What is Neuropathy? 
Definition, Symptoms, Causes, and Diagnosis

Sara Austin, MD 
Neurologist in Austin, TX
1 in 15 Americans have neuropathy

= 20 million people

<table>
<thead>
<tr>
<th>Condition</th>
<th>Number</th>
</tr>
</thead>
<tbody>
<tr>
<td>Alzheimer’s Disease</td>
<td>5,400,000</td>
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<tr>
<td>Epilepsy</td>
<td>3,000,000</td>
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<tr>
<td>Rheumatoid Arthritis</td>
<td>1,300,000</td>
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<tr>
<td>HIV</td>
<td>1,200,000</td>
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<tr>
<td>Parkinson’s Disease</td>
<td>500,000</td>
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<tr>
<td>Multiple Sclerosis</td>
<td>400,000</td>
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<tr>
<td><strong>Total</strong></td>
<td><strong>11,800,000</strong></td>
</tr>
</tbody>
</table>

Sources: The Foundation for Peripheral Neuropathy, Alzheimer’s Association, Epilepsy Foundation Statistics, American College of Rheumatology, Parkinson’s Disease Foundation, National Multiple Sclerosis Society.
Nervous System: Central vs. Peripheral

Neuro = nerve
Pathy = sickness
Types of Peripheral Nerves

Motor nerves — from spinal cord to muscles

Sensory nerves — from sensory organelle to spinal cord
  Large fiber- position sense and balance
  Small fiber- pain and temperature

Autonomic nerves — control blood pressure, sweating, bladder function, heart rate, digestion, etc.
Symptoms

Where

• Feet, legs
• Hands, arms
• Internal organs
Symptoms – Motor Nerves Affected

- Weakness
- Cramping and muscle twitch
- Muscle or bone loss
- Loss of ankle reflexes, foot drop
- Changes in skin, hair, and nails
Symptoms – Sensory Nerves Affected

Damage to large fiber myelinated nerves:

• Impaired sense of:
  – Proprioception (position sense)
  – Balance, especially when eyes are closed
  – Vibration
  – Light touch discrimination

• Will show up in Nerve Conduction Study
Symptoms – Sensory Nerves Affected

Damage to small fiber nerves (don’t have myelin):

- Pain - loss of pain or hypersensitive (mostly in feet)
- Impaired sense of temperature
- Not picked up on Nerve Conduction Study- skin biopsy can diagnose properly
- Most common cause: early diabetes
Symptoms – Autonomic Nerves Affected

- Digestive problems – constipation, diarrhea, difficulty eating
- Bladder dysfunction
- Dry eyes or mouth
- Reduced or inability to sweat, heat intolerance
- Dizziness, lightheadedness due to blood pressure
- Difficulty breathing or heart problems
- Impotence
Effects of Neuropathy

• Limits ability to function
• Limits independence
• Promotes **falls** and slow wound healing
• Causes **problems with sleep**
• Can lead to **depression and social isolation**
Top Causes of Neuropathy

- Diabetic 33%
- Idiopathic 33%
- Other 34%

Over 100 different causes
Causes for Neuropathy

- Diabetes and Impaired Glucose Tolerance – 33%  
  - Over 50% of individuals with diabetes will have mild to severe forms of neuropathy  
  - Severity is related to how long nerves are exposed to hyperglycemia
Causes for Neuropathy

• Other – 34%

• Inherited disorders (Charcot-Marie Tooth)
• Toxins – alcohol, heavy metals chemicals
• Vitamin Deficiencies (B-12, D, Thiamine, E)
• Underactive thyroid gland
• Auto-Immune Disease/Inflammation
• Chemotherapy medications
• Metabolic disease (kidney or liver)
• Infections - HIV, Lyme disease, shingles
• Trauma or pressure on nerves
• Tumor
Causes - continued

• Idiopathic – 33%
Can Nerves Regenerate?

- **Axonal neuropathies** - hard to grow back

- **Demyelinated neuropathies** - can grow back with treatment
Proper Diagnosis is Key

• Board Certified Neuromuscular Trained Neurologist

• History and Physical Tests—help to identify source
  • Family History, time course, current medications taken, habits, other medical problems, symptoms
  • Sharp/light touch – determine nerves affected, reflexes and strength tests
Proper Diagnosis is Key

Based on your symptoms you may have some of these routine blood tests:

- Metabolic profile
- Complete blood count
- Lipid profile
- Serum/urine immunofixation
- Quantitative immunoglobulins
- Anti-sulfatide antibodies
- Thiamine, B12, B6
- ANA, SS-A/SS-B, RF, ESR
- Cryoglobulins
- **Glucose Tolerance Test**, HGBA1C, TSH
Proper Diagnosis is Key

Additional tests:

• Liver, kidney, electrolytes, ESR, thyroid, inflammatory markers, protein study, Vitamins D & B12
• HIV, Anti-Hu antibodies, Anti-Endomysial/Anti-Gliadin antibodies
• Urine for heavy metals
• Aminolevulinic acid/Porphobilinogen
• (24-hour urine)
• Galactosidase assay
• Lipoproteins
Proper Diagnosis is Key

Additional studies

• EMG/Nerve Conduction Study
• Transthyretin Gene test
• Skin biopsy if suspect small fiber neuropathy
• Spinal tap/ Lumbar puncture if suspect auto-immune
• Nerve and muscle biopsy if other tests fail to identify cause
• Sweat test
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