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| --- | --- | --- | --- |
| Table e-1. CADD scores in figure 6. |  |  |  |
| Age of onset | Report | Zygosity | Mutation(s) | CADD score |
| Infant | van Grunsven EG, 199816) | homozygote | G16S | 18.23 |
|  | Nakano K, 200123) | compound heterozygote | R106P / V404Efs\*3 | 33 / 32 |
|  | Ferdinandusse S, 200621) | homozygote | T15A | 18.37 |
|  | Ferdinandusse S, 200621) | homozygote | A17V | 18.79 |
|  | Ferdinandusse S, 200621) | homozygote | L21F | 23.6 |
|  | Ferdinandusse S, 200621) | homozygote | A26P | 23.4 |
|  | Ferdinandusse S, 200621) | homozygote | N72K | 24.2 |
|  | Ferdinandusse S, 200621) | compound heterozygote | D74\_I94del / A348T | 21.8 / 28.9 |
|  | Ferdinandusse S, 200621) | homozygote | R104M | 31 |
|  | Ferdinandusse S, 200621) | homozygote | G126V | 29.5 |
|  | Ferdinandusse S, 200621) | homozygote | S153L | 31 |
|  | Ferdinandusse S, 200621) | homozygote | A162D | 28.8 |
|  | Ferdinandusse S, 200621) | homozygote | S177F | 24.3 |
|  | Ferdinandusse S, 200621) | homozygote | V218L | 23.8 |
|  | Ferdinandusse S, 200621) | compound heterozygote | V218L / W273C | 23.8 / 33 |
|  | Ferdinandusse S, 200621) | homozygote | W224\* | 38 |
|  | Ferdinandusse S, 200621) | homozygote | E232K | 24 |
|  | Ferdinandusse S, 200621) | homozygote | A241T | 27.9 |
|  | Ferdinandusse S, 200621) | homozygote | G242E | 29.2 |
|  | Ferdinandusse S, 200621) | compound heterozygote | R248C / W249G | 34 / 25.4 |
|  | Ferdinandusse S, 200621) | homozygote | R248C | 34 |
|  | Ferdinandusse S, 200621) | homozygote | E366G | 31 |
|  | Ferdinandusse S, 200621) | homozygote | L405P | 28.5 |
|  | Ferdinandusse S, 200621) | compound heterozygote | N457D / I516T | 23.6 / 27.4 |
|  | Ferdinandusse S, 200621) | compound heterozygote | N457Y / I516T | 24.1 / 27.4 |
|  | Ferdinandusse S, 200621) | homozygote | R506C | 34 |
|  | Ferdinandusse S, 200621) | homozygote | R506H | 33 |
|  | Ferdinandusse S, 200621) | homozygote | D510Y | 31 |
|  | Ferdinandusse S, 200621) | homozygote | P529L | 31 |
|  | Ferdinandusse S, 200621) | homozygote | H532R | 25.8 |
|  | Ferdinandusse S, 200621) | homozygote | G533R | 29.5 |
|  | Ferdinandusse S, 200621) | homozygote | Q677\* | 40 |
|  | Tsuchida S, 201425) | compound heterozygote | G525V / G658\* | 33 / 36 |
|  | Konkol’ová J, 201522) | homozygote | N457D | 23.6 |
|  | Konkol’ová J, 201522) | compound heterozygote | N457D / R506C | 23.6 / 34 |
|  | Nascimento J, 201524) | homozygote | N457Y | 24.1 |
|  | Yubero D, 201627) | homozygote | R132W | 27.1 |
| Juvenile | Pierce SB, 201011) | compound heterozygote | Y217C / Y568\* | 27.4 / 38 |
|  | McMillan HJ, 20128) | compound heterozygote | A34V / I516T | 12.91 / 27.4 |
|  | Lines MA, 20146) | compound heterozygote | P513T / R543P | 26.8 / 22.8 |
|  | Amor DJ, 20164) | compound heterozygote | I62Mfs\*12 / H227R | 26.7 / 24.1 |
|  | Chen K, 20179) | homozygote | A100S | 27.3 |
|  | Matsukawa T, 20177) | compound heterozygote | R132W / A175T | 27.1 / 11.6 |
| Adult | Amor DJ, 20164) | compound heterozygote | c.58+1G>A / N98S | 22.1 / 25.8 |
|  | Amor DJ, 20164) | compound heterozygote | G378\* / L221F | 40 / 19.69 |
|  | This study | homozygote | A175T | 11.6 |