

Supplementary Table 1: Summary of all reported CSF1R mutations, exons, age at onset and age at death.

| Author | Year | Mutation | Exon | Age at onset | Age at death | Disease duration |
|-------------------------|------|------------------------|-------|--------------|--------------|------------------|
| Rademakers ¹ | 2011 | G589E | 13 | 47 | 58 | 11 |
| Rademakers | 2011 | G589E | 13 | 58 | 61 | 3 |
| Rademakers | 2011 | E633K | 14 | 42 | 46 | 4 |
| Rademakers | 2011 | E633K | 14 | 67 | 74 | 7 |
| Rademakers | 2011 | E633K | 14 | 78 | 84 | 6 |
| Rademakers | 2011 | M766T | 17 | 18 | N/A | N/A |
| Rademakers | 2011 | M766T | 17 | 43 | N/A | N/A |
| Rademakers | 2011 | I755N | 17 | 48 | N/A | N/A |
| Rademakers | 2011 | A770P | 17 | 52 | 63 | 11 |
| Rademakers | 2011 | I794T | 18 | 35 | N/A | N/A |
| Rademakers | 2011 | F849del | 19 | 63 | 67 | 4 |
| Rademakers | 2011 | D837Y | 19 | N/A | N/A | N/A |
| Rademakers | 2011 | F849S | 19 | N/A | N/A | N/A |
| Rademakers | 2011 | P878T | 20 | 39 | 49 | 10 |
| Rademakers | 2011 | P878T | 20 | 39 | 43 | 4 |
| Rademakers | 2011 | M875T | 20 | 41 | N/A | N/A |
| Rademakers | 2011 | M875T | 20 | 46 | N/A | N/A |
| Rademakers | 2011 | L868P | 20 | 55 | 63 | 8 |
| Rademakers | 2011 | M875T | 20 | 58 | N/A | N/A |
| Rademakers | 2011 | M875T | 20 | 71 | N/A | N/A |
| Rademakers | 2011 | M875T | 20 | N/A | N/A | N/A |
| Rademakers | 2011 | Gly585_lys619delinsAla | IVS12 | 36 | 40 | 4 |
| Rademakers | 2011 | Gly585_lys619delinsAla | IVS12 | 38 | 41 | 3 |
| Rademakers | 2011 | C774_N814delins | IVS18 | 23 | N/A | N/A |
| Rademakers | 2011 | cys774_asn814del | IVS18 | 50 | 55 | 5 |
| Kinoshita ² | 2012 | R782H | 18 | 51 | N/A | N/A |
| Kleinfeld ³ | 2013 | I794T | 18 | 37 | N/A | N/A |
| Kleinfeld | 2013 | F828S | 19 | 39 | N/A | N/A |
| Kleinfeld | 2013 | F828S | 19 | 47 | N/A | N/A |
| Kondo ⁴ | 2013 | K793T | 18 | 40 | N/A | N/A |
| Mitsui ⁵ | 2012 | C653Y | 14 | 43 | N/A | N/A |
| Mitsui | 2012 | C653Y | 14 | 48 | N/A | N/A |
| Mitsui | 2012 | R777W | 18 | 38 | N/A | N/A |
| Mitsui | 2012 | I794T | 18 | 52 | N/A | N/A |
| Mitsui | 2012 | I794T | 18 | 60 | N/A | N/A |
| Mitsui | 2012 | R777W | 18 | 65 | N/A | N/A |
| Ahmed ⁶ | 2013 | A781V | 18 | 32 | N/A | N/A |
| Ahmed | 2013 | A781V | 18 | 54 | N/A | N/A |

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|-------------------------|------|------------|-------|-----|-----|-----|
| Ahmed | 2013 | A781V | 18 | 58 | N/A | N/A |
| Ahmed | 2013 | A781V | 18 | 60 | N/A | N/A |
| Ahmed | 2013 | A781V | 18 | 67 | N/A | N/A |
| Fujioka ⁷ | 2013 | G589E | 13 | 46 | N/A | N/A |
| Fujioka | 2013 | G589E | 13 | 47 | N/A | N/A |
| Fujioka | 2013 | G589E | 13 | 58 | N/A | N/A |
| Fujioka | 2013 | G589E | 13 | 59 | N/A | N/A |
| Guerreiro ⁸ | 2013 | T567fsX44 | 12 | 24 | N/A | N/A |
| Guerreiro | 2013 | L630R | 14 | 45 | N/A | N/A |
| Guerreiro | 2013 | E633K | 14 | 46 | N/A | N/A |
| Guerreiro | 2013 | R777Q | 18 | 44 | N/A | N/A |
| Guerreiro | 2013 | R777W | 18 | 46 | N/A | N/A |
| Guerreiro | 2013 | I794T | 18 | 46 | N/A | N/A |
| Guerreiro | 2013 | R777Q | 18 | 58 | N/A | N/A |
| Guerreiro | 2013 | R777Q | 18 | 60 | N/A | N/A |
| Guerreiro | 2013 | L817P | 19 | 36 | N/A | N/A |
| Guerreiro | 2013 | I827T | 19 | 44 | N/A | N/A |
| Guerreiro | 2013 | E847D | 19 | 44 | N/A | N/A |
| Guerreiro | 2013 | Y856H | 20 | 39 | N/A | N/A |
| Guerreiro | 2013 | Y856H | 20 | 42 | N/A | N/A |
| Guerreiro | 2013 | Y856H | 20 | 42 | N/A | N/A |
| Guerreiro | 2013 | Y856H | 20 | 47 | N/A | N/A |
| Guerreiro | 2013 | Y856H | 20 | 55 | N/A | N/A |
| Guerreiro | 2013 | P901S | 21 | 20 | N/A | N/A |
| Guerreiro | 2013 | c2655-2A>G | IVS20 | 50 | N/A | N/A |
| Inui ⁹ | 2013 | A777Gly | 18 | 24 | N/A | N/A |
| Karle ¹⁰ | 2013 | I794T | 18 | 29 | N/A | N/A |
| Karle | 2013 | R777Q | 18 | 38 | N/A | N/A |
| Karle | 2013 | I794T | 18 | 40 | N/A | N/A |
| Karle | 2013 | A781V | 18 | 50 | N/A | N/A |
| Karle | 2013 | I843N | 19 | 40 | N/A | N/A |
| Karle | 2013 | V838L | 19 | 53 | N/A | N/A |
| Karle | 2013 | Q877X | 20 | 28 | N/A | N/A |
| Nicholson ¹¹ | 2013 | M766T | 17 | 51 | N/A | N/A |
| Nicholson | 2013 | R782H | 18 | N/A | N/A | N/A |
| Saitoh ¹² | 2013 | I794T | 18 | 28 | N/A | N/A |
| Sundal ¹³ | 2013 | G589E | 13 | 58 | N/A | N/A |
| Sundal | 2013 | E633K | 14 | 42 | N/A | N/A |
| Sundal | 2013 | E633K | 14 | 67 | N/A | N/A |
| Sundal | 2013 | M766T | 17 | 18 | N/A | N/A |
| Sundal | 2013 | M766T | 17 | 27 | N/A | N/A |

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|----------------------------|------|------------------------|-------|-----|-----|-----|
| Sundal | 2013 | M766T | 17 | 43 | N/A | N/A |
| Sundal | 2013 | I755Asn | 17 | 48 | N/A | N/A |
| Sundal | 2013 | A770P | 17 | 52 | N/A | N/A |
| Sundal | 2013 | I794T | 18 | 35 | N/A | N/A |
| Sundal | 2013 | I794T | 18 | 57 | N/A | N/A |
| Sundal | 2013 | M875T | 20 | 58 | N/A | N/A |
| Sundal | 2013 | M875T | 20 | 71 | N/A | N/A |
| Sundal | 2013 | Gly585_lys619delinsAla | IVS12 | 36 | N/A | N/A |
| Sundal | 2013 | Gly585_lys619delinsAla | IVS12 | 38 | N/A | N/A |
| Sundal | 2013 | cys774_asn814del | IVS18 | 23 | N/A | N/A |
| Sundal | 2013 | cys774_asn814del | IVS18 | 50 | N/A | N/A |
| Terasawa ¹⁴ | 2013 | A823V | 19 | 52 | N/A | N/A |
| Battisti ¹⁵ | 2014 | C653R | 14 | 48 | N/A | N/A |
| Battisti | 2014 | I843F | 19 | 56 | N/A | N/A |
| Battisti | 2014 | I1906T | 21 | 37 | N/A | N/A |
| Hoffmann ¹⁶ | 2014 | R777Q | 18 | 22 | N/A | N/A |
| Hoffmann | 2014 | R777Q | 18 | 29 | N/A | N/A |
| Hoffmann | 2014 | R777Q | 18 | 30 | N/A | N/A |
| Hoffmann | 2014 | R777Q | 18 | 40 | N/A | N/A |
| Kitani Morii ¹⁷ | 2014 | I794T | 18 | N/A | N/A | N/A |
| Konno ¹⁸ | 2014 | S688Efx13 | 15 | 41 | N/A | N/A |
| Konno | 2014 | G765D | 17 | 37 | N/A | N/A |
| Konno | 2014 | A781E | 18 | 36 | N/A | N/A |
| Konno | 2014 | I794T | 18 | 40 | N/A | N/A |
| Konno | 2014 | I794T | 18 | 55 | N/A | N/A |
| Konno | 2014 | P842S | 19 | 45 | N/A | N/A |
| Konno | 2014 | C2442+1G>T | IVS18 | 53 | 57 | 4 |
| La Piana ¹⁹ | 2014 | V784M | 18 | 39 | N/A | N/A |
| Levin ²⁰ | 2014 | V838L | 19 | N/A | N/A | N/A |
| Schuberth ²¹ | 2014 | L582P | 12 | 54 | N/A | N/A |
| Schuberth | 2014 | E633K | 14 | 41 | N/A | N/A |
| Schuberth | 2014 | V838L | 19 | 45 | N/A | N/A |
| Sundal ²² | 2015 | Asn854Lys | 20 | 29 | N/A | N/A |
| Sundal | 2015 | Asn854Lys | 20 | 38 | N/A | N/A |
| Sundal | 2015 | Asn854Lys | 20 | 42 | N/A | N/A |
| Sundal | 2015 | Asn854Lys | 20 | 44 | N/A | N/A |
| Sundal | 2015 | Asn854Lys | 20 | 45 | N/A | N/A |
| Sundal | 2015 | Asn854Lys | 20 | 50 | N/A | N/A |
| Kim ²³ | 2015 | M766V | 17 | 39 | N/A | N/A |
| Kim | 2015 | R782H | 18 | 37 | 42 | 5 |
| Kim | 2015 | A781V | 18 | 41 | N/A | N/A |

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|-------|------|-------------|-------|----|-----|-----|
| Kim | 2015 | A781V | 18 | 44 | 49 | 5 |
| Lynch | 2015 | V596M | 13 | 31 | 36 | 5 |
| Lynch | 2015 | E633K | 14 | 29 | N/A | N/A |
| Lynch | 2015 | A763P | 17 | 45 | 51 | 6 |
| Lynch | 2015 | G825L | 19 | 42 | N/A | N/A |
| Lynch | 2015 | c2442+1 G>a | IVS18 | 44 | N/A | N/A |
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SUPPLEMENTARY REFERENCES

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