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New Evidence for Secondary Axonal Degeneration in Demyelinating Neuropathies

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Abstract

Development of peripheral nervous system (PNS) myelin involves a coordinated series of events between growing axons and the Schwann cell (SC) progenitors that will eventually ensheath them. Myelin sheaths have evolved out of necessity to maintain rapid impulse propagation while accounting for body space constraints. However, myelinating SCs perform additional critical functions that are required to preserve axonal integrity including mitigating energy consumption by establishing the nodal architecture, regulating axon caliber by organizing axonal cytoskeleton networks, providing trophic and potentially metabolic support, possibly supplying genetic translation materials and protecting axons from toxic insults. The intermediate steps between the loss of these functions and the initiation of axon degeneration are unknown but the importance of these processes provides insightful clues. Prevalent demyelinating diseases of the PNS include inherited neuropathies Charcot-Marie-Tooth Disease, Type 1 (CMT1) and Hereditary Neuropathy with Liability to Pressure Palsies (HNPP) and the inflammatory diseases Acute Inflammatory Demyelinating Polyneuropathy (AIDP) and Chronic Inflammatory Demyelinating Polyneuropathy (CIDP). Secondary axon degeneration is a common feature of demyelinating neuropathies and this process is often correlated with clinical deficits and long-lasting disability in patients. There is abundant electrophysiological and histological evidence for secondary axon degeneration in patients and rodent models of PNS demyelinating diseases. Fully understanding the involvement of secondary axon degeneration in these diseases is essential for expanding our knowledge of disease pathogenesis and prognosis, which will be essential for developing novel therapeutic strategies.

Keywords

Acute Inflammatory Demyelinating Polyneuropathy; AIDP; Charcot-Marie-Tooth Disease; Chronic Inflammatory Demyelinating Polyneuropathy; CIDP; CMT; CMT1A; CMT1B; CMT1C; CMT1D; CMT1E; CMT1F; CMT1X; Cx32; Demyelination; EGR2; GBS; GJB1; Guillain-Barré Syndrome; Hereditary Neuropathy with Liability to Pressure Palsies; HNPP; LITAF/SIMPLE; MPZ; Myelin; NEFL; Peripheral Neuropathy; PMP22; Secondary Axon Degeneration

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1. Introduction

1.1. Myelinating Schwann Cells

Myelin sheaths arose from evolutionary pressure to achieve rapid impulse propagation without dramatically increasing axon diameters due to vertebrate organism space constraints. Peripheral nervous system (PNS) axons are ensheathed by myelinating Schwann cells (SC) in a symbiotic developmental program. Schwann cell precursors (SCPs) provide trophic support for growing axons and axons in turn support SCP survival and migration [158]. While axons are still growing towards their innervation targets, SCPs begin transitioning into immature Schwann cells (iSCs), which cease migrating and deposit basal lamina [398]. iSCs initiate myelination in a multi-step process called radial sorting. Three to eight iSCs surround bundles of axons to form units sharing a common basal lamina and then iSC lamellipodia-like processes invade the axons to categorize them by caliber [158]. Large caliber axons are surrounded by promyelinating Schwann cells in a 1:1 relationship and small caliber axons remain in Remak bundles that are engulfed by SC cytoplasm [158]. Myelinating SC then continue to polarize radially and longitudinally to ensheath axons; a process that requires precise expression and localization of myelin proteins and lipids. Myelin sheath thickness is regulated by several factors including Neuregulin-1 Type III, which activates SC Erb-b2 Receptor Tyrosine Kinase 2/3 and downstream signaling pathways, and internode length regulation is also complex and includes mechanical activation of Crumbs Cell Polarity Complex Component 3 and downstream Hippo signaling [158, 569]. Basal lamina signaling, involving Periaxin (PRX), is also involved in regulating internode length [569].

Less is known about the mechanisms involved in maintaining peripheral nerve myelin sheath integrity through adulthood [158]. However, advances in our knowledge of the pathomechanisms involved in demyelinating neuropathies are shedding light on this topic. Classic demyelination, the loss of myelin sheaths after their proper development, is typically observed in the acquired demyelinating neuropathies AIDP and CIDP. Whereas dysmyelination, when myelin sheaths likely never develop properly and may undergo a process resembling classic demyelination later in disease progression, is usually observed in the inherited demyelinating neuropathies CMT1A-F and -X. For the purpose of simplicity, classic demyelination and dysmyelination will both be referred to as demyelination for the remainder of this review.

1.2. Effects of Myelination and Demyelination on Axons

PNS myelin sheaths exert a number of effects on their axons including establishing the molecular architecture of the nodal region, organizing the cytoskeleton, supplying trophic support and providing protection from insults. The nodal region consists of three morphologically and molecularly distinct domains: (1) the node of Ranvier, which is the ~1μm gap between two myelinating SCs that contains high concentrations of voltage-gated sodium channels, (2) the paranode, which is immediately adjacent to the node and consists of cellular junctions between the axon and the edge of each myelin sheath layer, and (3) the juxtaparanode, which flanks the paranode and contains high concentrations of voltage-gated

potassium channels [440]. Establishing these domains requires cooperative interactions between axons and myelinating SCs and demyelination has been demonstrated to disrupt this architecture. Demyelinated axons consequently demonstrate slowed or blocked conduction, increased refractory periods and consume much greater quantities of Adenosine Triphosphate (ATP) to restore ion gradients [162, 481, 546]. These changes are driven by increased axonal expression and accumulation of voltage-gated sodium channels which leads to excessive sodium influx and axon degeneration by a calpain-mediated mechanism [162]. The axonal cytoskeleton also contributes to establishing the nodal architecture. Axonal actin is organized in ring-like structures that wrap around the perimeter of the axon shaft and are evenly spaced along the length of axon by adjoining spectrin tetramers [628]. This subcortical actin/spectrin scaffold is involved in establishing both the node of Ranvier, through interactions with AnkyrinG which cluster voltage-gated sodium channels, and the paranode, by helping to establish the paranodal lateral diffusion barrier [647]. Additionally, axonal neurofilament organization and function is affected by myelination. Neurofilaments are highly phosphorylated at myelinated internodes which increases neurofilament spacing through electrostatic repulsion and reduces neurofilament transport leading to filament accumulation and increased axon caliber [440]. Conversely, neurofilaments at nodes of Ranvier are less phosphorylated, more concentrated and less numerous and demyelinated axons have been suggested to have a similar organization [440]. Interestingly, myelination has not been demonstrated to regulate microtubule organization but does affect the distribution of cargo transported on microtubules. Mitochondria are transported on microtubules and accumulate at nodes of Ranvier and demyelination alters mitochondrial dynamics [98, 274]. Impaired transport of axonal cargoes due to neurofilament accumulation has been suggested to cause axon degeneration [162].

SCs and their progenitors have clearly been demonstrated to provide trophic support to developing axons, but this function is less understood for the long-term maintenance of axon viability. SCs secrete a number of neurotrophins, including neurotrophin-3 (NT-3) , Insulin-like Growth Factor 1, Ciliary Neurotrophic Factor (CNTF) and Erythropoietin, and SC-derived neurotrophic factor depletion is correlated with axon degeneration [42]. However, it remains unclear whether SC-mediated tropic support of axons is independent of myelination as studies from central nervous system (CNS) myelin suggest [42]. Regardless, myelinating SCs likely rely on the contact points with axons, the inner mesaxon, Schmidt-Lanterman incisures and paranodal loops, for supplying trophic support. These non-compact myelin structures have also been implicated in the transfer of mRNA, proteins and occasionally ribosomes by vesicular transport [345, 538]. Metabolic substrates may also be transported from myelinating SCs to axons to meet energy demands. Indeed, CNS myelin provides monocarboxylates to axons through transporters in the adaxonal oligodendrocyte membrane but a similar mechanism is less defined in the PNS [168]. However, there is evidence that myelinating SCs can supply axons with lactate [29, 70, 476]. Although these studies clearly demonstrate that SCs provide axons with nutritive substances, these functions appear to be independent from myelination [70]. Nonetheless, demyelination leads to a separation of the symbiotic relationship between axons and SCs so it is likely that the demyelination-induced absence of these functions leads to axon degeneration. Additionally, PNS myelin sheaths have been suggested to protect axons from toxic insults [42]. Chemotherapy treatment has

been demonstrated to exacerbate symptoms of demyelinating diseases (discussed below) but evidence demonstrating that demyelinated axons are more sensitive to insults than myelinated axons is inadequate.

1.3. Axon Degeneration Pathways

Neurons possess a cell-autonomous axon degeneration program that occurs when axons are transected (Wallerian Degeneration, WD) and in some types of neurological disease (Wallerian-like Degeneration, WLD) [105]. This pathway has been extensively reviewed but briefly, depletion of the Nicotinamide Adenine Dinucleotide (NAD^+) synthesizing enzyme Nicotinamide Mononucleotide Adenylyl Transferase 2 activates Sterile Alpha and TIR Motif Containing 1 (SARM1), which leads to a further reduction in NAD^+ levels due to the NADase activity of SARM1 [105]. Depletion of NAD^+ leads to the loss of ATP, an energetic failure and calpain-mediated proteolysis of axon structural proteins [105]. Axon degeneration can also occur by apoptotic and pruning mechanisms [179]. The apoptotic program leads to degeneration of both the axon and soma and has been best characterized in models of global neurotrophic factor deprivation [180]. Briefly, activation of Duel Leucine Zipper Kinase/c-Jun N-terminal Kinase signaling leads to upregulated expression of proapoptotic genes, BCL2 associated X, apoptosis regulator activation and translocation to mitochondria, release of cytochrome C and activation of cell-destructive caspases and calpains [179, 180]. The pruning program leads to degeneration of axons or axon segments and has been best established in models of local neurotrophic factor deprivation [180]. This pathway is identical to the apoptotic program except for a few components but how the destructive caspases and calpains are spatially restricted is unknown [180].

WLD has been implicated in demyelinating peripheral nerve diseases but inhibiting this pathway generally only modestly and temporarily rescues axon degeneration indicating that additional mechanisms are involved [106, 401]. Additionally, the initiation events for demyelination-induced axon degeneration are undefined but clues are beginning to emerge. The role of PNS myelin sheaths to mitigate energy consumption, regulate axon caliber, influence mitochondria distribution, provide trophic and metabolic support, supply genetic translation materials and insulate axons from insults suggests that the absence of these functions may trigger axon degeneration. The evidence for secondary axon degeneration in demyelinating diseases and models and progress made towards understanding the mechanisms involved will be examined below.

2. Secondary Axon Degeneration in Demyelinating Neuropathies

2.1. Charcot-Marie-Tooth Disease (CMT) and Hereditary Neuropathy with Liability to Pressure Palsies (HNPP)

CMT is a diverse group of inherited peripheral nerve disorders that are most commonly categorized by the primary cells involved in pathogenesis. CMT Type 1 (CMT1) is caused by myelinating SC dysfunction and CMT Type 2 (CMT2) is caused by axonal deficits [249]. However, both CMT1 and CMT2 patients present with foot deformities, absent reflexes and progressive distal weakness and sensory loss, which often begin during adolescence [249]. The similarities in symptoms indicate that axon degeneration, secondary in CMT1 and

primary in CMT2, is likely the driver of functional deficits in all CMT patients. Additionally, HNPP is frequently categorized as a demyelinating CMT distinct from CMT1 subtypes [73]. HNPP patients present with symptoms similar to CMT1 and CMT2 patients suggesting that secondary axon degeneration is also involved in the pathogenesis of this disease [25].

2.1.1. CMT1A—CMT1A is the most common form of CMT and is caused by duplication of a 1.5 Mb region (17p 11.2-p12) containing the *Peripheral Myelin Protein 22* (*PMP22)* gene [73, 614]. Patient histological and electrophysiological evaluation reveals myelin deficits including loss of myelinated fibers, thin myelin sheaths, de- and remyelination, prevalent onion bulb formation, occasional tomacula (focal myelin thickenings) and slowed nerve conduction velocity (NCV) [94, 129, 214, 293, 593] (Supplemental Table 1). The nature of the genetic defect in CMT1A patients suggests excess PMP22 expression but the pathogenic mechanism appears to be more complex given that PMP22 protein levels are often variable, fluctuating between comparable levels to healthy controls to elevated expression [324]. PMP22 is thought to function as a structural component of compact myelin and increased gene copy number in CMT1A patients has been suggested to be detrimental to myelinating SCs due to disrupted stoichiometry of compact myelin constituents, disturbed secretory pathway function and dramatic alterations to the transcriptome [408, 614]. There is electrophysiological evidence for secondary axon degeneration in CMT1A patients which is suggested to drive clinical deficits (Table 1). Compound muscle action potential (CMAP) and sensory nerve action potential (SNAP) amplitudes correlate with clinical disability whereas motor conduction velocity (MCV) and sensory conduction velocity (SCV) generally do not [39, 214, 224, 270, 288, 292, 293, 368, 526, 593, 596, 645]. Additionally, histological evidence for secondary axon degeneration includes axon atrophy and loss, active axonal degeneration and axon sprouts/clusters of regenerating axons observed in sural, peroneal and radial nerve biopsies from CMT1A patients (Table 1). Several rodent PMP22 overexpression models have been generated to model CMT1A and there is electrophysiological and histological evidence for secondary axon degeneration in these models (Table 1). There are several clues about the mechanisms causing secondary axon degeneration in CMT1A. Voltage-gated potassium channel organization is disturbed in peripheral nerve axons from C61 *PMP22* transgenic mice suggesting that nodal architecture is disrupted in CMT1A [284]. Additionally, neurofilaments were hypophosphorylated and exhibited increased density in peripheral nerve axons from CMT1A rodent models [157, 192, 423, 500]. Mouse sciatic nerve axons regenerated through sural nerve grafts from CMT1A patients also demonstrated increased neurofilament densities as well as accumulated mitochondria and reduced microtubule densities [489, 492]. Accumulated membranous organelles were observed in peripheral nerve axons of *PMP22* transgenic mouse models as well [592]. These findings indicate that axonal cytoskeletal organization and cargo transport are altered in CMT1A. Dysfunctional trophic support has also been implicated in CMT1A. CNTF expression is reduced in CMT1A patient sural nerve biopsies and model SCs and treating CMT1A model dorsal root ganglia cultures with recombinant CNTF reduces neurofilament hypophosphorylation [422, 597]. Additionally, NT-3 improved growth and myelination of axons regenerated through sural nerve grafts from CMT1A patients and a pilot clinical trial with recombinant

methionyl human NT-3 yielded promising improvements in clinical disability scores and sensory function [494]. There are also reports of chemotherapy treatment exacerbating CMT1A symptoms suggesting that patient axons are more vulnerable to toxic insults like chemotherapy agents, likely due to their dysfunctional myelin sheaths [9, 102, 191, 241].

2.1.2. HNPP—HNPP is the third most common form of CMT and is caused by deletion of the same 1.5 Mb region that is duplicated in CMT1A (17p11.2-p12), which contains the *PMP22* gene [25, 73]. Patient histological and electrophysiological evaluation reveals myelin deficits including loss of myelinated fibers, thin myelin sheaths, de- and remyelination, prominent tomacula formation, occasional onion blubs and generally focally slowed NCV [119, 189, 351, 439] (Supplemental Table 1). As discussed previously, PMP22 likely functions as a structural component of compact myelin and haploinsufficiency in HNPP patients has been suggested to be pathogenic due to loss of PMP22 function [73]. There is electrophysiological and histological evidence for secondary axon degeneration in HNPP patients, which is suggested to correlate with clinical deficits [285, 467] (Table 1). Sural, peroneal and radial nerve biopsies from HNPP patients revealed axon atrophy and loss, clusters of regenerating axons and occasional myelin ovoids (Table 1). PMP22 heterozygous knockout mice and PMP22 heterozygous super enhancer deletion mice are used to model HNPP and there is electrophysiological and histological evidence for secondary axon degeneration in these mice (Table 1). Interestingly, these mice show more severe deficits when homozygous and also exhibit electrophysiological and histological evidence for secondary axon degeneration [8, 442, 500, 651]. Insights into mechanisms causing secondary axon degeneration in HNPP are beginning to emerge. Increased neurofilament and microtubule density has been observed in superficial peroneal nerve biopsy axons from an HNPP patient and compressed axons are frequently detected in peripheral nerve biopsies from HNPP patients and PMP22 heterozygous knockout mice [7, 124, 216, 474]. These findings suggest that axonal cytoskeletal organization and potentially cargo transport are disturbed in HNPP. Additionally, chemotherapy treatment has been demonstrated to exacerbate symptoms in an HNPP patient, indicating that their axons are more vulnerable to toxic insults [253].

2.1.3. CMT1B—CMT1B is the second most common form of CMT1 and is caused by mutations in the *Myelin Protein Zero* (*MPZ*) gene [73]. Numerous *MPZ* point mutations have been identified and they are primarily localized to the single extracellular domain of the MPZ protein [73, 527]. Interestingly, the majority of CMT1B patients can be divided into three distinct groups: infantile-, adolescent- and adult-onset. Infantile- and adolescent-onset patient histological and electrophysiological evaluation reveals myelin deficits including loss of myelinated fibers, myelin sheaths ranging from thin to nearly absent, de- and remyelination, variable onion bulb formation, occasional tomacula, myelin uncompaction and slowed NCV [502] (Supplemental Table 1). However, deficits are generally more prominent in patients with infantile-onset CMT1B. Interestingly, adult-onset patients often have undetectable NCV and myelin changes. However, occasional thin myelin sheaths, de- and remyelination, onion bulb formation, tomacula, myelin uncompaction and moderately slowed NCV have been observed [502] (Supplemental Table 1). Loss of myelinated fibers were not included as evidence of myelin deficits in these patients due to the prominent

observation of axon loss which is likely reflected in this assessment. MPZ is a myelin-specific glycoprotein that is required to form the compact multilamellar structure of peripheral nerve myelin sheaths [73]. In general, mutations that cause infantile-onset disease are suggested to dramatically disrupt the structure of MPZ, which activates the unfolded protein response due to accumulation of misfolded MPZ in the endoplasmic reticulum (ER) and consequently disrupts myelin sheath compaction due to deficiency of MPZ in the myelin lamellae [408]. Adolescent-onset disease mutations are predicted to confer more subtle changes to the structure of MPZ presumably leading to milder disruptions in myelin sheath compaction [408]. Mutations that cause adult-onset disease have been suggested to result in subtle changes to myelin sheaths, likely involving disrupted contact between myelinating SCs and their axons [408]. However, axon degeneration is likely the underlying cause of functional deficits in all CMT1B patients given that CMAP amplitudes correlate with clinical disability but MCV does not [151, 214]. Axon degeneration is secondary to the prominent demyelination in infantile- and adolescent-onset patients but it is also likely secondary in adult-onset patients even though demyelination is often undetectable and these patients are frequently classified as CMT2 [408]. Electrophysiological data supporting secondary axon degeneration is available for several MPZ mutations belonging to each disease subtype (Table 1). Histological data demonstrating secondary axon degeneration is also available, especially for adult-onset disease mutations. Sural and peroneal nerve biopsies from infantile-, adolescent- and adult-onset CMT1B patients revealed axon atrophy and loss, active axonal degeneration, axon sprouts/clusters of regenerating axons, bands of Büngner and myelin ovoids (Table 1). Interestingly, a sural nerve biopsy from an infantile-onset patient (p.R98C) showed a minor reduction in fiber density as compared to a sural nerve biopsy harvested 20 years prior from the same patient [32]. These findings were consistent with the clinical deficits and indicate that disease progression was minimal following the initial rapid deterioration during infancy [32]. Although there are a limited number of CMT1B rodent models, there is evidence for secondary axon degeneration in infantile- (p.R98C knockin mice) and adolescent-onset (p.S63del transgenic mice) disease models [505, 619] (Table 1). Intriguingly, MPZ knockout and overexpression mice also develop demyelinating neuropathies and exhibit electrophysiological and histological evidence for secondary axon degeneration [163, 184, 374, 396, 620, 651]. There are several clues pertaining to the mechanisms that cause secondary axon degeneration in CMT1B. Disrupted nodal architecture is observed in biopsied sural nerves from an infantile-onset (p.R98C) patient and autopsied peripheral nerves from an adult-onset (p.H39P) patient; Contactin Associated Protein-like 1 (CASPR) and Contactin-1 (CNTN1) organization is disrupted and voltage-gated sodium channel subtype is altered [32, 325]. Remarkably, MPZ has recently been shown to play a direct role in maintaining nodal architecture by interacting with Neurofascins (NF) [79]. Additionally, increased neurofilament density was detected in sural nerve biopsy axons from an infantile-onset (p.R98C) patient and an adult-onset (p.T124M) patient [207, 387]. Peripheral nerve axons in MPZ knockout mice also have increased neurofilament density and contain accumulated mitochondria [163, 184]. These findings suggest that axonal cytoskeletal organization and cargo transport are altered in CMT1B, a notion that is further supported by the presence of compressed axons in patient nerves [118, 325].

2.1.4. CMT1C—CMT1C is a rare form of CMT that is caused by mutations in the *Lipopolysaccharide Induced TNF Factor* (*LITAF*; also known as Small Integral Membrane Protein of Lysosome/Late Endosome [SIMPLE]) gene [73, 97]. Multiple *LITAF* point mutations have been identified and they are primarily localized to the C-terminal cysteine-rich domain, which is involved in endosome membrane binding [73, 97]. Patient histological and electrophysiological evaluation reveals myelin deficits including loss of myelinated fibers, thin myelin sheaths, variable onion bulb formation and slowed NCV [195, 244] (Supplemental Table 1). LITAF is a regulator of endosomal trafficking and related signaling processes and CMT1C mutations have been suggested to be pathogenic due to mislocalization of LITAF [97, 305, 331]. There is electrophysiological and histological evidence for secondary axon degeneration in CMT1C patients (Table 1). Sural nerve biopsies from p.T49M and p.P135R patients revealed axon loss and clusters of regenerating axons [103, 495]. There is also electrophysiological and histological evidence for secondary axon degeneration in a p.W116G transgenic mouse model (Table 1). Clues about the mechanisms causing secondary axon degeneration in CMT1C are beginning to emerge. CASPR and voltage-gated potassium channel organization is disrupted in p.W116G transgenic mouse sciatic nerve axons [316]. Additionally, mitochondria and other electron-dense organelles accumulate at the paranode, the initial site of the myelin sheath dysfunction, in these mice [316]. These findings suggest that nodal architecture and axonal cargo transport are disturbed in CMT1C.

2.1.5. CMT1D—CMT1D is a rare form of CMT that is caused by mutations in the *Early Growth Response 2 (EGR2*) gene [73]. Several *EGR2* point mutations have been identified and they are predominantly localized to one of the three zinc finger domains of the protein which confer DNA binding [73, 194]. Patient histological and electrophysiological evaluation reveals myelin deficits including loss of myelinated fibers, myelin sheaths ranging from thin to nearly absent, de- and remyelination, variable onion bulb formation, irregularly folded myelin sheaths and slowed NCV [73] (Supplemental Table 1). EGR2 is a SC transcription factor that regulates expression of essential myelin genes (*PMP22, gap junction protein beta 1* (*GJB1*), *PRX* and indirectly *MPZ*) and CMT1D mutations have been suggested to be pathogenic due to disrupted DNA binding and transcriptional activation [73]. There is electrophysiological and histological evidence for secondary axon degeneration in CMT1D patients (Table 1). Sural nerve biopsies from p.D355V, p.R359W and p.E412G patients revealed axon loss, active axonal degeneration and clusters of regenerating axons (Table 1). Insights into mechanisms causing secondary axon degeneration in CMT1D are emerging. CASPR and voltage-gated potassium channel organization is disrupted in sciatic nerve axons of p.I268N knockin mice [34]. Additionally, increased neurofilament density was detected in sural nerve biopsy axons from a p.S382R + p.D383Y patient and increased neurofilament and microtubule density and occasional axon compression were observed in sural nerve biopsy axons from a p.R359W patient [564, 613]. These findings suggest that nodal architecture, axonal cytoskeletal organization and potentially axonal cargo transport are disturbed in CMT1D. Additionally, chemotherapy treatment has been demonstrated to exacerbate symptoms in a CMT1D patient, indicating that their axons are more vulnerable to toxic insults [415].

2.1.6. CMT1E—CMT1E is a rare form of CMT that is caused by mutations in the *PMP22* gene [73, 328]. Numerous *PMP22* point mutations have been identified and they are localized throughout the protein but predominantly reside in or near the transmembrane domains [328]. Patient histological and electrophysiological evaluation reveals myelin deficits including loss of myelinated fibers, myelin sheaths ranging from thin to nearly absent, de- and remyelination, generally prominent onion bulbs, variable tomacula formation, occasional myelin uncompaction and slowed NCV [328] (Supplemental Table 1). As discussed previously, PMP22 likely functions as a structural component of compact myelin and CMT1E mutations have been suggested to be pathogenic due to two distinct mechanisms; (1) a toxic gain of function which likely involves ER stress due to misfolded PMP22 or (2) a loss of function similar to HNPP [328]. There is electrophysiological and histological evidence for secondary axon degeneration in CMT1E patients (Table 1). Sural nerve biopsies from CMT1E patients revealed axon atrophy and loss, active axonal degeneration, axon swellings, clusters of regenerating axons and myelin ovoids (Table 1). Multiple CMT1E mouse models also demonstrate electrophysiological and histological evidence for secondary axon degeneration (Table 1). Clues about mechanisms causing secondary axon degeneration in CMT1E are as follows. Nodal architecture is disturbed, particularly voltage-gated potassium channel organization, in Trembler (p.G150D) and TremblerJ (p.L16P) mice [128, 477, 610]. Trembler and TremblerJ mice have been instrumental in understanding the effects of demyelination on axons revealing cytoskeletal defects, including reduced neurofilament phosphorylation, increased neurofilament density, and altered cargo transport (Table 1). Additionally, increased neurofilament density was detected in sural nerve biopsy axons from a p.V30M patient and mouse sciatic nerve axons regenerated through sural nerve grafts from this patient also exhibited increased neurofilament densities [491]. Taken together, these findings suggest that nodal architecture, axonal cytoskeletal organization and axonal cargo transport are disturbed in CMT1E. Additionally, CNTF expression is reduced and injury-induced BDNF expression is diminished in TremblerJ sciatic nerves and NT-3 treatment improved myelination of regenerating TremblerJ axons suggesting that trophic support provided by CMT1E SCs is dysfunctional [165, 494].

2.1.7. CMT1F—CMT1F is a rare form of CMT that is caused by mutations in the *Neurofilament Light* (*NEFL*) gene [73]. Several *NEFL* point mutations have been identified and they are localized throughout the protein including in the head, rod and tail domains [225]. Patient histological and electrophysiological evaluation reveals myelin deficits including loss of myelinated fibers, thin myelin sheaths, de- and remyelination, variable onion bulb formation, occasional tomacula and slowed NCV [225] (Supplemental Table 1). Interestingly, NEFL is a neuronal cytoskeletal protein that is involved in regulating axon caliber (discussed previously) and CMT1F mutations are suggested to be pathogenic due to disrupted formation of intermediate filament networks [225, 453]. Given that NEFL is an axonal protein, the role of demyelination in CMT1F pathogenesis remains debatable. In fact, the slowed NCVs have been attributed to reduced axon caliber and not demyelination [454, 459, 642]. However, there is electrophysiological and histological evidence for axon degeneration in CMT1F patients (Table 1). Sural nerve biopsies from CMT1F patients revealed axon atrophy and loss, active axonal degeneration, axon swellings, clusters of

regenerating axons, bands of Büngner and myelin ovoids (Table 1). Although there are a limited number of CMT1F rodent models, there is evidence for axon degeneration in p.N98S knockin mice and p.E396K transgenic mice [308, 599] (Table 1). There are several clues about the mechanisms causing axon degeneration in CMT1F. As expected, sural nerve biopsy axons from CMT1F patients have disrupted neurofilament networks, which often causes dramatic accumulation of neurofilaments and axonal swellings [45, 134, 142, 143, 167, 359, 642, 653]. Additionally, proximal neurofilament accumulation is correlated with disturbed organelle trafficking and dysregulated density of neurofilaments (decreased) and microtubules (increased) in distal axons [141, 143, 167, 213, 359, 642, 653]. Similar findings were observed in CMT1F rodent models (Table 1). These results suggest that axonal cytoskeletal organization and cargo transport are disturbed in CMT1F. Further studies are needed to determine whether demyelination contributes to CMT1F pathogenesis or if it is merely a consequence of axonal damage.

2.1.8. CMT1X—CMT1X is the most common form of X-linked CMT and is caused by mutations in the *GJB1* gene [69, 276]. Numerous *GJB1* point mutations have been identified and they are localized throughout the protein including in the intracellular, transmembrane and extracellular domains [69, 276]. Patient histological and electrophysiological evaluation reveals myelin deficits including loss of myelinated fibers, thin myelin sheaths, occasional de- and remyelination, generally modest onion bulb formation and slowed NCV [276] (Supplemental Table 1). *GJB1* encodes Connexin 32 (Cx32), a gap junction channel, and CMT1X mutations have been suggested to be pathogenic due to loss of expression or function of Cx32 [69, 529]. Although a primary role for demyelination in CMT1X pathogenesis has been questioned, it is likely that myelin sheath function is disturbed given the important role for this protein in myelinating SCs [104]. However, there is electrophysiological and histological evidence for secondary axon degeneration in CMT1X patients, which often correlates with clinical deficits [214, 222, 332, 441, 529] (Table 1). Sural, peroneal and radial nerve biopsies from CMT1X patients revealed axon atrophy and loss, active axonal degeneration, axon sprouts/clusters of regenerating axons and myelin ovoids (Table 1). Cx32 knockout mice are the best characterized model of CMT1X and they also exhibit electrophysiological and histological evidence for secondary axon degeneration [21, 507] (Table 1). Clues about the mechanisms causing secondary axon degeneration in CMT1X are accumulating. Increased neurofilament and decreased microtubule densities as well as organelle accumulation have been observed in sural nerve biopsied axons from p.E102G and p.E208K CMT1X patients [203, 204, 490]. Similar results were observed in mouse sciatic nerve axons regenerated through sural nerve grafts from p.E102G CMT1X patients [490]. Additionally, peripheral nerve axons from Cx32 knockout mice demonstrate reduced neurofilament phosphorylation, increased neurofilament density and altered cargo transport [507, 589]. Interestingly, these changes were evident before demyelination was detectable supporting the notion that loss of Cx32 confers subtle changes to myelin sheath function [589]. These results suggest that axonal cytoskeletal organization and cargo transport are disturbed in CMT1X. Additionally, chemotherapy treatment has been demonstrated to exacerbate symptoms in a CMT1X patient, indicating that their axons are more vulnerable to toxic insults [464].

2.2. Demyelinating Inflammatory Neuropathies

Guillain-Barré syndrome (GBS) and Chronic Inflammatory Demyelinating Polyneuropathy (CIDP) are two of the most common inflammatory diseases targeting the PNS [576]. GBS has multiple subtypes of which Acute Inflammatory Demyelinating Polyneuropathy (AIDP) is the most prevalent in North America and Europe [576]. AIDP and CIDP present with similar symptoms of progressive appendicular weakness, variable sensory loss and diminished or absent reflexes [576]. Although these diseases primarily target peripheral nerve myelin sheaths, secondary axon degeneration is a common feature of both and has been suggested to correlate with long-term disability in patients (discussed below).

2.2.1. AIDP—AIDP is a demyelinating autoimmune disorder that targets components of peripheral nerves and nerve roots and is typically triggered by an acute infectious event [520]. Disease typically begins 10-14 days after an upper or lower respiratory illness or gastroenteritis and patients present with progressive weakness in their extremities that develops over a duration of less than four weeks with a variable age of onset [520]. The antigenic targets in AIDP patients remain unclear but galactocerebroside and ganglioside autoantibodies have been implicated in a small fraction of patients [187, 303]. Patient histological and electrophysiological evaluation of peripheral nerves reveals myelin deficits including loss of myelinated fibers, thin myelin sheaths, de- and remyelination and slowed MCV and prolonged distal and F-wave latencies [520] (Supplemental Table 1). However, distinguishing AIDP from axonal GBS by electrophysiological methods is difficult particularly early in disease progression [46]. There are genuine examples of AIDP with electrophysiological and histological evidence for secondary axon degeneration though (Table 1). Biopsied sural nerves and autopsied peripheral nerves from AIDP patients revealed axon loss, active axonal degeneration, clusters of regenerating axons, bands of Büngner and myelin ovoids (Table 1). Correlating secondary axon degeneration to patient prognosis in AIDP is challenging but some studies suggest that axonal involvement in GBS is associated with greater clinical disability [588]. Experimental Autoimmune Neuritis (EAN) rodent models recapitulate key features of AIDP and multiple models demonstrate electrophysiological and histological evidence for secondary axon degeneration (Table 1). Although clues pertaining to the mechanisms involved in secondary axon degeneration in AIDP are extremely limited, there is evidence supporting disturbed nodal architecture and cytoskeletal organization. The organization of voltage-gated sodium channels, AnkyrinG and often voltage-gated potassium channels are disrupted in guinea pig myelin-induced EAN rats prior to the loss of Neurofascin-186 (NF-186) and Gliomedin from the node of Ranvier [343]. Interestingly, these mice generate autoantibodies against NF-186 and Gliomedin suggesting a potential pathogenic mechanism for AIDP [343]. Additionally, increased neurofilament density has been observed in peripheral nerve autopsied axons from an AIDP patient [84].

2.2.2. CIDP—CIDP is a demyelinating autoimmune disorder that is caused by an improper immune response targeting components of peripheral nerves and nerve roots [377]. The mechanism triggering disease onset is unknown, but it is not expected to involve an infectious event [377]. Several phenotypic variants of CIDP exist but patients generally exhibit proximal and distal weakness of extremities due to a relapsing or progressive

neuropathy that develops over a duration of more than eight weeks with a variable age of onset [377]. Patient histological and electrophysiological evaluation of peripheral nerves reveals myelin deficits including loss of myelinated fibers, de- and remyelination, thin and occasionally absent myelin sheaths, variable onion bulb formation, slowed MCV and prolonged distal and F-wave latencies [377] (Supplemental Table 1). Interestingly, although ample evidence suggests an autoimmune mechanism the target antigen in most CIDP patients has not been identified even though antibodies against integral peripheral myelin components (i.e. MPZ, Peripheral Myelin Protein 2 (PMP2) and PMP22) are relatively common [377]. These compact myelin component autoantibodies are frequently insufficient for triggering CIDP but recent attention has been given to autoantibodies targeting components of the node and paranode (CASPR, CNTN1, Neurofascin-155 [NF-155]) given their implication in a small fraction of CIDP patients [132, 377, 607]. Patients with CASPR, CNTN1 and NF-155 autoantibodies are thought to fall into a distinct disease category but remain under the CIDP umbrella given that they respond to some immunomodulatory therapies [108, 126, 132, 472]. Although mechanisms triggering CIDP remain unclear, there is electrophysiological and histological evidence for secondary axon degeneration in CIDP patients, which often correlates with clinical deficits [71, 296, 413] (Table 1). Biopsied sural, peroneal and radial nerves and autopsied peripheral nerves from CIDP patients revealed axon loss, active axonal degeneration, axon sprouts/clusters of regenerating axons and myelin ovoids (Table 1). EAN rodent models that demonstrate CIDP-like pathology have been developed and there is electrophysiological and histological evidence for secondary axon degeneration in multiple models (Table 1). Clues about the mechanisms causing secondary axon degeneration in CIDP are accumulating. Superficial peroneal nerve biopsy axons from an unclassified CIDP patient with unknown autoantibodies exhibit disturbed nodal architecture with disorganized voltage-gated sodium channels, paranodin and voltage-gated potassium channels [101]. Voltage-gated sodium channels organization, as well as CASPR and NF organization, are also disturbed in sural nerve biopsy axons from an unclassified CIDP patient with CASPR IgG4 autoantibodies [132]. Similarly, CASPR, CNTN1 and NF-155 organization was disrupted in peripheral nerve axons of rats treated with CNTN1 IgG4 antibodies [369]. Additionally, increased neurofilament and microtubule densities and accumulated mitochondria have been observed in sural, peroneal and radial nerve biopsied axons from unclassified CIDP patients with unknown autoantibodies [468, 478, 604]. Intriguingly, increased neurofilament phosphorylation as well as reduced neurofilament and mitochondria densities have also been observed in CIDP patient peripheral nerve axons [164, 413, 468]. Taken together, these findings suggest that nodal architecture, axonal cytoskeletal organization and axonal cargo transport are disturbed in CIDP.

3. Conclusions and Challenges

The role of secondary axon degeneration in the pathogenesis of demyelinating neuropathies is becoming more apparent. There is abundant evidence for the occurrence for secondary axon degeneration in CMT1. Classification of CMT is difficult given that mutations within the same gene can cause different forms of disease and limitations of our current electrophysiological and histological methods to detect primarily myelin versus axonal

effects. However, it seems certain that myelinating SCs are the primary manifestation of disease when the mutated gene is a myelinating SC-specific or -enriched gene (i.e. *PMP22, MPZ, EGR2* and *GJB1*). Dysregulated expression of additional myelin genes including *myelin associated glycoprotein* (*MAG*) and *NF* (specifically, isoform NF-155) in mouse models also results in secondary axon degeneration. Histological evidence of secondary axon degeneration has been observed in MAG knockout mice accompanied by increased axonal neurofilament densities and disrupted nodal architecture [166, 633]. Electrophysiological evidence of secondary axon degeneration has been reported in SC-specific NF-155 knockout mice along with disrupted nodal architecture and axonal cargo transport [458]. Additionally, mitochondrial and miRNA processing dysfunction in SCs causes myelin deficits and secondary axon degeneration in rodent models [452, 594, 595]. It is also evident that secondary axon degeneration is a common feature of demyelinating inflammatory neuropathies. Serum IgG from a large proportion of AIDP and CIDP patients targets the nodes or paranodes of peripheral nerve axons suggesting that disrupted nodal architecture and paranodal myelin-axonal contacts contributes to AIDP and CIDP pathogenesis [576]. Further advances are needed to fully comprehend the mechanisms that cause myelin deficits in AIDP and CIDP. Although there is strong support for axon integrity being dependent upon proper myelinating SC function, the precise mechanisms that lead up to and trigger axon degeneration upon demyelination remain unclear. Additionally, teasing out the myelin dependent versus independent functions of SCs and the developmental versus degenerating effects of myelin dysfunction on axon integrity will be insightful. Advances in these inquiries will greatly expand our knowledge of disease pathogenesis and prognosis and aid therapy development.

Supplementary Material

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Abbreviations

AIDP	Acute Inflammatory Demyelinating Polyneuropathy
ATP	Adenosine Triphosphate
CNS	Central Nervous System
CMT	Charcot-Marie-Tooth Disease
CMT1	Charcot-Marie-Tooth Disease, Type 1
CMT1A	Charcot-Marie-Tooth Disease, Type 1A
CMT1B	Charcot-Marie-Tooth Disease, Type 1B
CMT1C	Charcot-Marie-Tooth Disease, Type 1C

CMT1D	Charcot-Marie-Tooth Disease, Type 1D
CMT1E	Charcot-Marie-Tooth Disease, Type 1E
CMT1F	Charcot-Marie-Tooth Disease, Type 1F
CMT1X	Charcot-Marie-Tooth Disease, Type 1X
CMT2	Charcot-Marie-Tooth Disease, Type 2
CIDP	Chronic Inflammatory Demyelinating Polyneuropathy
iSCs	Immature Schwann Cells
CNTF	Ciliary Neurotrophic Factor
CMAP	Compound Muscle Action Potential
Cx32	Connexin 32
CNTN1	Contactin-1
CASPR	Contactin Associated Protein-like 1
DADS	Distal Acquired Demyelinating Symmetric Neuropathy
EGR2	Early Growth Response 2
ER	Endoplasmic Reticulum
EAN	Experimental Autoimmune Neuritis
GJB1	Gap Junction Protein Beta 1
GBS	Guillain-Barré Syndrome
HNPP	Hereditary Neuropathy with Liability to Pressure Palsies
LITAF	Lipopolysaccharide Induced TNF Factor
MCV	Motor Conduction Velocity
MADSAM	Multifocal Acquired Demyelinating Sensory and Motor Neuropathy
MAG	Myelin Associated Glycoprotein
MPZ	Myelin Protein Zero
NCV	Nerve Conduction Velocity
NF	Neurofascin
NF-155	Neurofascin-155
NF-186	Neurofascin-186
NEFL	Neurofilament Light

NT-3	Neurotrophin-3
NAD^+	Nicotinamide Adenine Dinucleotide
PRX	Periaxin
PMP2	Peripheral Myelin Protein 2
PMP22	Peripheral Myelin Protein 22
PNS	Peripheral Nervous System
SC	Schwann Cell
SCPs	Schwann Cell Precursors
SCV	Sensory Conduction Velocity
SNAP	Sensory Nerve Action Potential
SARM1	Sterile Alpha and TIR Motif Containing 1
WD	Wallerian Degeneration
WLD	Wallerian-like Degeneration

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Highlights

- Summary of the role of myelin and myelinating Schwann cells
- Review of the effects of myelination and demyelination on axon integrity and axon degeneration pathways
- Review of secondary axon degeneration in demyelinating Charcot- Marie-Tooth disease
- Review of axon degeneration in acquired demyelinating neuropathies, GBS and CIDP

Table 1.

Electrophysiological and histological evidence for secondary axon degeneration in demyelinating neuropathy patients and rodent models. Nerve conduction studies with evoked potentials were included as electrophysiological evidence; reduced compound muscle action potential (CMAP) amplitudes (↓CMAP), reduced sensory nerve action potential (SNAP) amplitudes (↓SNAP), reduced CMAP & SNAP Amplitudes (↓CMAP & ↓SNAP), reduced compound action potentials (↓CAP) and not specified (NS). Peripheral nerve biopsies demonstrating features of axon degeneration were included as histological evidence; axonal atrophy (AA), active axon degeneration (AD), axonal loss (AL), sxonal swellings (AS), axon sprouts/clusters of regenerating axons (ASC), bands of Büngner (BB) and myelin ovoids (MO). Reports inaccessible due to language barriers were excluded. Additional abbreviations: distal acquired demyelinating symmetric neuropathy (DADS), deletion (del), duplication (dup), experimental autoimmune neuritis (EAN), GM1 ganglioside (GM1), heterozygous (het), insertion (ins), knockout (KO), knockin (KI), multifocal acquired demyelinating sensory and motor neuropathy (MADSAM), not applicable (N/A), no reports (NR), super enhancer (SE), transgenic (Tg).

Disease	Subtype	Sample	Electrophysiological Evidence for Secondary Axon Degeneration	Histological Evidence for Secondary Axon Degeneration	Mechanisms Involved
CMT1A (17p11.2-p12 Duplication [*PMP22* Duplication])	N/A	Patients	**↓CMAP** [4, 39, 50, 61, 148, 173, 240, 270, 288, 297, 311, 323, 416, 419, 433, 451, 581, 585, 596, 623] **↓SNAP** [563] **↓CMAP & ↓SNAP** [27, 48, 49, 59, 94, 129, 137, 176, 195, 214, 223, 224, 228, 231, 293, 368, 370, 378, 424, 465, 509, 521, 593, 602, 634]	**AA** [562] **AD** [421] **AL** [214, 378] **ASC** [85, 214, 501, 563, 623]	Trophic support [494, 597], vulnerable to toxic insults [9, 102, 191, 241]
		Rodents	**↓CMAP:** *PMP22* Tg Mice (C61 [279, 284], TgN248 [364]), *PMP22* Tg Rats [159, 192] **↓CMAP & ↓SNAP:** *PMP22* Tg Mice (C3-PMP [592], C22 [592]), *PMP22* Tg Rats [514]	**AA:** *PMP22* Tg Mice (C3-PMP [592], C22 [480, 592]), *PMP22* Tg Rats [192] **AD:** *PMP22* Tg Rats [514] **AL:** My41 *PMP22* Tg Mice [480], *PMP22* Tg Rats [159] **ASC:** C61 *PMP22* Tg Mice [279] **BB:** *PMP22* Tg Mice (C22 [480], My41 [480])	Nodal architecture [284], cytoskeletal organization [157, 192, 423, 489, 492, 500], cargo transport [489, 492, 592], trophic support [422, 494, 597]
HNPP (17p11.2-p12 Deletion [*PMP22* Deletion])	N/A	Patients	**↓CMAP** [55, 80, 109, 177, 217, 240, 294, 429, 591, 618] **↓SNAP** [19, 66, 110, 113, 185, 236, 299, 309, 319, 321, 342, 344, 351, 397, 404, 412, 439, 443, 460, 467, 567, 575, 615, 632] **↓CMAP & ↓SNAP** [16, 17, 20, 23, 40, 53, 56, 88, 111, 112, 119, 152-154, 160, 186, 189, 193, 201, 210, 216, 219, 223, 226, 260, 268, 269, 285, 327, 367, 431, 435, 448, 455, 519, 533, 573] **NS** [36, 438, 515, 603]	**AA** [226, 562] **AD** [226, 285, 351, 533, 603] **AL** [299, 351] ASC [216, 351, 515, 573, 603] **MO** [603]	Cytoskeletal organization [124], vulnerable to toxic insults [253]
		Rodents	**↓CMAP:** *PMP22* Het KO Mice [7, 229, 651]	**AL:** *PMP22* SE Het del Mice [442]	NR
CMT1B (*MPZ* Point Mutation[a])	Infantile-onset	Patients	**↓CMAP:** p.I30S [393], p.I30T [161], p.T65N [77], p.H81Q[b] [99], p.H81R[b] [151], p.R98C [30, 32], P.G103W [77], p.S121F [516], p.G123D [72], p.T124K [282], p.C127Y [144], p.D128N[b] [151], p.K130R [635], p.N131K [280],	**AL:** p.R98C [32], p.G167R [560] **AD:** p.D90E[b] [60, 358], p.R98C [612], p.K130R [635] **ASC:** p.H81Q[b] [99], p.R98C	Nodal architecture [32], cytoskeletal organization [387]

Disease	Subtype	Sample	Electrophysiological Evidence for Secondary Axon Degeneration	Histological Evidence for Secondary Axon Degeneration	Mechanisms Involved
			p.P132L [135], p.D134E [30], p.I135T [135], p.G137R [532], p.L175SfsX74 [612], p.L184AfsX50 [553], p.L184AfsX51 [537], p.A209EfsX24 [30], p.Q215X [366] **↓CMAP &** **↓SNAP:** p.P105L [346], p.N131S [232], p.G137_K149del [559], p.K207X [652] **NS:** p.D90E[b] [358]	[317], p.G167R [560] **MO:** p.D90E[b] [60]	
		Rodents	**↓CMAP:** p.R98C KI Mice [505]	NR	NR
	Adolescent-onset	Patients	**↓CMAP:** p.V58D [317], p.T65A [281], p.R98H[b] [307, 540], p.Q100X [393], p.V102CfsX11[b] [107], p.D109N [307], p.I135M [336], p.Q187PfsX63 [336] **↓SNAP:** p.V102CfsX11[b] [107] **↓CMAP & ↓SNAP:** p.K48Q[b] [77], p.E97AfsX5[b] [77], p.K214M [35], p.K236del [541], c.645 + 1_G>T[b,d] [77, 275] **NS:** p.G123S[b] [318]	**AL:** p.V58D [317], p.R98H[b] [307] **ASC:** p.R98H[b] [307], p.D109N [307], p.G123S[b] [318], p.S233RfsX18 [317]	NR
		Rodents	**↓CMAP:** p.S63del Tg Mice [619]	**AD:** p.S63del Tg Mice [619]	NR
	Adult-onset	Patients	**↓CMAP:** p.H39P [325, 539], p.S51F [640], p.S54VfsX5[d] [92], p.I62M [26], p.E71X [306], p.S78L[b] [150, 151, 262, 381, 640], p.H81L [339], p.F95L [427], p.I99T [131], p.T124A [365], p.T124M[c] [301, 302, 314, 339], p.L144RfsX18[534], p.Y145LfsX4 [365], p.Y145S[c] [313, 543], p.G163R [140] **↓SNAP:** p.S20F [156], p.D104TfsX14 [116] **↓CMAP & ↓SNAP:** p.D35N [72], p.R36G [114], p.R36W [81], p.H39P [266], p.V46M [77], p.S55I [275], p.E56K [283], p.D60H [27], p.P70S [310], p.D75V [394], p.Y82H [58], p.D104TfsX14[c] [353, 445], p.P105T [251], p.R106C [375], p.D109E [503], p.N116S [275], p.D121N [136], p.T124M [74, 91, 118, 174, 207, 394, 513, 547, 565, 570], p.V136G[c] [475], p.T143R [77], p.Y145X [363], p.V146G [77], p.V150 S195del [486], p.P151AfsX3 [456], p.D224Y[c] [147, 508], p.H225QfsX10[b] [215], p.R227G [523]	**AA:** p.D61G [513], p.Y119C [513], p.T124M [394, 513] **AL:** p.S20F [156], p.D35Y [376], p.S51F [640], p.D61G [513], p.I99T [131], p.Y119C [513], p.T124M [32, 91, 118, 207, 301, 513], p.D224Y [508], **AD:** p.D35Y [376], p.D61G [513], p.P70S [310], p.Y119C [513], p.T124M [91, 301, 302, 394, 513], p.D224Y [508] **ASC:** p.R36G [114], p.H39P [266, 325], p.S44F [151], p.D61G [513], p.P70S [310], p.E71X [306], p.Y82H [58], p.I99T [131], p.D104TfsX14[c] [445], p.Y119C [513], p.T124M [32, 74, 91, 118, 151, 207, 301, 394, 513, 547, 638], p.D224Y [508], p.R227G [523] **BB:** p.D61G [513], p.Y119C [513], p.T124M [513], p.R227G [523] **MO:** p.R36G [114], p.T124M [301], p.R227G [523]	Nodal architecture [325], cytoskeletal organization [207]
		Rodents	NR	NR	NR
CMT1C (*LITAF* Point Mutation)	N/A	Patients	**↓CMAP:** p.T49M[f] [495], p.G112S[g] [244] **↓SNAP:** p.G112S [265] **↓CMAP & ↓SNAP:** p.I92V [506], P.G112S [495], p.P135R [103], p.V144M [182]	**AL:** p.T49M[f] [495], p.P135R [103] **ASC:** p.P135R [103]	NR
		Rodents	**↓CMAP:** p.W116G Tg Mice [316]	**AD:** p. W116G Tg Mice [316]	Nodal architecture

Disease	Subtype	Sample	Electrophysiological Evidence for Secondary Axon Degeneration	Histological Evidence for Secondary Axon Degeneration	Mechanisms Involved
					[316], cargo transport [316]
CMT1D (*EGR2* Point Mutation)	N/A	Patients	**↓CMAP:** p. I268N[c] [613], P.R359W [100, 564], p.R381C [637], p.R381H [447], p.S382R + p.D383Y [613], p.D411G [194] **↓SNAP:** p.R353G [415], p.R359W [178], p.T387N [522] **↓CMAP & ↓SNAP:** p.R359Q [391], p.R381C [75], p.R409Q [517], p.R409W [322], p.E412G [487, 568] **NS:** p.R381C [611]	**AL:** p.R359W [100, 564], pE412G [568] **AD:** pE412G [568] **ASC:** p.D355V [411], pE412G [568]	Cytoskeletal organization [564, 613], vulnerable to toxic insults [415]
		Rodents	N/R	N/R	Nodal architecture [34]
CMT1E (*PMP22* Point Mutation)	N/A	Patients	**↓CMAP:** p.S7YfsX30 [326], p.L18R [362], p.S22F [484], p.W28X [87], p.W39C [557], p.S72L [371, 531], p.L78P [461], p.Q86X [335], p.T99_G100del [290], p.Q103X [461], p.I104FfsX7 [335], p.G107VfsX4 [215], p.C109R [149], p.V110_I116dup [290], p.S112R [252], p.A115_T118del [499], p.T118M[c,h] [528], p.Y136_A139del [598], p.G150D [237], p.X161WextX10[c] [644], c.178 + 2T>C [169], c.179 - 1G>A [261], c.319 + 1G>A [335], c.417 + 2T>G [609], 17p11.2-p12 del/p.W61X [248], 17p11.2-p12 del/p.T118M[h] [528], 17p11.2-p12 del/Exon 2+3 del[i] [14], 17p11.2-p12 del/Exon 5 del[i] [1, 113] **↓SNAP:** p.S7YfsX30 [420], p.C42R [335], p.R95QfsX128 [321], p.G100EfsX11 [403], p.I116TfsX5[558], p.L145PfsX78 [646], Exon 4+5 del[i] [552, 587] **↓CMAP & ↓SNAP:** p.L4RfsX3 [405], p.S22F [277], p.T23R [356], p.S76TfsX35 [350], p.R95QfsX128 [120], p.Y97TfsX14 [643], p.C109X [3], p.T118M[h] [484], p.S131C [484], p.L145RfsX10 [55], p.R159C [183], c.78 + 5G>A[c] [622], c.79 - 2A>G [290], c.179 + 1G>C [43], Exon 5 del[i] [1, 86], 17p11.2-p12 del/p.T118M[h] [243]	**AA:** p.V30M [491], c.179 - 1G>C [389] **AL:** p.L18R [362], p.T23R [246], p.S149R [430], 17p11.2-p12 del/Exon 2+3 del[i] [14] **AD:** p.S72L [65], p.R159C [183] **AS:** p.R159C [183] **ASC:** p.S76TfsX35 [350], p.R95QfsX128 [120], p.C109X [3], p.R159C [183], c.179 - 1G>C [389], 17p11.2-p12 del/ p.R157G [425] **MO:** p.C109X [3]	Cytoskeletal organization [3, 491]
		Rodents	**↓CMAP:** TremblerJ (p.L16P) Mice [128, 384, 385, 493]	**AA:** TremblerJ (p.L16P) Mice [480] **AD:** Trembler (p.G150D) Mice [220, 361] **AL:** TremblerJ (p.L16P) Mice [479], Trembler-m1H (p.H12R) Mice [238], Trembler-m2H (p.Y153X) Mice [238], Trembler-m3H (p.S72T) [238] **BB:** TremblerJ (p.L16P) Mice [480]	Nodal architecture [128, 198, 418, 477, 482, 610], cytoskeletal organization [121-123, 220, 263, 273, 347, 463, 491, 493, 544], cargo transport [121-123, 463, 482], trophic support [165, 494]

Disease	Subtype	Sample	Electrophysiological Evidence for Secondary Axon Degeneration	Histological Evidence for Secondary Axon Degeneration	Mechanisms Involved
CMT1F (*NEFL* Point Mutation)	N/A	Patients	**↓CMAP:** p.P8L [247], p.P8Q [247], p.P8R [117, 225, 247, 337, 392], p.P22S [142, 181, 337], p.E90K [247], p.L93P [392], p.N98S [54, 225, 247, 354, 630, 639], p.E163X[c] [167], p.L268P [143], p.L311P [225], p.Q333P [388], p.Y389C [213], p.E396K [141, 337, 459] **↓CMAP & ↓SNAP:** p.P22R [524], p.P22S [57, 359], p.P22T [639], p.E210X[c] [642], p.C322_N326del [143], p.E396K [52, 143], p.E397K [653], p.R421X [10]	**AA:** p.P22S [142], p.E396K [141] **AD:** p.C322_N326del [143], p.E396K [141], p.E397K [653] **AL:** p.P8R [225], p.N98S [225, 354], p.N272K [134] **AS:** p.P22S [142, 359], p.N98S [354], p.L268P [143], p.N272K [134], p.C322_N326del [143], p.E397K [653], p.P440L [45] **ASC:** p.P8R [225], p.P22S , p.E90K [2, 247], p.N98S [2], p.E140X[c] [2], p.E163X[c] [167], p.E210X[c] [642], p.L268P [143], p.N272K [134], p.C322_N326del [143], p.Y389C [213], p.E396K [2, 141, 143], p.E397K [653], p.P440L [45] **BB:** p.E397K [653] **MO:** p.E90K [247], p.P440L [45]	Cytoskeletal organization [45, 134, 142, 143, 167, 359, 642, 653], cargo transport [141, 143, 213, 359, 653]
		Rodents	**↓CAP:** p.N98S KI Mice [308]	**AD:** p.N98S KI Mice [308] **AL:** p.N98S KI Mice [308], p.E396K Tg Mice [599] **ASC:** p.N98S KI Mice [308]	Cytoskeletal organization [6, 127, 308, 504, 648], cargo transport [127, 308, 504, 648]
CMT1X (*GJB1* Point Mutation or Deletion[j])	N/A	Patients	**↓CMAP:** p.M1I [572], p.T4K [393], p.L6S [572], p.L9F [78], p.S11C [93], p.N14K [93], p.R15L [349], p.R15Q [82, 203], p.R15W [259], p.H16L [262], p.I20F [572], p.G21D [349], p.W24C [315], p.S26L [203, 550, 572], p.S26W [138, 640], p.R32G [262], p.V35L [334], p.A39S [349], p.S49Y [572], p.I52TfsX31 [393], p.T55I [203, 259], p.T55R [315], p.V63I [561], p.C64F [640], p.C64Y [257, 550], p.Y65C [349], p.Y65H [510], p.S72F [332], p.R75P [395], p.V91M [63, 572], p.M93K [393], p.V95M [399], p.I101RfsX8 [572], p.E102X [203, 554, 640], p.E109X [315], p.V120E [259], P.V125D [315], p.I127M [332], p.V139M [203], p.R142Q [93], p.R142W [349], p.L144del [572], p.F153L [572], p.Y160H [262], p.R164K [437], p.R164Q [259], p.R164W [349, 525, 640], p.V170D [262], p.C173Y [572], p.V177del [650], p.C179G [402], p.R183C [203, 572], p.R183H [349, 572], p.E186K [349, 561], p.T191fs[j] [315], p.I202D [341], p.E208K [203], p.Y211X [203], p.L212F [78], p.R215P [572], P.R215W [550, 640], p.R220X [572], c.-5413_-49del [298], c.-529 T>C [571], c.-146-90_-146-89insT [566], c.-17 + 1G>T [566], c.*15 C>T [95], *GJB1* Deletion[j] [11, 414] **↓SNAP:** p.N2S [340], p.H16L [606], p.E102del [616], p.F153S [315], p.S198A [393] **↓CMAP & ↓SNAP:** p.MIR [78], p.N2K [96],	**AA:** p.N2K [96], p.R15W [511], p.R22Q [511], p.E102G [490], p.I127S [605] **AL:** p.R15W [617], p.D46G [449], p.L108P [65], p.Y211H [63], p.F235C [333] **AD:** p.H16L [606], p.R22Q [561], p.S26L [203], p.V63I [561], p.P87L [203, 300], p.H94R [600], p.V95M [600], p.I127S [605], p.L156AfsX37 [579], p.R183C [203, 600], p.E186K [561], p.E208K [203], p.Y211X [203, 205], p.R215W [600], p.R219C [600], c.-17 G>A [409], *GJB1* Deletion[j] [203, 414, 561] **ASC:** p.N2K [96], p.R15Q [82, 203], p.R15W [200, 511, 512], p.H16L [606], p.R22Q [511, 512, 561], p.R22X [62], p.A39V [512], p.F69L [638], p.P87L [300], p.E102G [490], p.I127F[k] [470], p.I127S [605], 561], p.S128X [638], p.V140E [278], p.L156AfsX37 [579], p.P158A [62], p.D178G[k] [470], p.E208K [203, 204], p.Y211X [205], p.P227SfsX16 [512], c.-5413_-49del [298], c.-459 C>T [329], c.-215 G>A [624], c.-19 C>G [45], c.-17 G>A [409, 566], *GJB1* Deletion[j] [11, 203, 414] **MO:** c.-17 G>A [409] **NS:** p.V91M [390], p.T191fs[j] [315]	Cytoskeletal organization [203, 204, 490], cargo transport [490], vulnerable to toxic insults [464]

Disease	Subtype	Sample	Electrophysiological Evidence for Secondary Axon Degeneration	Histological Evidence for Secondary Axon Degeneration	Mechanisms Involved
			p.W3G [352], p.W3X [340], p.T8I [130], p.N14S [542], p.R15Q [550], p.R15W [200, 511], p.I20T [349], p.R22Q [511, 561], p.R32K [33], p.I33N [382, 616], p.V35M [332], p.V37GfsX47 [338], p.V38A [258], p.A40T [360], p.E41D [410], p.N54H [536], p.N54S [616], p.C60Y [22], p.V63F [78], p.I71S [626], p.R75CfsX8 [72], p.L81P [340], p.P87L [300], p.L90P [340], p.A88D [349], p.M93R [616, 649], p.V95M [550], p.A96V [190], p.Q99_H100insQ [488], p.H100Q [257], p.H100Y [197], p.L106P [340], p.R107MfsX10 [485], P.I127S [605], p.I127F [349], p.I127N [349], p.I127T [230], p.W133GfsX63 [496, 616], p.Y135LfsX12 [349], p.V137A [551], p.V140E [278], p.R142E [334], p.R142Q [548], p.L143P [278], p.L144W [190], p.F145C [340], p.A147PfsX49 [386], p.F149L [627], p.L156AfsX37 [579], p.R164Q [550, 616], p.R164W [550], p.L165Q [190], p.V170F [257], p.P172L [550], p.C179Y [72], p.S182T [332], p.R183C [349], p.T188I [334], p.T191_F193dup [590], p.F193S [190], p.A197V [349], p.L204F [550], p.N205S [31], p.Y211X [205], p.R224L [190], p.S277GfsX128 [393], p.R238C [373], p.K260E [257], p.I262TfsX13 [67], c.-529 T>C [41], c.-459 C>T [329, 571], c.-215 G>A [624], c.-170 T>G [357], c.-103 C>T [566], c.-17 G>A [409, 566], c.*15 C>T [566], *GJB1* Deletion[i] [83, 561] **NS:** p.Y211H [63], p.S62R [330], p.S128L [380], p.R142Q [212], p.L156R [621], p.V177A [18]		
		Rodents	**↓CMAP:** Cx32 KO Mice [21]	**AL:** Cx32 KO Mice [507] **ASC:** Cx32 KO Mice [21, 507]	Cytoskeletal organization [490, 507, 589], cargo transport [490, 589]
AIDP[1] (Acute Immune Response Targeting PNS)	N/A	Patients	**↓CMAP:** [47, 227, 239, 254, 264, 400, 444, 535, 549] **↓SNAP:** [76, 304, 578] **↓CMAP & ↓SNAP:** [13, 15, 37, 51, 89, 125, 175, 196, 436, 518, 582, 608, 629] **NS:** [469]	**AD:** [47, 51, 84, 202, 295, 348, 406, 469] **AL:** [51, 84, 406] **ASC:** [47, 51] **BB:** [51, 406] **MO:** [254, 406, 469]	Cytoskeletal organization [84]
		Rodents	**↓CMAP:** Severe EAN Mice [625] **↓SNAP:** AIDP Patient IG Mice [115] **↓CAP:** Guinea Pig Myelin EAN Rats [343]	**AL:** Severe EAN Mice [625] **AD:** Guinea Pig Myelin EAN Rats [343], PMP2 EAN Rats [245, 343]	Nodal architecture [343]
CIDP (Chronic Immune Response Targeting PNS)	Mixed/Not Classified	Patients	**↓CMAP:** Unknown Ig [188, 233, 444, 473] **↓CMAP & ↓SNAP:** CNTN1 IgG4 [289, 471], NF-155 IgG4 [289, 428], Unknown Ig [38, 199, 211, 289, 428, 432, 586] **NS:** GM1 IgM [255], IgGγ [601], IgGκ [601], IgMγ [601], IgMκ [601], IgGκ + IgMκ [601]	**AD:** CASPR IgG4 [132], IgGγ [601], IgGκ [601], IgMγ [601], IgMκ [601], IgGκ + IgMκ [601], Unknown Ig [28, 38, 139, 188, 209, 271, 295, 355, 413, 432, 457, 468, 497] **AL:** CASPR IgG4 [132], CNTN1 IgG4 [289], Unknown Ig [71, 164, 188, 296, 355] **ASC:** Unknown Ig [71, 101, 271, 296, 355, 457, 468, 586,	Nodal architecture [101, 132], cytoskeletal organization [164, 413, 468, 478, 604], cargo transport [604]

Disease	Subtype	Sample	Electrophysiological Evidence for Secondary Axon Degeneration	Histological Evidence for Secondary Axon Degeneration	Mechanisms Involved
				604] **MO:** Unknown Ig [604]	
	Typical	Patients	↓**CMAP:** Unknown Ig [155] ↓**CMAP & ↓SNAP:** Unknown Ig [234]	**AD:** CNTN1 IgG4 [133, 286], NF-155 IgG4 [286], Unknown Ig [234, 286] **AL:** CNTN1 IgG4 [133, 286], NF-155 IgG4 [286], Unknown Ig [286] **ASC:** Unknown Ig [234, 584] **MO:** CNTN1 IgG4 [286], NF-155 IgG4 [286]	NR
	DADS	Patients	↓**CMAP:** Unknown Ig [155, 434] ↓**CMAP & ↓SNAP:** Unknown Ig [234]	**AD:** NF-155 IgG4 [286], Unknown Ig [234, 286, 434] **AL:** NF-155 IgG4 [286], Unknown Ig [286] **ASC:** Unknown Ig [234, 584] **MO:** NF-155 IgG4 [286], Unknown Ig [434]	NR
	MADSAM	Patients	↓**CMAP & ↓SNAP:** Unknown Ig [234]	**AD:** Unknown Ig [234, 286] **AL:** Unknown Ig [286] **ASC:** Unknown Ig [234]	NR
	Pure Motor	Patients	↓**CMAP:** IgE [272]	NR	NR
	Pure Sensory	Patients	↓**CMAP & ↓SNAP:** Unknown Ig [234]	**AD:** Unknown Ig [234, 286] **ASC:** Unknown Ig [584]	NR
	N/A	Rodents	↓ **CMAP:** B7-2–Deficient Nod Mice [498, 577], Bovine Peripheral Nerve Myelin Immunized Rats [250], PMP2 EAN Rats [218] ↓ **CAP:** CNTN1 IgG4 Treated Rats [369]	**AD:** B7-2–Deficient Nod Mice [577], Bovine Peripheral Nerve Myelin Immunized Rats [250], PMP2 EAN Rats [483] **AL:** B7-2–Deficient Nod Mice [577]	Nodal architecture [369]

[a] MPZ amino acid number corresponds to the full-length protein containing the leader peptide.

[b] The same MPZ mutation occasionally causes a spectrum of disease onset. These mutations were included in the most common or earliest disease onset group in the table: infantile- and adolescent-onset (p.H81Q, p.H81R, p.D90E), adolescent- and adult-onset (p.K48Q, p.E97AfsX5, p.V102CfsX11, p.G123S, p.H225QfsX10, c.645 + 1G>T) and infantile-, adolescent- and adult-onset (p.S78L, p.R98H, p.D128N).

[c] Mutations are generally heterozygous but homozygous mutations has also been identified and they frequently present with more severe CMT than their heterozygous relatives.

[d] Some patients were also diagnosed with well-controlled diabetes.

[e] Insufficient details were provided to accurately determine disease onset age.

[f] LITAF p.T49M pathogenicity is controversial [41].

[g] A PRX variant (p.R187C) of unknown significance in was also identified in this patient.

[h] PMP22 p.T118M pathogenicity is controversial [417, 641].

[i] Proper nomenclature could not be established based on reported mutation.

[j] Both male and female CMT1X patients were included.

[k] The abstracts of some reports that were inaccessible due to language barriers contained sufficient information to be included. However, it is unclear if the full text provides additional evidence.

[l] Reports demonstrating a lack of denervation by electromyography without supporting histological evidence for secondary axon degeneration were excluded.