  |  |  |  |  |  |  | 3 | 10y | 22y/M | PKD | MWA |
| 2013 | Liu XR | AD | 579dupA | E194RfsX6 | 2 | F1 | 1 | 7y/5m | 42y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | 13y/5m | 40/F | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | 5y/5m | 34y/F | PKD/IC |  |
|  |  |  |  |  |  |  | 4 | 13y/6m | 22y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 5 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 6 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 7 | - | NA/M | - |  |
| 2013 | Liu XR | AD | 649dupC | R217PfsX8 | 2 | F2 | 1 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 17y/8m | 32y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | 20y/8m | 30y/F | PKD/IC |  |
|  |  |  |  |  |  |  | 4 | 9m/8m | 3y/F | IC/INCS |  |
| 2013 | Liu XR | AD | C872T | A291V | 2 | F3 | 1 | 7m | 38y/M | FC |  |
|  |  |  |  |  |  |  | 2 | 14y/7m | 15y/M | PKD/FC |  |
|  |  |  |  |  |  |  | 3 | - | NA/M | - |  |
| 2013 | Liu XR | AD | 649dupC | R217PfsX8 | 2 | F4 | 1 | 8y | 56y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 13y/7m | 26y/M | PKD/IC |  |
| 2013 | Liu XR | AD | 649dupC | R217PfsX8 | 2 | F5 | 1 | 6y/5m | 55y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | IC |  |
|  |  |  |  |  |  |  | 5 | 9y | 24y/M | PKD |  |
| 2013 | Liu XR | AD | 649dupC | R217PfsX8 | 2 | F6 | 1 | 20y | 48y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 13y/5m | 23y/M | PKD/NC |  |
| 2013 | Liu XR | AD | 649dupC | R217PfsX8 | 2 | F7 | 1 | 10y | 48y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 9y/10m | 22y/M | PKD/INCS |  |
| 2013 | Liu XR | AD | 649dupC | R217PfsX8 | 2 | F8 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 10y/12m | 24y/M | PKD/FC |  |
| 2013 | Liu XR | AD | C649T | R217X | 2 | F9 | 1 | NA | NA/M | IC |  |
|  |  |  |  |  |  |  | 2 | 15y/3m | 31y/M | PKD/IC | GTCS |
| 2013 | Liu XR | AR | 510dupT/C647G | L171SfsX3/P216R | 2 | F10 | 1 | 7y/3m | 14y/M | PKD/IC | INCS |
| 2013 | Liu XR | AD | 649dupC | R217PfsX8 | 2 | F11 | 1 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 6y/7m | 12y/M | PKD/IC |  |
| 2013 | Liu XR | S | 649dupC | R217PfsX8 | 2 | F12 | 1 | 11y | 20y/M | PKD |  |
| 2013 | Liu XR | S | 649dupC | R217PfsX8 | 2 | F13 | 1 | 5y/8m | 37y/M | PKD/FC |  |
| 2013 | Liu XR | S | 649dupC | R217PfsX8 | 2 | F14 | 1 | 8y | 31y/M | PKD |  |
| 2013 | Liu XR | S | 117delA | V41TfsX49 | 2 | F15 | 1 | 12y | 26y/M | PKD |  |
| 2013 | Becker F | S | 649dupC | R217PfsX8 | 2 | Dys6 | 1 | 10y | NA/M | PKD |  |
| 2013 | Becker F | AD | 649dupC | R217PfsX8 | 2 | Dys62 | 1 | 9y | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | - | NA/NA | - | Incomplete |
| 2013 | Becker F | S | 649dupC | R217PfsX8 | 2 | Dys36 | 1 | 6y | NA/M | PKD |  |
| 2013 | Becker F | AD | C919T | Q307X | 3 | Dys19 | 1 | 17y | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | - | NA/NA | - | Incomplete |
| 2013 | Becker F | AD | 388delG | A130PfsX46 | 2 | Dys39 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | 10y | NA/F | PKD |  |
| 2013 | Becker F | S | G884A | R295Q | 3 | Dys66 | 1 | 40y | NA/F | PKD | PNKD |
| 2013 | Becker F | AD | 291delC | N98TfsX17 | 2 | ICCA1 | 1 | 4m/7m | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | NA/5m | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | NA/6m | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 4 | NA/d3 | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 5 | NA/6m | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 6 | NA/6m | NA/F | PKD/IC |  |
| 2013 | Becker F | AD | 649dupC | R217PfsX8 | 2 | ICCA2 | 1 | 5m/d1 | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | NA/d10 | NA/F | PKD/IC |  |
| 2013 | Becker F | AD | 649dupC | R217PfsX8 | 2 | ICCA3 | 1 | 7y/8m | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | NA/6m | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | 9y/8m | NA/M | PKD/IC |  |
| 2013 | Becker F | AD | 649dupC | R217PfsX8 | 2 | ICCA4 | 1 | 6y/6m | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | NA/6m | NA/F | PKD/IC |  |
| 2013 | Becker F | AD | 649dupC | R217PfsX8 | 2 | ICCA5 | 1 | 7y/6m | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | 7y/5m | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | NA/5m | NA/F | PKD/IC |  |
| 2013 | Becker F | AD | C649T | R217X | 2 | ICCA6 | 1 | 5y/5m | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | NA/NA | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | Child/4m | NA/F | PKD/IC |  |
| 2013 | Becker F | AD | 649dupC | R217PfsX8 | 2 | ICCA7 | 1 | 4y/6m | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | - | NA/M | - | Incomplete |
| 2013 | Becker F | AD | 649dupC | R217PfsX8 | 2 | K1 | 1 | 4m | NA/M | seizure |  |
|  |  |  |  |  |  |  | 2 | 5m | NA/F | GTCS |  |
|  |  |  |  |  |  |  | 3 | 13y/6m | NA/F | PKD/IC | PNKD |
|  |  |  |  |  |  |  | 4 | Puberty/6m | NA/F | PKD/IC | PNKD |
|  |  |  |  |  |  |  | 5 | 5y/4m | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 6 | 6m | NA/F | IC |  |
|  |  |  |  |  |  |  | 7 | 4m | NA/M | IC |  |
| 2013 | Becker F | AD | 649dupC | R217PfsX8 | 2 | K7 | 1 | 17y | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | seizure |  |
|  |  |  |  |  |  |  | 3 | 8m | NA/M | seizure |  |
|  |  |  |  |  |  |  | 4 | 11y/3.5m | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 5 | 13y/6m | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 6 | 4m | NA/F | IC |  |
|  |  |  |  |  |  |  | 7 | 8.5m | NA/F | seizure |  |
| 2013 | Labate A | AD | 649dupC | R217PfsX8 | 2 | F1 | 1 | 5m | 43y/F | BFIE | FC |
|  |  |  |  |  |  |  | 2 | 8m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 4m | 21y/F | BFIE | FC |
| 2013 | Labate A | AD | 649dupC | R217PfsX8 | 2 | F2 | 1 | 3m | 30/M | BFIE | FC, MR |
|  |  |  |  |  |  |  | 2 | 4m | NA/M | BFIE | AFS |
| 2013 | Labate A | AD | C718T | R240X | 2 | F3 | 1 | 3m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 3m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 4m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | 5m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | 9m | NA/F | BFIE |  |
| 2013 | Zara F | AD | 291delC | N98TfsX17 | 2 | F11 | 1 | 6m | 41y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6m | 20y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 7m | 18y/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | 14m | 15y/M | FC |  |
|  |  |  |  |  |  |  | 5 | 5m | 12y/M | BFIE |  |
|  |  |  |  |  |  |  | 6 | 4m | 8y/M | BFIE |  |
| 2013 | Zara F | AD | 291delC | N98TfsX17 | 2 | F12 | 1 | NA | 44y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6m | 24y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 6m | 14y/M | BFIE |  |
| 2013 | Zara F | AD | 649dupC | R217PfsX8 | 2 | F13 | 1 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | 71y/M | NA |  |
|  |  |  |  |  |  |  | 3 | 6m | 33y/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | 5m | 7y/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | 6m | 3y/F | BFIE |  |
| 2013 | Zara F | AD | 649dupC | R217PfsX8 | 2 | F15 | 1 | 4m | 78y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 5m | 46y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 4m | 10y/F | BFIE |  |
| 2013 | Zara F | AD | 649dupC | R217PfsX8 | 2 | F16 | 1 | <1y | 67y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | <1y | 43y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 4m | 15y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 5m | 13y/F | BFIE |  |
| 2013 | Zara F | AD | 649dupC | R217PfsX8 | 2 | F17 | 1 | NA | 38y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6m | 34y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 5m | 10y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 5m | 6y/M | BFIE |  |
|  |  |  |  |  |  |  | 5 | 5m | 20y/M | BFIE |  |
| 2013 | Zara F | AD | 649dupC | R217PfsX8 | 2 | F18 | 1 | 5m | 11y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 5m | 7y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 4m | 7y/M | BFIE |  |
| 2013 | Zara F | AD | 649dupC | R217PfsX8 | 2 | F19 | 1 | <1y | 43y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6m | 8y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 6m | 7y/M | BFIE |  |
| 2013 | Zara F | AD | 649dupC | R217PfsX8 | 2 | F110 | 1 | 6m | 15y/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | 7m | 11y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 8m | 11y/M | BFIE |  |
| 2013 | Zara F | AD | 649dupC | R217PfsX8 | 2 | F114 | 1 | 5m | 6y/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | 5m | 5y/F | BFIE |  |
| 2013 | Zara F | AD | 649dupC | R217PfsX8 | 2 | F115 | 1 | 6m | 46y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 4m | 8y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 6m | 10y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 5m | 7y/M | BFIE |  |
| 2013 | Zara F | AD | 649dupC | R217PfsX8 | 2 | F122 | 1 | <1y | 71y/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | 13y/<1y | 37y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | 11y/7m | 13y/M | PKD/IC |  |
| 2013 | Zara F | AD | 649dupC | R217PfsX8 | 2 | F123 | 1 | 6m | 52y/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | 5m | 43y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | <1y | 41y/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | 6m | 23y/M | BFIE |  |
|  |  |  |  |  |  |  | 5 | 5.5m | 10y/M | BFIE |  |
|  |  |  |  |  |  |  | 6 | 6m | 8y/M | BFIE |  |
| 2013 | Zara F | AD | 649dupC | R217PfsX8 | 2 | F124 | 1 | NA | 91y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | 59y/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | NA | 41y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 6m | 34y/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | 8m | 13y/F | BFIE |  |
| 2013 | Zara F | AD | 649dupC | R217PfsX8 | 2 | F125 | 1 | NA | 92y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6m | 53y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 6m | 36y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 8m | 24y/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | 5m | 32y/F | BFIE |  |
|  |  |  |  |  |  |  | 6 | 6.5m | 8y/F | BFIE |  |
| 2013 | Zara F | AD | 649dupC | R217PfsX8 | 2 | F127 | 1 | 8m | 12y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 7m | 7y/F | BFIE |  |
| 2013 | Zara F | AD | 649dupC | R217PfsX8 | 2 | F128 | 1 | <1y | 32y/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6m | 7y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 6.5m | 3y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 4m | 1y/M | BFIE |  |
| 2013 | Zara F | AD | 649dupC | R217PfsX8 | 2 | F129 | 1 | 12y/4m | 40y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | 5m | 12y/M | BFIE |  |
| 2013 | Silveira-Moriyama L | S | G913T | G305W | 3 | F1 | 1 | 8y | 20y/F | PKD |  |
| 2013 | Silveira-Moriyama L | S | 649dupC | R217PfsX8 | 2 | F2 | 1 | 9y/4y | 16y/F | PKD/IC |  |
| 2013 | Silveira-Moriyama L | S | Microdeletion | Gene deletion | Gene | F3 | 1 | 5y | 19y/M | PKD |  |
| 2013 | Silveira-Moriyama L | AD | 649dupC | R217PfsX8 | 2 | F4 | 1 | 10y | 57y/F | PKD | HM |
|  |  |  |  |  |  |  | 2 | 11y | 35y/M | PKD | HM |
|  |  |  |  |  |  |  | 3 | NA | NA/F | GS |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | GS |  |
| 2013 | Silveira-Moriyama L | AD | 649dupC | R217PfsX8 | 2 | F5 | 1 | 7y | 66y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 10y | 16y/M | PKD |  |
| 2013 | Silveira-Moriyama L | AD | 649dupC | R217PfsX8 | 2 | F6 | 1 | 9y | 29y/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | Migraine |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | IC |  |
| 2013 | Silveira-Moriyama L | S | 649dupC | R217PfsX8 | 2 | F7 | 1 | 11y | 17y/F | PKD |  |
| 2013 | Silveira-Moriyama L | AD | C649T | R217X | 2 | F8 | 1 | 5y | 14y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 8y | 10y/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | IC |  |
| 2013 | Chen YC | AD | 649dupC | R217PfsX8 | 2 | F1 | n=6 | 1-19y | 27-57y/NA | PKD/IC |  |
| 2013 | Chen YC | AD | 601\_604delCACT | S202HfsX26 | 2 | F2 | n=3 | 1-17y | 25-54y/NA | PKD/IC | Incomplete |
| 2013 | Chen YC | AD | G529T | E177X | 2 | F3 | 1 | 12y/NA | 34y/NA | PKD/IC | Incomplete |
| 2013 | Chen YC | AD | 649dupC | R217PfsX8 | 2 | F4 | n=3 | 6-10y | 23-53y/NA | PKD | Incomplete |
| 2013 | Chen YC | AD | 649dupC | R217PfsX8 | 2 | F5 | n=3 | 11-12y/NA | 27-57y/NA | PKD/IC | Incomplete |
| 2013 | Chen YC | AD | 649dupC | R217PfsX8 | 2 | F7 | n=4 | 6-16y/NA | NA/NA | PKD/IC | Incomplete |
| 2013 | Chen YC | AD | 649dupC | R217PfsX8 | 2 | F9 | n=2 | 9-13y | 27-35y/NA | PKD | Incomplete |
| 2013 | Chen YC | AD | 649dupC | R217PfsX8 | 2 | F11 | 1 | 9y | 27y/NA | PKD | Incomplete |
| 2013 | Chen YC | AD | 649dupC | R217PfsX8 | 2 | S1 | 1 | 9y | 27/NA | PKD | Incomplete |
| 2013 | Shi CH | AD | 649dupC | R217PfsX8 | 2 | F1 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
| 2013 | Shi CH | AD | 649dupC | R217PfsX8 | 2 | F2 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
| 2013 | Shi CH | AD | 649dupC | R217PfsX8 | 2 | F3 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
| 2013 | Shi CH | S | 649dupC | R217PfsX8 | 2 | Case 1 | 1 | NA | NA/NA | PKD |  |
| 2013 | Shi CH | S | 649dupC | R217PfsX8 | 2 | Case 2 | 1 | NA | NA/NA | PKD |  |
| 2013 | Chen YP | S | 649dupC | R217PfsX8 | 2 | 2 | 1 | 12y | NA/NA | PKD |  |
| 2013 | Chen YP | S | 649dupC | R217PfsX8 | 2 | 25 | 1 | 15y | NA/NA | PKD |  |
| 2013 | Chen YP | S | 649dupC | R217PfsX8 | 2 | 30 | 1 | 13y | NA/NA | PKD |  |
| 2013 | Chen YP | S | 649dupC | R217PfsX8 | 2 | 32 | 1 | 10y | NA/NA | PKD |  |
| 2013 | Chen YP | S | 649dupC/C412G | R217PfsX8/P138A | 2 | 12 | 1 | 12y | NA/NA | PKD |  |
| 2013 | Chen YP | S | C981G | I327M | 2 | 13 | 1 | 13y | 15y/NA | PKD |  |
| 2013 | Chen YP | AD | 649dupC | R217PfsX8 | 2 | F 17 | 1 | 7.5y | NA/NA | PKD |  |
| 2013 | Chen YP | AD | 649dupC | R217PfsX8 | 2 | F 18 | 1 | 12y | NA/NA | PKD |  |
| 2013 | Chen YP | AD | 649dupC | R217PfsX8 | 2 | F 27 | 1 | 12y | NA/NA | PKD |  |
| 2013 | Chen YP | AD | 649dupC | R217PfsX8 | 2 | F 28 | 1 | 10y | NA/NA | PKD |  |
| 2013 | Chen YP | AD | C412G | P138A | 2 | F19 | 1 | 15y | NA/NA | PKD |  |
| 2013 | Chen YP | AD | G439C/G640C | D147H/A214P | 2 | F23 | 1 | 8y | NA/NA | PKD |  |
| 2013 | Jing XY | AD | 322delA | T108QfsX7 | 2 | Pedigree 1 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 7y | 23y/F | PKD |  |
| 2013 | Jing XY | AD | 649dupC | R217PfsX8 | 2 | Pedigree 2 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | 10y | 60y/M | PKD |  |
|  |  |  |  |  |  |  | 3 | 14y | 52y/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 5 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 6 | 9y | 32y/M | PKD |  |
|  |  |  |  |  |  |  | 7 | >16y | 24y/M | PKD |  |
| 2013 | Jing XY | AD | 649dupC | R217PfsX8 | 2 | Pedigree 3 | 1 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 17y | 24y/M | PKD |  |
|  |  |  |  |  |  |  | 3 | 16y | 22y/M | PKD |  |
| 2013 | Jing XY | AD | 649dupC | R217PfsX8 | 2 | Pedigree 4 | 1 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 5 | 11y | 21y/F | PKD |  |
| 2013 | Jing XY | AD | 649dupC | R217PfsX8 | 2 | Pedigree 5 | 1 | 10y | 40y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 11y | 13y/M | PKD |  |
| 2013 | Jing XY | AD | 649dupC | R217PfsX8 | 2 | Pedigree 6 | 1 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
| 2013 | Tan LCS | NA | 649dupC | R217PfsX8 | 2 | 1 | 1 | 10.9±2.8y | NA/NA | PKD | 12M3F  IC4 |
| 2013 | Tan LCS | NA | 649dupC | R217PfsX8 | 2 | 2 | 1 | 10.9±2.8y | NA/NA | PKD |
| 2013 | Tan LCS | NA | 649dupC | R217PfsX8 | 2 | 3 | 1 | 10.9±2.8y | NA/NA | PKD |
| 2013 | Tan LCS | NA | 649dupC | R217PfsX8 | 2 | 4 | 1 | 10.9±2.8y | NA/NA | PKD |
| 2013 | Tan LCS | NA | 649dupC | R217PfsX8 | 2 | 5 | 1 | 10.9±2.8y | NA/NA | PKD |
| 2013 | Tan LCS | NA | 649dupC | R217PfsX8 | 2 | 6 | 1 | 10.9±2.8y | NA/NA | PKD |
| 2013 | Tan LCS | NA | 649dupC | R217PfsX8 | 2 | 7 | 1 | 10.9±2.8y | NA/NA | PKD |
| 2013 | Tan LCS | NA | 649dupC | R217PfsX8 | 2 | 8 | 1 | 10.9±2.8y | NA/NA | PKD |
| 2013 | Tan LCS | NA | 649dupC | R217PfsX8 | 2 | 9 | 1 | 10.9±2.8y | NA/NA | PKD |
| 2013 | Tan LCS | NA | 649dupC | R217PfsX8 | 2 | 10 | 1 | 10.9±2.8y | NA/NA | PKD |
| 2013 | Tan LCS | NA | 649dupC | R217PfsX8 | 2 | 11 | 1 | 10.9±2.8y | NA/NA | PKD |
| 2013 | Tan LCS | NA | 649delC | R217EfsX12 | 2 | 12 | 1 | 10.9±2.8y | NA/NA | PKD |
| 2013 | Tan LCS | NA | C649T | R217X | 2 | 13 | 1 | 10.9±2.8y | NA/NA | PKD |
| 2013 | Tan LCS | NA | 604delT/609\_611delACC | S202HfsX27/S202Hfs10X | 2 | 14 | 1 | 10.9±2.8y | NA/NA | PKD, Intellectual disability |
| 2013 | Tan LCS | NA | 697\_698delAG | S233WfsX5 | 2 | 15 | 1 | NA | NA/NA | NA |
| 2013 | Fabbri M | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | NA | NA/F | Migraine |  |
|  |  |  |  |  |  |  | 2 | 12y | 85y/F | PKD |  |
|  |  |  |  |  |  |  | 3 | 9y | 43y/F | PKD |  |
|  |  |  |  |  |  |  | 4 | 10y | 53y/F | PKD | MWA |
|  |  |  |  |  |  |  | 5 | 6y | 17y/F | PKD |  |
| 2013 | Wang JL | AD | 649dupC | R217PfsX8 | 2 | A | 1 | 7m | 80y/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | 8m | 62y/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 6m | 57y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 4m | 56y/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | 6m | 40y/F | BFIE |  |
|  |  |  |  |  |  |  | 6 | 5m | 36y/M | BFIE |  |
|  |  |  |  |  |  |  | 7 | 6m | 30y/M | BFIE |  |
|  |  |  |  |  |  |  | 8 | 6m | 34y/F | BFIE |  |
|  |  |  |  |  |  |  | 9 | 3m | 37y/F | BFIE |  |
|  |  |  |  |  |  |  | 10 | 6m | 18y/M | BFIE |  |
|  |  |  |  |  |  |  | 11 | 6m | 14y/M | BFIE |  |
| 2013 | Wang JL | AD | 649dupC | R217PfsX8 | 2 | B | 1 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 10m | 70y/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 10m | 51y/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | 6m | 48y/M | BFIE |  |
|  |  |  |  |  |  |  | 6 | 6m | 21y/M | BFIE |  |
|  |  |  |  |  |  |  | 7 | 3m | 24y/F | BFIE |  |
| 2013 | Fusco C | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | 9y/6m | 10y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | childhood | NA/F | BFIE |  |
| 2013 | Weber C | S | Microdeletion | Gene deletion | Wholexon | 1 | 1 | 5y/infancy | NA/M | PKD/IC | Developmental delay |
| 2014 | Yang XL1 | AD | 649dupC | R217PfsX8 | 2 | 8 | 1 | - | NA/M | - | In Chinese |
|  |  |  |  |  |  |  | 2 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 4 | 4m | 7y/M | BFIE |  |
| 2014 | Yang XL1 | AD | 649dupC | R217PfsX8 | 2 | 11 | 1 | NA | NA/M | FC | In Chinese |
|  |  |  |  |  |  |  | 2 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 4m | 1y/M | BFIE |  |
| 2014 | Yang XL1 | AD | 649dupC | R217PfsX8 | 2 | 12 | 1 | NA | NA/M | FC | In Chinese |
|  |  |  |  |  |  |  | 2 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 5m | 1y/M | BFIE |  |
| 2014 | Yang XL1 | AD | 323\_324delCA | T108SfsX25 | 2 | 16 | 1 | NA | NA/M | BFIE | In Chinese |
|  |  |  |  |  |  |  | 2 | 4m | 9m/F | BFIE |  |
| 2014 | Torisu H | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | 6y | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 14m | NA/F | Seizure |  |
|  |  |  |  |  |  |  | 3 | 6m | NA/F | BFIE |  |
| 2014 | Djémié T | AD | 649dupC | R217PfsX8 | 2 | FA | 1 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 5-18m | NA/M | NA |  |
|  |  |  |  |  |  |  | 3 | 5-18m | NA/F | NIE |  |
|  |  |  |  |  |  |  | 4 | 5-18m | NA/M | FC |  |
|  |  |  |  |  |  |  | 5 | 5-18m | NA/M | FC |  |
|  |  |  |  |  |  |  | 6 | 5-18m | NA/M | FC |  |
|  |  |  |  |  |  |  | 7 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 8 | 5-18m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 9 | 5-18m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 10 | - | NA/F | - |  |
|  |  |  |  |  |  |  | 11 | 5-18m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 12 | 5-18m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 13 | 7m | NA/M | West syndrome |  |
|  |  |  |  |  |  |  | 14 | - | NA/F | - |  |
|  |  |  |  |  |  |  | 15 | 5-18m | NA/M | FC |  |
|  |  |  |  |  |  |  | 16 | 12y | NA/F | PKD |  |
|  |  |  |  |  |  |  | 17 | - | NA/F | - |  |
|  |  |  |  |  |  |  | 18 | 5-18m | NA/M | BFIE | NIE |
|  |  |  |  |  |  |  | 19 | 5-18m | NA/F | BFIE | NIE |
|  |  |  |  |  |  |  | 20 | 5-18m | NA/F | BFIE |  |
| 2014 | Djémié T | AD | 649dupC | R217PfsX8 | 2 | FB | 1 | 14y/6m | NA/F | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | 14y/6m | NA/M | PKD/IC |  |
| 2014 | Djémié T | S | 649dupC | R217PfsX8 | 2 | FC | 1 | 12y/5m | NA/F | PKD/IC |  |
| 2014 | Djémié T | AD | 649dupC | R217PfsX8 | 2 | FD | 1 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 6m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 7m | NA/F | BFIE |  |
| 2014 | Djémié T | AD | 649dupC | R217PfsX8 | 2 | FE | 1 | 4-8m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 4-8m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 4-8m | NA/M | BFIE |  |
| 2014 | Djémié T | AD | 649dupC | R217PfsX8 | 2 | FF | 1 | NA | NA/F | NA |  |
|  |  |  |  |  |  |  | 2 | 5m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 5.5m | NA/F | BFIE |  |
| 2014 | Djémié T | AD | 649dupC | R217PfsX8 | 2 | FG | 1 | 4-6m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 4-6m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 4-6m | NA/F | BFIE |  |
| 2014 | Djémié T | AD | 697\_698delAG | S233WfsX5 | 2 | FH | 1 | 6.5-8m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 6.6-8m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 6.5-8m | NA/M | BFIE |  |
| 2014 | Djémié T | AD | 879+1G>T | Splice site | Intron 2 | FI | 1 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 6m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 6m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | 6m | NA/M | BFIE |  |
| 2014 | Djémié T | AD | T1021C | X341fsX28 | 4 | FJ | 1 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 6-11m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | 6-11m | NA/M | BFIE |  |
| 2014 | Djémié T | AD | 649delC | R217EfsX12 | 2 | FK | 1 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 5m | NA/M | BFIE |  |
| 2014 | Djémié T | AD | 649dupC | R217PfsX8 | 2 | FL | 1 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 6m | NA/M | BFIE |  |
| 2014 | Djémié T | S | 649dupC | R217PfsX8 | 2 | FM | 1 | 7m | NA/F | BFIE |  |
| 2014 | Djémié T | AD | 649dupC | R217PfsX8 | 2 | FN | 1 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 5.5m | NA/F | BFIE |  |
| 2014 | Djémié T | AD | T1021C | X341fsX28 | 4 | FO | 1 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 7m | NA/F | BFIE |  |
| 2014 | Djémié T | AD | 649dupC | R217PfsX8 | 2 | FP | 1 | NA | NA/F | FC | NIE |
|  |  |  |  |  |  |  | 2 | NA | NA/M | FC |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | FC |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | FC |  |
|  |  |  |  |  |  |  | 5 | NA | NA/M | FC | NIE |
|  |  |  |  |  |  |  | 6 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 7 | NA | NA/M | PKD |  |
| 2014 | Yang X1 | AD | 649dupC | R217PfsX8 | 2 | F1 | 1 | <12m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | <12m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | <12m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | <12m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | <12m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 6 | 6m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 7 | <6m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 8 | <6m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 9 | <6m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 10 | 6m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 11 | 6m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 12 | 6m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 13 | 6m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 14 | 6m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 15 | 6m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 16 | <12m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 17 | 6m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 18 | <6m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 19 | 4.5m | 4.5y/F | BFIE |  |
| 2014 | Yang X1 | AD | 649dupC | R217PfsX8 | 2 | F2 | 1 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 2 | <12m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | <12m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | - | NA/F | - |  |
|  |  |  |  |  |  |  | 5 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 6 | <12m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 7 | 5m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 8 | 2m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 9 | <12m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 10 | <12m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 11 | 5m | 1.4y/F | BFIE |  |
| 2014 | Yang X1 | AD | 649dupC | R217PfsX8 | 2 | F3 | 1 | <12m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | <12m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | <12m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 4 | <12m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | <12m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 6 | <12m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 7 | <12m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 8 | <12m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 9 | <12m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 10 | 4m | 1y/F | BFIE |  |
| 2014 | Yang X1 | AD | 649dupC | R217PfsX8 | 2 | F4 | 1 | - | NA/F | - |  |
|  |  |  |  |  |  |  | 2 | 5m | NA/M | BFIE | Incomplete |
|  |  |  |  |  |  |  | 3 | 4.5m | 2.5y/F | BFIE |  |
| 2014 | Yang X1 | AD | 649dupC | R217PfsX8 | 2 | F5 | 1 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 6m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | 7m | 1.2/F | BFIE |  |
| 2014 | Yang X1 | AD | 649dupC | R217PfsX8 | 2 | F6 | 1 | <12m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 3 | <12m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | <12m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 5 | 6m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 6 | 4.5m | 43y/F | BFIE |  |
| 2014 | Yang X1 | AD | 649dupC | R217PfsX8 | 2 | F7 | 1 | 6m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | 2m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 3.5m | 54y/F | BFIE |  |
| 2014 | Yang X1 | AD | 649dupC | R217PfsX8 | 2 | F8 | 1 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 2 | <12m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 3.5m | 1.5y/M | BFIE |  |
| 2014 | Yang X1 | AD | 649dupC | R217PfsX8 | 2 | F9 | 1 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 5m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | 5m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | 5m | 5y/F | BFIE |  |
| 2014 | Yang X1 | AD | 649delC | R217EfsX12 | 2 | F10 | 1 | 4.5m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 3.5m | 6.33y/F | BFIE |  |
| 2014 | Yang X1 | AD | 649delC | R217EfsX12 | 2 | F11 | 1 | <12m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | <12m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 3 | 3m | 3.66y/M | BFIE |  |
| 2014 | Yang X1 | AD | 649delC | R217EfsX12 | 2 | F12 | 1 | 1m | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 2 | 4m | 1.25y/M | BFIE |  |
| 2014 | Yang X1 | AD | 904dupG | D302GfsX39 | 3 | F13 | 1 | 3m | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 2 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 3 | 6m | 3.5y/M | BFIE |  |
| 2014 | Yang X1 | AD | 649dupC | R217PfsX8 | 2 | F23 | 1 | Adolescence | 50y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 10y | 65y/F | PKD |  |
|  |  |  |  |  |  |  | 3 | 9y/<12m | 42y/F | PKD/IC |  |
|  |  |  |  |  |  |  | 4 | 8y/<12m | 38y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 5 | 10y/7m | 11y/M | PKD/IC |  |
| 2014 | Yang X1 | AD | 649dupC | R217PfsX8 | 2 | F24 | 1 | 7y/<12m | 34y/F | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | <12m | 33y/M | BIE |  |
|  |  |  |  |  |  |  | 3 | 7y/6m | 9y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 4 | 4.5m | 0.5y/M | BIE |  |
| 2014 | Yang X1 | AD | 649dupC | R217PfsX8 | 2 | F25 | 1 | 14y | 51y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 12y | 29y/F | PKD |  |
|  |  |  |  |  |  |  | 3 | 3.5m | 9m/M | BIE |  |
| 2014 | Yang X1 | AD | 649delC | R217EfsX12 | 2 | F26 | 1 | 11y | 42y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 8y/8m | 16y/F | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | 5m | 14y/F | BIE |  |
| 2014 | Yang X1 | AD | 649delC | R217EfsX12 | 2 | F27 | 1 | 15y/<12m | 50y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | 10y | 42y/F | PKD |  |
|  |  |  |  |  |  |  | 3 | 10y | 26y/F | PKD |  |
|  |  |  |  |  |  |  | 4 | 8y/5.5m | 13y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 5 | 10y | 22y/F | PKD |  |
|  |  |  |  |  |  |  | 6 | 4.5m | 3y/F | BIE |  |
| 2014 | Yang X1 | AD | 649delC | R217EfsX12 | 2 | F28 | 1 | 5y | 35y/F | PKD |  |
|  |  |  |  |  |  |  | 2 | 5.5y/4m | 12y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | 4m | 5y/F | BIE |  |
| 2014 | Yang X1 | AD | 514\_517delTCTG | S172RfsX3 | 2 | F29 | 1 | 16y/4.5m | 40y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | <12m | 36y/M | BIE |  |
|  |  |  |  |  |  |  | 3 | 3m | 5y/F | BIE |  |
| 2014 | Yang X1 | AD | A1023T | X341C | 3 | F30 | 1 | <12m | 83y/F | BIE |  |
|  |  |  |  |  |  |  | 2 | 3m | 50y/F | BIE |  |
|  |  |  |  |  |  |  | 3 | 12y/3m | 41y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 4 | 4m | 3.75y/M | BIE |  |
| 2014 | Youn J | AD | 649dupC | R217PfsX8 | 2 | FA | 1 | 11±2.8y/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | 11±2.8y | NA/M | PKD |  |
| 2014 | Youn J | AD | 649dupC | R217PfsX8 | 2 | FB | 1 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 11±2.8y | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | 11±2.8y | NA/M | PKD |  |
| 2014 | Youn J | AD | 649dupC | R217PfsX8 | 2 | FC | 1 | 11±2.8y | NA/M | PKD | MWOA |
|  |  |  |  |  |  |  | 2 | 11±2.8y | NA/M | PKD | Writers’s cramp |
| 2014 | Youn J | S | 649delC | R217EfsX12 | 2 | 1 | 1 | 11±2.8y/N | NA/M | PKD/IC |  |
| 2014 | Youn J | S | 629dupC | A211SfsX14 | 2 | 1 | 1 | 11±2.8y | NA/M | PKD |  |
| 2014 | Guo XN | AD | 649dupC | R217PfsX8 | 2 | F1 | 1 | 5y/NA | 29y/F | PKD/IC |  |
| 2014 | Guo XN | AD | 649dupC | R217PfsX8 | 2 | F3 | 1 | 9y | 21y/M | PKD |  |
| 2014 | Guo XN | AD | 649dupC | R217PfsX8 | 2 | F4 | 1 | 13y/NA | 23y/M | PKD/IC |  |
| 2014 | Guo XN | AD | 649dupC | R217PfsX8 | 2 | F5 | 1 | 5y | 23y/M | PKD |  |
| 2014 | Guo XN | AD | 1020insATAAG | X341KfsX31 | 4 | F7 | 1 | 13y | 40y/M | PKD |  |
| 2014 | Guo XN | AD | 324dupA | V109SfsX25 | 2 | F8 | 1 | 10y/NA | 21y/M | PKD/IC |  |
| 2014 | Guo XN | S | 649dupC | R217PfsX8 | 2 | S15 | 1 | 11y | 19y/M | PKD |  |
| 2014 | Guo XN | S | 649dupC | R217PfsX8 | 2 | S24 | 1 | 5y | 26y/M | PKD |  |
| 2014 | Guo XN | S | C981G | I327M | 3 | S26 | 1 | 12y | 14y/M | PKD |  |
| 2014 | Guo XN | S | 649delC | R217EfsX12 | 2 | S28 | 1 | 12y | 23y/F | PKD |  |
| 2014 | Mao CY | S | 649dupC | R217PfsX8 | 2 | 1 | 1 | 10y | NA/M | PKD |  |
| 2014 | Mao CY | AD | 649dupC | R217PfsX8 | 2 | 2 | 1 | 12y | NA/F | PKD |  |
| 2014 | Mao CY | AD | 649dupC | R217PfsX8 | 2 | 4 | 1 | 12y | NA/F | PKD |  |
| 2014 | Mao CY | S | 649dupC | R217PfsX8 | 2 | 9 | 1 | 11y | NA/M | PKD |  |
| 2014 | Mao CY | AD | 649dupC | R217PfsX8 | 2 | 11 | 1 | 12y | NA/M | PKD |  |
| 2014 | Mao CY | AD | 649dupC | R217PfsX8 | 2 | 12 | 1 | 4y | NA/M | PKD |  |
| 2014 | Mao CY | S | 649dupC | R217PfsX8 | 2 | 17 | 1 | 11y | NA/M | PKD |  |
| 2014 | Mao CY | S | 649dupC | R217PfsX8 | 2 | 18 | 1 | 12y | NA/F | PKD |  |
| 2014 | Mao CY | AD | 649dupC | R217PfsX8 | 2 | 20 | 1 | 20y | NA/M | PKD |  |
| 2014 | Mao CY | S | 649dupC | R217PfsX8 | 2 | 24 | 1 | 10y | NA/M | PKD |  |
| 2014 | Mao CY | S | 649dupC | R217PfsX8 | 2 | 25 | 1 | 12y | NA/M | PKD |  |
| 2014 | Mao CY | AD | 649dupC | R217PfsX8 | 2 | 26 | 1 | 11y | NA/M | PKD |  |
| 2014 | Mao CY | AD | 649dupC | R217PfsX8 | 2 | 27 | 1 | 14y | NA/M | PKD |  |
| 2014 | Mao CY | AD | 649dupC | R217PfsX8 | 2 | 30 | 1 | 8y | NA/M | PKD |  |
| 2014 | Mao CY | AD | 649dupC | R217PfsX8 | 2 | 32 | 1 | 9y | NA/M | PKD |  |
| 2014 | Mao CY | AD | 649dupC | R217PfsX8 | 2 | 33 | 1 | 8y | NA/M | PKD |  |
| 2014 | Brueckner F | AD | 649delC | R217EfsX12 | 2 | 1 | 1 | Infancy | 38y/M | FC |  |
|  |  |  |  |  |  |  | 2 | infancy | 39/F | FC |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | Headache |  |
|  |  |  |  |  |  |  | 4 | infancy | 15y/M | BFIE |  |
|  |  |  |  |  |  |  | 5 | 12y/6m | 16y/F | PKD/IC |  |
| 2014 | Matsumoto N | AD | C981G | I327M | 3 | 1 | 1 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 12y | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | 10y | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | - | NA/F | - |  |
|  |  |  |  |  |  |  | 5 | 6y | 24y/M | PKD |  |
|  |  |  |  |  |  |  | 6 | 7m | 7y/M | BFIE |  |
|  |  |  |  |  |  |  | 7 | 7m | 5y/M | BFIE |  |
| 2014 | Ji Z | AD | 186\_187delGC | P63QfsX70 | 2 | 1 | 1 | NA | 76/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 10y | 45y/F | PKD |  |
| 2014 | Guerrero-López R | AD | 649delC | R217EfsX12 | 2 | FA | 1 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 2 | - | NA/F | - |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 5 | infancy | 47y/F | NIE |  |
|  |  |  |  |  |  |  | 6 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 7 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 8 | - | NA/F | - |  |
|  |  |  |  |  |  |  | 9 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 10 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 11 | - | NA/- | - |  |
|  |  |  |  |  |  |  | 12 | Infancy | NA/F | BFIE | GTCS |
|  |  |  |  |  |  |  | 13 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 14 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 15 | 3m | 32y/M | FS |  |
|  |  |  |  |  |  |  | 16 | NA | NA/M | Other seizure |  |
|  |  |  |  |  |  |  | 17 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 18 | - | NA/M | - |  |
|  |  |  |  |  |  |  | 19 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 20 | NA | NA/F | BFIE |  |
| 2014 | Guerrero-López R | AD | 649delC | R217EfsX12 | 2 | FB | 1 | NA | NA/M | BFIE | FC |
|  |  |  |  |  |  |  | 2 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 3 | <12m | NA/F | FC |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | BFIE |  |
| 2014 | Guerrero-López R | AD | 649dupC | R217PfsX8 | 2 | FC | 1 | NA | NA/F | - | Incomplete |
|  |  |  |  |  |  |  |  | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | BFIE |  |
| 2014 | Guerrero-López R | AD | 649dupC | R217PfsX8 | 2 | FD | 1 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 2 | NA/Infancy | NA/M | PKD/IC | GTCS |
|  |  |  |  |  |  |  | 3 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | BFIE |  |
|  |  |  |  |  |  |  | 5 | NA | NA/F | PKD | NIE |
| 2014 | Guerrero-López R | AD | 121\_122delGT | V41YfsX93 | 2 | FE | 1 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 2 | NA/NA | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | 6m | 10y/M | NIE |  |
|  |  |  |  |  |  |  | 4 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 5 | NA | NA/M | BFIE |  |
|  |  |  |  |  |  |  | 6 | NA | NA/M | BFIE |  |
| 2014 | Li HF | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 11y/2m | 21y/M | PKD/FC |  |
| 2014 | Ebrahimi-Fakhari D | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | 15y | NA/F | PKD | Migraine |
|  |  |  |  |  |  |  | 2 | childhood | NA/F | PKD | Migraine |
| 2014 | Zhang LM | AD | 604\_607delTCAC | S202HfsX26 | 2 | 1 | 1 | 3y | 68y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 3y/24m | 41y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 4 | 7y/9m | 12y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 5 | 2y/12m | 3y/F | PKD/IC |  |
| 2014 | Yang X2 | AD | 649dupC | R217PfsX8 | 2 | F1 | 1 | <1y | 55y/F | BFIE | In Chinese |
|  |  |  |  |  |  |  | 2 | 11y | 51y/M | PKD |  |
|  |  |  |  |  |  |  | 3 | 13y | 41y/F | PKD |  |
|  |  |  |  |  |  |  | 4 | 12y | 39y/F | PKD |  |
|  |  |  |  |  |  |  | 5 | 12y/<1y | 29y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 6 | 11y/<1y | 28y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 7 | 12y | 29y/M | PKD |  |
|  |  |  |  |  |  |  | 8 | 12y | 19y/M | PKD |  |
|  |  |  |  |  |  |  | 9 | 5y | 17y/M | PKD |  |
|  |  |  |  |  |  |  | 10 | 5m | 10m/M | BFIE |  |
| 2014 | Yang X2 | AD | 649dupC | R217PfsX8 | 2 | 2 | 1 | 12m | 52y/M | BFIE | In Chinese |
|  |  |  |  |  |  |  | 2 | 6y/3m | 29y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | 4m | 1.25y/M | BFIE |  |
| 2014 | Yang X2 | AD | 649dupC | R217PfsX8 | 2 | 4 | 1 | 7y | 77y/M | PKD | In Chinese |
|  |  |  |  |  |  |  | 2 | 8y/3m | 47y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 3 | 13y | 43y/F | PKD |  |
|  |  |  |  |  |  |  | 4 | 10y | 40y/F | PKD |  |
|  |  |  |  |  |  |  | 5 | 11y/12m | 17y/M | PKD/IC |  |
| 2014 | Prabhakara S | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 2 | 17y | 27y/M | PKD |  |
| 2014 | Chou IC | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | 10y | 46y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 10y | 14y/M | PKD |  |
|  |  |  |  |  |  |  | 3 | 10y | 12y/M | PKD |  |
| 2014 | He ZW | S | C412G | P138A | 2 | 89 | 1 | 2y | 7y/M | FS+ |  |
| 2014 | He ZW | S | C623A | S208Y | 2 | 123 | 1 | 13y | 18y/F | FS+ |  |
| 2014 | He ZW | S | C412G | P138A | 2 | 174 | 1 | 2y | 16y/M | FS+ |  |
| 2014 | He ZW | S | C412G | P138A | 2 | 114 | 1 | 2y | 6y/M | FS+ |  |
| 2014 | He ZW | S | G439C | D147H | 2 | 192 | 1 | 1y | 13y/M | FS+ |  |
| 2014 | He ZW | S | C412G | P138A | 2 | 311 | 1 | 4y | 6y/M | FS+ |  |
| 2014 | He ZW | S | C412G | P138A | 2 | 313 | 1 | 3y | 6y/M | FS+ |  |
| 2014 | He ZW | AD | 649dupC | R217PfsX8 | 2 | SN540 | 1 | 5m | 18y/M | DS |  |
| 2014 | He ZW | S | 649dupC/G439C | R217PfsX8/D147H | 2 | 363 | 1 | 9y | 33y/M | FS+ |  |
| 2014 | He ZW | S | C412G | P138A | 2 | 737 | 1 | 4y | 7y/M | FS+ |  |
| 2014 | He ZW | S | C412G | P138A | 2 | 853 | 1 | 8y | 17y/M | FS+ |  |
| 2014 | He ZW | S | G439C | D147H | 2 | HH69 | 1 | 3y | 15y/M | FS+ |  |
| 2014 | He ZW | S | G439C | D147H | 2 | SN252 | 1 | 4y | 24y/M | FS+ |  |
| 2014 | He ZW | S | C412G | P138A | 2 | SN488 | 1 | 13y | 20y/F | FS+ |  |
| 2014 | He ZW | S | 649delC | R217EfsX12 | 2 | SN275 | 1 | 3y | 12y/F | FS+ |  |
| 2014 | He ZW | AD | C412G | P138A | 2 | 812 | 1 | 10y | 32y/M | GEFS+ |  |
| 2014 | He ZW | AD | C412G | P138A | 2 | 1232 | 1 | 2y | 15y/M | GEFS+ |  |
| 2014 | He ZW | AD | 649dupC | R217PfsX8 | 2 | SN854 | 1 | 8y | 36y/M | GEFS+ |  |
| 2014 | He ZW | S | G439C | D147H | 2 | 576 | 1 | 5m | 5y/F | DS |  |
| 2014 | He ZW | S | C412G | P138A | 2 | 1186 | 1 | 8m | 10y/F | DS |  |
| 2014 | He ZW | S | C412G | P138A | 2 | 872 | 1 | 9m | 3y/F | DS |  |
| 2014 | He ZW | S | 649delC | R217fsX12 | 2 | SN676 | 1 | 5m | 32y/M | DS |  |
| 2014 | He ZW | S | C412G | P138A | 2 | SN740 | 1 | 40d | 25y/F | DS |  |
| 2014 | He ZW | AD | 649dupC | R217PfsX8 | 2 | SN540 | 1 | 5m | 18y/M | DS |  |
| 2014 | He ZW | S | C412G | P138A | 2 | 428 | 1 | 11m | 12y/M | DS |  |
| 2015 | Yang X | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | 4-13y | NA/F | PKD | In Chinese |
|  |  |  |  |  |  |  | 2 | 4-13y | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 4 | 4-13y | NA/F | PKD |  |
|  |  |  |  |  |  |  | 5 | 4-13y | NA/M | PKD |  |
|  |  |  |  |  |  |  | 6 | 4-13y | NA/M | PKD |  |
|  |  |  |  |  |  |  | 7 | 10y | 27y/M | PKD |  |
| 2015 | Yang X | AD | 649dupC | R217PfsX8 | 2 | 2 | 1 | 4-13y | NA/F | PKD | In Chinese |
|  |  |  |  |  |  |  | 2 | 4y | 15y/M | PKD |  |
| 2015 | Yang X | AD | 649dupC | R217PfsX8 | 2 | 3 | 1 | 4-13y | NA/M | PKD | In Chinese |
|  |  |  |  |  |  |  | 2 | 13y | 36y/M | PKD |  |
| 2015 | Yang X | AD | 649dupC | R217PfsX8 | 2 | 4 | 1 | 4-13y | NA/M | PKD | In Chinese |
|  |  |  |  |  |  |  | 2 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 3 | - | NA/F | - |  |
|  |  |  |  |  |  |  | 4 | 4y | 30y/M | PKD |  |
| 2015 | Yang X | AD | 649dupC | R217PfsX8 | 2 | NA | 1 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 4-13y | NA/M | PKD | In Chinese |
| 2015 | Yang X | AD | 649dupC | R217PfsX8 | 2 | NA | 1 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 4-13y | NA/M | PKD | In Chinese |
| 2015 | Delcourt M | S | 649dupC/Microdeletion | R217PfsX8/gene deletion | 2 | Patient 1 | 1 | 6m/1.5y | 15y/M | AFS/EA | HM |
| 2015 | Delcourt M | AD | 649dupC/649dupC | R217PfsX8 | 2 | Patient 2 | 1 | 25m/3m | 20y/M | PKD/IC | HM, Learning difficulty |
| 2015 | Delcourt M | AD | 649dupC/649dupC | R217PfsX8 | 2 | Patient 3 | 1 | 3m/6y | 15y/F | AFS/EA |  |
| 2015 | Delcourt M | AD | 649dupC/649dupC | R217PfsX8 | 2 | Patient 4 | 1 | child/3m/7y | 25y/F | PNKD/Seizure/EA | Migraine |
| 2015 | Delcourt M | AD | G913A/G913A | G305R | 3 | Patient 2 | 1 | 3m | 5.5y/F | Seizure | EA |
| 2015 | Termsarasab | S | Microdeletion | deletion | Gene | 1 | 1 | 10.5y | 12y/M | PKD |  |
| 2015 | Chen GH | AD | 649dupC | R217PfsX8 | 2 | F1 | 1 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 10y | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | 9y | NA/M | PKD |  |
| 2015 | Chen GH | AD | 649dupC | R217PfsX8 | 2 | F2 | 1 | 10y | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 8y | NA/F | PKD |  |
| 2015 | Chen GH | AD | 133\_136delCCAG | P45RfsX44 | 2 | F3 | 1 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 5y | NA/M | PKD |  |
| 2015 | Lamperti C | S | 649dupC | R217PfsX8 | 2 | F1 | 1 | 5y/NA | 19y/F | PKD/IC |  |
| 2015 | Lamperti C | AD | 27insT | S9X | 2 | F2 | 1 | 17y | 20y/F | PKD |  |
|  |  |  |  |  |  |  | 2 | - | NA/M | - | Incomplete |
| 2015 | Lamperti C | AD | G967A | G323R | 3 | F3 | 1 | 12y | 16y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | 12y | NA/M | PKD |  |
| 2015 | Lamperti C | AD | 215\_216delCA | Y72RfsX62 | 2 | F4 | 1 | 8y | 11y/M | PKD | Migraine |
|  |  |  |  |  |  |  | 2 | 12y | NA/M | PKD |  |
| 2015 | Lamperti C | S | 649dupC | R217PfsX8 | 2 | F5 | 1 | 7.5y | 19y/F | PKD | epilepsy |
|  |  |  |  |  |  |  | 2 | 11y | 17y/F | PKD | epilepsy |
|  |  |  |  |  |  |  | 3 | infancy | NA/M | PKD |  |
| 2015 | Lamperti C | S | 649dupC | R217PfsX8 | 2 | F6 | 1 | 9y/NA | 11y/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | 13y/NA | NA/F | PKD/IC |  |
| 2015 | Lamperti C | S | 649dupC | R217PfsX8 | 2 | F7 | 1 | 14y | 17y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
| 2015 | Lamperti C | S | 649dupC | R217PfsX8 | 2 | F8 | 1 | 10y | NA/F | PKD |  |
| 2015 | Lamperti C | S | 649dupC | R217PfsX8 | 2 | F9 | 1 | 8y | 29y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | - | NA/F | - | Incomplete |
| 2015 | Lamperti C | S | 649dupC | R217PfsX8 | 2 | F10 | 1 | 12y | 34y/F | PKD |  |
|  |  |  |  |  |  |  | 2 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 3 | 13y | NA/F | PKD |  |
| 2015 | Huang XJ | S | C787T | Q263X | 2 | FA | 1 | NA | NA/M | PKD |  |
| 2015 | Huang XJ | AD | G797A | R266Q | 2 | FB | 1 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
| 2015 | Huang XJ | S | C931T/C931T | R311W/R311W | 3 | PA0234 | 1 | 2y/7m | 25y/F | PKD/IC | Uniparental disomy |
| 2015 | Gardiner AR | AD | 510dupT | L171LfsX3 | 2 | F1 | n=2 | 12-13y | 24-27y/1M1F | PKD | Seizure |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F2 | n=2 | 7-8y | 12-16y/1M1F | PKD | Incomplete |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F3 | n=2 | 0.5-27y | 29-51y/1M1F | PKD | Incomplete |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F4 | n=4 | 6-11y | 18-49y/4M | PKD | MWA |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F5 | n=2 | 8y | 42-45y/1M1F | PKD |  |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F6 | n=2 | 1-6y | 12-62y/1M1F | PKD | HM, Migraine |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F7 | n=2 | 0.5-8y | 12-52y/2M2F | BFIE | HM |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F8 | n=2 | 14y | 31-33y/1M1F | PKD |  |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F9 | n=4 | 4-10y | 12-56y/2M2F | PKD |  |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F10 | n=4 | 6-11y | 20-68y/2M2F | PKD | Migraine |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F11 | n=2 | 6-16y | 8-38y/1M1F | PKD | Migraine |
| 2015 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | S12 | 1 | 5y | 16y/1M | PKD |  |
| 2015 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | S13 | 1 | 12y | 18y/1F | PKD |  |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F14 | n=3 | 10y | 12-59y/1M2F | HM | EA |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F15 | n=2 | 8y | 40-42y/2M | PKD | MWA |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F16 | n=2 | 8-18y | 25-48y/1M1F | PKD | Migraine |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F17 | n=2 | 8y | 43y/2M | PKD | Headache |
| 2015 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | S18 | 1 | 27y | 48y/M | PKD |  |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F19 | n=2 | 9-12y | 42-47y/1M1F | PKD | Seizure |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F20 | n=3 | 6-14y | 12-42y/3M | PKD | Seizure,Migraine |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F21 | n=6 | 0.5-30y | 87y/3M3F | PKD | Incomplete, Migraine, HM, Epilepsy |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F22 | n=2 | 12-18y | 14-39y/1M1F | PKD, Migraine |  |
| 2015 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | S23 | 1 | 14y | 39y/M | PKD |  |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F24 | n=15 | 7-13y | 9-60y/9M6F | PKD, Seizure | Incomplete |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F25 | n=3 | 8y | 12-52y/2M1F | PKD, Migraine | Seizure |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F26 | n=13 | 8-12y | 28-76y/5M8F | PKD | HM, Migraine |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F27 | n=2 | 6-7y | 9-12y/2F | PKD | HM, Migraine |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F28 | n=3 | 6-8y | 12-49y/1M2F | PKD |  |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F29 | 1 | 9y/NA | 32y/F | PKD/IC | Migraine |
| 2015 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | S30 | 1 | 9y | 19y/1F | PKD |  |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F31 | 1 | 11y | 20y/1F | PKD |  |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F32 | n=2 | 10y | 18-69y/2M | PKD |  |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F33 | n=2 | 6-12y/NA | 31-59y/1M1F | PKD/IC | HM |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F34 | n=2 | 0.5-5y/NA | 8-40y/2M1F | PKD/IC, HM | Incomplete |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F35 | n=2 | 0.5y | 2y/2M | IC | PKD |
| 2015 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | S36 | 1 | 39y | 63y/M | PKD | EA with dysarthria |
| 2015 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | S37 | 1 | 3y | 8y/M | PKD |  |
| 2015 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | S38 | 1 | 12y | 29y/M | PKD |  |
| 2015 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | S39 | 1 | 6y | 16y/M | PKD |  |
| 2015 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | S40 | 1 | 8y | 15y/M | PKD | HM |
| 2015 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | S41 | 1 | 8y | 27y/M | PKD |  |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F 42 | 1 | 5y | 10y/2M | PKD | HM |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F 43 | n=2 | 7-14y | 22-49y/1M1F | PKD |  |
| 2015 | Gardiner AR | S | 649dupC | R217PfsX8 | 2 | S44 | 1 | 14y | 33y/M | PKD |  |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F 45 | n=2 | 7-12y | 9-32y/2F | PKD | HM |
| 2015 | Gardiner AR | AD | 649dupC | R217PfsX8 | 2 | F 46 | n=2 | 8-14y | 12-37y/1M1F | PKD |  |
| 2015 | Gardiner AR | S | C718T | R240X | 2 | S47 | 1 | 11y | 21y/M | PKD |  |
| 2015 | Gardiner AR | S | G913T | G305W | 3 | S48 | 1 | NA | 23y/1F | PKD |  |
| 2015 | Gardiner AR | AD | 997\_998insATG | C332\_V333insD | 3 | F49 | n=2 | 14y | 46y/2M | PKD | Seizure, Migraine |
| 2015 | Gardiner AR | AD | C1011T | splice site | 3 | F50 | 1 | 2y | 15y/F | HM | Benign Seizure |
| 2015 | Gardiner AR | AD | 1020\_1021insG | X341Lext27 | 4 | F51 | n=2 | 12-18y | 16-35y/2M2F | PKD | MWA |
| 2016 | Le X | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | - | NA/F | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 7.5y | 10.75y/M | PKD | In Chinsese |
| 2016 | Bi GH | AD | G797A | R266Q | 2 | 1 | 1 | - | NA/M | - | Incomplete |
|  |  |  |  |  |  |  | 2 | 17y | 43y/M | PKD | In Chinese |
|  |  |  |  |  |  |  | 3 | 13y | 18y/M | PKD |  |
| 2016 | Zhang H | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | 6y | 48y/M | PKD | In Chinese |
|  |  |  |  |  |  |  | 2 | 2y | 26y/M | PKD |  |
| 2016 | Igarashi A | AD | T841C | W281R | 2 | 1 | 1 | Infancy | NA/F | IC |  |
|  |  |  |  |  |  |  | 2 | Infancy | NA/F | IC |  |
|  |  |  |  |  |  |  | 3 | childhood/Infancy | NA/M | PKD/IC |  |
|  |  |  |  |  |  |  | 4 | 5m | 5y/F | BIE | Down’s syndrome |
|  |  |  |  |  |  |  | 5 | 5m | 4y/M | BIE |  |
| 2014 | Zheng W | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | 9m | 66y/F | FS |  |
|  |  |  |  |  |  |  | 2 | 12m | 45y/M | FS |  |
|  |  |  |  |  |  |  | 3 | 7m | 43y/M | FS |  |
|  |  |  |  |  |  |  | 4 | 11m | 24y/M | FS |  |
| 2016 | Maini I | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | 8m/1m | 16m/M | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | <12m | NA/M | PKD |  |
| 2016 | Weber A | AD | 880-35G>A | S294LfsX29 | Intron 2 | 1 | 1 | 6m | 18m/F | IC |  |
|  |  |  |  |  |  |  | 2 | 9y | 39y/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
| 2016 | Trump N | NA | 649dupC | R217PfsX8 | 2 | 1 | 1 | NA | NA/M | PKD |  |
| 2016 | Trump N | NA | T1021C | X341fsX28 | 4 | 1 | 1 | NA | NA/F | IC |  |
| 2016 | Liu XR | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | 10m | 24y/M | PHD | GTCS |
|  |  |  |  |  |  |  | 2 | 12y | NA/M | PKD |  |
| 2016 | LiuXR | AD | G640C | A214P | 2 | 1 | 1 | 2y | 18y/M | PHD | GTCS |
|  |  |  |  |  |  |  | 2 | - | NA/F | - | Incomplete |
| 2016 | Castelnovo G | AD | 224delC | P75RfsX15 | 2 | 1 | 1 | childhood | 44y/M | IC | Ataxia |
|  |  |  |  |  |  |  | 2 | NA | NA/F | Epilepsy |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | Epilepsy |  |
| 2016 | Law CY | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | childhood | NA/F | seizure |  |
|  |  |  |  |  |  |  | 2 | childhood | NA/F | seizure |  |
|  |  |  |  |  |  |  | 3 | childhood | 47y/F | PKD |  |
|  |  |  |  |  |  |  | 4 | 10m | NA/F | IC |  |
|  |  |  |  |  |  |  | 5 | childhood | NA/M | seizure |  |
| 2016 | Law CY | AD | 649dupC | R217PfsX8 | 2 | 2 | 1 | 8y/7m | 26y/F | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/M | PKD |  |
| 2016 | Law CY | AD | 649dupC | R217PfsX8 | 2 | 3 | 1 | 1.5y | NA/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | NA/NA | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/NA | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/NA | PKD |  |
|  |  |  |  |  |  |  | 5 | NA | NA/NA | PKD |  |
|  |  |  |  |  |  |  | 6 | NA | NA/NA | PKD |  |
| 2016 | Law CY | AD | 649dupC | R217PfsX8 | 2 | 4 | 1 | 9y | 33y/M | PKD | Migraine |
|  |  |  |  |  |  |  | 2 | 7m | NA/F | IC |  |
|  |  |  |  |  |  |  | 3 | 3m | NA/F | IC |  |
| 2016 | Law CY | S | 649dupC | R217PfsX8 | 2 | 5 | 1 | 9y | 14y/M | PKD |  |
| 2016 | Law CY | AD | 379delG | Q127SfsX49 | 2 | 6 | 1 | 29y | 3y/M | PKD |  |
|  |  |  |  |  |  |  | 2 | NA | Teenage/M | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/F | PKD |  |
| 2016 | Seo SY | AD | 649dupC | R217PfsX8 | 2 | 1 | 1 | 10y/6m | 7y/F | PKD/IC |  |
|  |  |  |  |  |  |  | 2 | NA | NA/F | PKD |  |
|  |  |  |  |  |  |  | 3 | NA | NA/M | PKD |  |
|  |  |  |  |  |  |  | 4 | NA | NA/F | PKD |  |

†mean value; AAO: age at onset; d: days; m, months; y: years; NA: not available; M: Male; F: Female; AD: autosomal dominant; AR: autosomal recessive; S: Sporadic; PKD: paroxysmal kinesigenic dyskinesia; PNKD: paroxysmal non-kinesigenic dyskinesia; PED: paroxysmal exercise-induced dyskinesia; PHD: paroxysmal hypnogenic dyskinesia; IC: infantile convulsion; PKD/IC: infantile convulsions with paroxysmal choreoathetosis; BFIE: benign familial infantile epilepsy; BIE: benign infantile epilepsy; BFNC: benign familial neonatal convulsion; GTCS: general tonic clonus seizure; FS: febrile seizures; FS+: febrile seizures plus; AFS: afebrile seizures; GEFS+: genetic epilepsy with febrile seizures plus; NIE: No n-infantile epilepsy; HM: hemiplegic migraine; CAE: childhood absence epilepsy; DS: Dravet syndrome; GS: generalized seizure; MWA: Migraine with aura; MWOA: Migraine without aura; INCS: infantile nonconvulsive seizure; NC: nocturnal convulsion; MR: mental retardation; EA: episodic ataxia.