Neuropathic pain phenotyping by international consensus (NeuroPPIC) for genetic studies: a NeuPSIG systematic review, Delphi survey, and expert panel recommendations.

For genetic research to contribute more fully to furthering our knowledge of neuropathic pain, we require an agreed, valid, and feasible approach to phenotyping, to allow collaboration and replication in samples of sufficient size. Results from genetic studies on neuropathic pain have been inconsistent and have met with replication difficulties, in part because of differences in phenotypes used for case ascertainment. Because there is no consensus on the nature of these phenotypes, nor on the methods of collecting them, this study aimed to provide guidelines on collecting and reporting phenotypes in cases and controls for genetic studies. Consensus was achieved through a staged approach: (1) systematic literature review to identify all neuropathic pain phenotypes used in previous genetic studies; (2) Delphi survey to identify the most useful neuropathic pain phenotypes and their validity and feasibility; and (3) meeting of experts to reach consensus on the optimal phenotype(s) to be collected from patients with neuropathic pain for genetic studies. A basic "entry level" set of phenotypes was identified for
any genetic study of neuropathic pain. This set identifies cases of "possible" neuropathic pain, and controls, and includes: (1) a validated symptom-based questionnaire to determine whether any pain is likely to be neuropathic; (2) body chart or checklist to identify whether the area of pain distribution is neuroanatomically logical; and (3) details of pain history (intensity, duration, any formal diagnosis). This NeuroPPIC "entry level" set of phenotypes can be expanded by more extensive and specific measures, as determined by scientific requirements and resource availability.