# INC 6601 – Minimal Data Set – Diagnosis Information

Protocol Number: Participant ID:
Site: Date of Visit:
Person Completing Form:

| | | | | |
|---|---|---|---|---|
| 1. Diagnosis: | ○ | Charcot Marie Tooth Disease (CMT) | ○ | Hereditary Neuropathy with Liability to Pressure Palsies (HNPP) |
| | ○ | Hereditary Motor Neuropathy (HMN) | ○ | Hereditary Sensory Neuropathy (HSN) |
| | ○ | Unknown | ○ | Other |
| | ○ | Control | If Other, specify: (Use only capital letters. Use only 1 space to separate two words. Do not use commas.) _____ _____ | |
| 2. NCS Pattern: | ○ | Demyelinating | ○ | Axonal |
| | ○ | Intermediate | ○ | Unknown |
| 3. Inheritance: | ○ | Dominant | ○ | Recessive |
| | ○ | X-linked | ○ | Unknown |
| 4. Does the patient have Diabetes? | ○ | Yes | ○ | No |
| 4a. If yes, indicate diabetes type: | ○ | Type 1 | ○ | Type 2 |
| | ○ | Gestational Diabetes | ○ | Pre-diabetes |
| 5. Genetic Testing: | ○ | Not tested (pt or family) | ○ | Tested (pt or family) ○ Unknown |

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|---|---|---|---|---|---|---|
| 6. CMT Subtype (Primary Pathogenic Mutation)*: | ○ | Autosomal Dominant CMT1 | ○ | CMT1A (*PMP22* duplication) | ○ | CMT1E (*PMP-22* point mutation) |
| | | | ○ | CMT1B (*MPZ*) | ○ | CMT1F (*NEFL*) |
| | | | ○ | CMT1C (*LITAF*) | ○ | CMT 1 Plus: (*FBLN5*) |
| | | | ○ | CMT1D (*EGR2*) | ○ | Other |
| | | | If Other, specify: (Use only capital letters and no spaces within the name of the subtype or mutation. Use only 1 space to separate two names. Do not use commas.) _____ | | | |
| | ○ | HNPP (*PMP-22* deletion) | | | | |
| | ○ | Autosomal Recessive Demyelinating | ○ | CMT4A (*GDAP1*) | ○ | CMT4E (*EGR2*) |
| | | | ○ | CMT4B1 (*MTMR2*) | ○ | CMT4F (*PRX*) |
| | | | ○ | CMT4B2 (*MTMR13*) | ○ | CMT4G (*HK1*) |
| | | | ○ | CMT4B3 (*SBF1*) | ○ | CMT4H (*FGD4*) |
| | | | ○ | CMT4C (*SH3TC2*) | ○ | CMT4J (*FIG4*) |
| | | | ○ | CMT4D (*NDRG1*) | ○ | Other |
| | | | If Other, specify: (Use only capital letters and no spaces within the name of the subtype or mutation. Use only 1 space to separate two names. Do not use commas.) _____ | | | |
| | ○ | Autosomal Dominant CMT2 | ○ | CMT2A (*MFN2*) | ○ | CMT2M (*DNM2*) |
| | | | ○ | CMT2B/ HSAN1B (*RAB7*) | ○ | CMT2N: (*AARS*) |
| | | | ○ | CMT2C (*TRPV4*) | ○ | CMT2O: (*DYNC1H1*) |
| | | | ○ | CMT2D (*GARS*) | ○ | CMT2P (*LRSAM1*) |
| | | | ○ | CMT2E (*NEFL*) | ○ | CMT2P-Okinawa/HSMN-P (*TFG*) |
| | | | ○ | CMT2F (*HSPB1*) | ○ | CMT2Q (*DHTKD1*) |
| | | | ○ | CMT2I: late onset CMT2 (*MPZ*) | ○ | CMT2 (*MARS*) |
| | | | ○ | CMT2J: CMT2 with hearing loss and pupillary abnormalities (*MPZ*) | ○ | CMT2 (*HARS*) |

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|---|---|---|---|---|---|---|
| | | | ○ | CMT2K: (*GDAP1*) | ○ | SPG10: hereditary spastic paraplegia (*KIF5A*) |
| | | | ○ | CMT2L: (*HSPB8*) | ○ | CMT2 with or without pyramidal signs) (*MT-ATP6*) |
| | | | ○ | Other | | |
| | | | If Other, specify: (Use only capital letters and no spaces within the name of the subtype or mutation. Use only 1 space to separate two names. Do not use commas.) _____ | | | |
| | ○ | Autosomal Recessive Axonal | ○ | AR CMT2B1 (*LMNA*) | ○ | AR CMT2 (*KARS*) |
| | | | ○ | AR CMT2B2 (*MED25*) | ○ | NMAN: Neuromyotonia (*HINT1*) |
| | | | ○ | AR CMT2 (*GDAP1*) | ○ | AR-CMT2: infantile onset(*TRIM2*) |
| | | | ○ | AR-CMT2 (*MFN2*) | ○ | Lethal congenital contractural syndrome type 2 (*ERBB3*) |
| | | | ○ | AR-CMT2 (*NEFL*) | ○ | Early onset AR-CMT2 (*SMN1*) |
| | | | ○ | AR-CMT2 (*LRSAMI*) | ○ | Other |
| | | | If Other, specify: (Use only capital letters and no spaces within the name of the subtype or mutation. Use only 1 space to separate two names. Do not use commas.) _____ | | | |
| | ○ | X-linked CMT | ○ | CMT1X (*GJB1*) | ○ | Other |
| | | | ○ | CMTX6 (*PDK3*) | | |
| | | | If Other, specify: (Use only capital letters and no spaces within the name of the subtype or mutation. Use only 1 space to separate two names. Do not use commas.) _____ | | | |
| | ○ | Dominant intermediate CMT | ○ | CMTDIB/CMT2M (*DNM2*) | ○ | CMTDIE (*INF2*) |
| | | | ○ | CMTDIC (*YARS*) | ○ | CMTD1F (*GNB4*) |
| | | | ○ | CMTDID (*MPZ*) | ○ | Other |
| | | | If Other, specify: (Use only capital letters and no spaces within the name of the subtype or mutation. Use only 1 space to separate two names. Do not use commas.) _____ | | | |
| | ○ | Recessive intermediate CMT | ○ | CMTRIA (*GDAP1*) | ○ | RI-CMT (*PLEKHG5*) |
| | | | ○ | CMTRIB (*KARS*) | ○ | Other |
| | | | If Other, specify: (Use only capital letters and no spaces within the name of the subtype or mutation. Use only 1 space to separate two names. Do not use commas.) _____ | | | |
| | ○ | Hereditary Motor Neuropathy | ○ | HMN2A (*HSPB8*) | ○ | HMN7A (*SLC5A7*) |
| | | | ○ | HMN2B (*HSPB1*) | ○ | HMN7B (*DCTN1*) |
| | | | ○ | HMN2C (*HSPB3*) | ○ | SMAX3 (*ATP7A*) |
| | | | ○ | HMN with pyramidal features/ ALS4 (*SETX*) | ○ | SMALED (*BICD2*) |
| | | | ○ | DSMA5 (*DNAJB2*) | ○ | SMALED/CMT2O (*DYNC1H1*) |
| | | | ○ | HMN5A (*BSCL2*) | ○ | SPSMA (*TRPV4*) |
| | | | ○ | HMN5A (*GARS*) | ○ | HMN (*AARS*) |
| | | | ○ | HMN5B, SPG31 (*REEP1*) | ○ | Other |
| | | | ○ | HMN6/ SMARD1 (*IGHMBP2*) | | |
| | | | If Other, specify: (Use only capital letters and no spaces within the name of the subtype or mutation. Use only 1 space to separate two names. Do not use commas.) _____ | | | |
| | ○ | Hereditary Sensory Neuropathy | ○ | HSAN1A (*SPTLC1*) | ○ | HSAN3 (*IKBKAP*) |
| | | | ○ | HSAN1C (*SPTLC2*) | ○ | HSAN4 (*NTRK1*) |
| | | | ○ | CMT2B (*RAB7*) | ○ | HSAN5 (*NGF-B*) |
| | | | ○ | HSN1D (*ATL1*) | ○ | Insensitivity to pain/Paroxysmal extreme pain disorder/Primary erythermalgia (*SCN9A*) |
| | | | ○ | HSN1E (*DNMT1*) | ○ | HSAN and dementia (*PRNP*) |

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|---|---|---|---|---|---|---|
| | | | ○ | HSAN2A (*WNK1*) | ○ | HSAN+myopathic facies (*DST*) |
| | | | ○ | HSAN2B (*FAM134B*) | ○ | Other |
| | | | ○ | HSN2C (*KIF1A*) | | |
| | | | If Other, specify: (Use only capital letters and no spaces within the name of the subtype or mutation. Use only 1 space to separate two names. Do not use commas.) _____ | | | |
| | ○ | Syndromic CMT | ○ | CCFDN: congenital cataracts, facial dysmorphism and neuropathy (*CTDP1*) | ○ | HSAN+spastic paraparesis (*CCT5*) |
| | | | ○ | CMTX4 (Cowchock syndrome) (*AIFM1*) | ○ | Waardenburg-Shah syndrome: Hirshsprung disease (*SOX10*) |
| | | | ○ | CMTX5 (*PRPS1*) – syndromic neuropathy | ○ | Other |
| | | | ○ | PNMHH (*MYH14*) – *myopathy and hearing loss* | | |
| | | | If Other, specify: (Use only capital letters and no spaces within the name of the subtype or mutation. Use only 1 space to separate two names. Do not use commas.) _____ | | | |
| | ○ | Patient has multiple primary pathogenic mutations/CMT Subtypes. | | | | |
| | ○ | Unknown | | | | |

| | | | | | | |
|---|---|---|---|---|---|---|
| 7. Mutation Type*: | ○ | Duplication | ○ | Deletion | ○ | Unknown |
| | ○ | Sequence | ○ | Other (ex. Multiple Primary Pathogenic Mutations) | | |
| | If Other, specify:<br>- For the unusual cases in which disease causing mutations in multiple genes may be present, please specify the genes and their mutation types. Type the gene first, hyphen, then type using the abbreviations: DUP, DEL, SEQ (no spaces).<br>- Use only capital letters and no spaces within the name of a gene or mutation type. Use only 1 space to separate words. Do not use commas. | | _____<br>_____<br>_____<br>_____ | | | |
| Nucleotide Change: | _____ | | | | | |
| Amino Acid Change: | _____ | | | | | |

| | | | | | | |
|---|---|---|---|---|---|---|
| 8. Genes Tested & Results: | PMP22 | ○ | Positive | ○ | Negative | ○ | Not tested |
| | GJB1 (CX32) | ○ | Positive | ○ | Negative | ○ | Not tested |
| | MFN2 | ○ | Positive | ○ | Negative | ○ | Not tested |
| | GDAP1 | ○ | Positive | ○ | Negative | ○ | Not tested |
| | SH3TC2 | ○ | Positive | ○ | Negative | ○ | Not tested |
| | LITAF | ○ | Positive | ○ | Negative | ○ | Not tested |
| | EGR2 | ○ | Positive | ○ | Negative | ○ | Not tested |
| | RAB7 | ○ | Positive | ○ | Negative | ○ | Not tested |
| | GARS | ○ | Positive | ○ | Negative | ○ | Not tested |
| | NEFL | ○ | Positive | ○ | Negative | ○ | Not tested |
| | HSPB1 | ○ | Positive | ○ | Negative | ○ | Not tested |
| | HSPB8 | ○ | Positive | ○ | Negative | ○ | Not tested |
| | PRX | ○ | Positive | ○ | Negative | ○ | Not tested |
| | MPZ | ○ | Positive | ○ | Negative | ○ | Not tested |
| | BSCL2 | ○ | Positive | ○ | Negative | ○ | Not tested |
| | DMN2 | ○ | Positive | ○ | Negative | ○ | Not tested |

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|---|---|---|---|---|---|---|---|
| | FGD4 | ○ | Positive | ○ | Negative | ○ | Not tested |
| | FIG4 | ○ | Positive | ○ | Negative | ○ | Not tested |
| | LMNA | ○ | Positive | ○ | Negative | ○ | Not tested |
| | MTMR2 | ○ | Positive | ○ | Negative | ○ | Not tested |
| | NDRG1 | ○ | Positive | ○ | Negative | ○ | Not tested |
| | PRPS1 | ○ | Positive | ○ | Negative | ○ | Not tested |
| | SBF2 (MTMR13) | ○ | Positive | ○ | Negative | ○ | Not tested |
| | SOX10 | ○ | Positive | ○ | Negative | ○ | Not tested |
| | SPTLC1 | ○ | Positive | ○ | Negative | ○ | Not tested |
| | TRPV4 | ○ | Positive | ○ | Negative | ○ | Not tested |
| | YARS | ○ | Positive | ○ | Negative | ○ | Not tested |
| | If Other, Specify (Use only capital letters and no spaces within the gene name.): | | | | | | |
| | _____ | ○ | Positive | ○ | Negative | ○ | Not tested |
| | _____ | ○ | Positive | ○ | Negative | ○ | Not tested |
| | _____ | ○ | Positive | ○ | Negative | ○ | Not tested |
| | _____ | ○ | Positive | ○ | Negative | ○ | Not tested |
| | _____ | ○ | Positive | ○ | Negative | ○ | Not tested |

Notes: _____
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