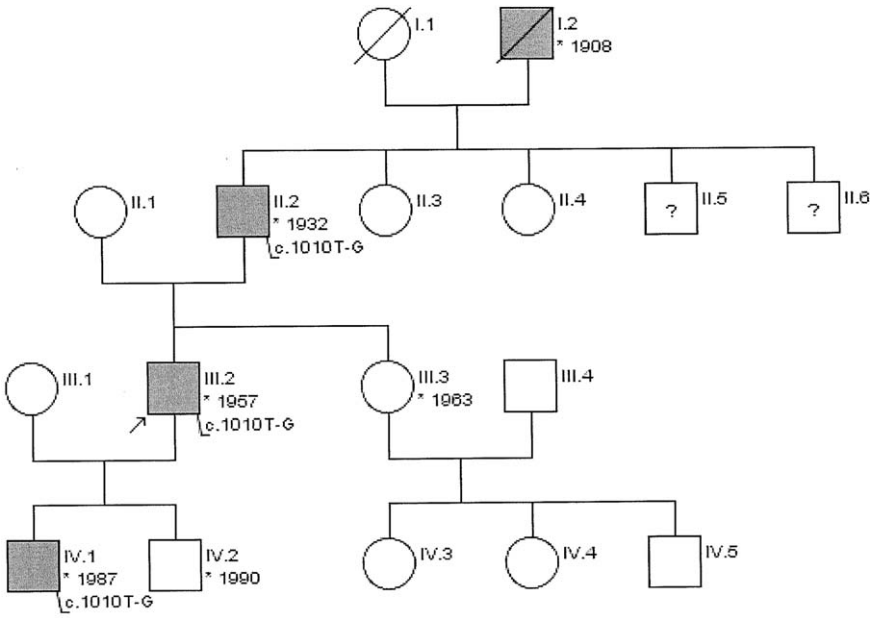

FIGURE 1. Pedigree of the family with a non-dystrophic myotonic syndrome and a new mutation in the *CLCN1* gene. The following individuals had DNA analysis, either by complete *CLCN1* sequencing (III-2), or by targeted sequencing of *CLCN1* exon 9 (II-2, IV-1, and IV-2).White symbols: healthy; white symbols with ?: history of myotonia is unclear; gray symbols: clinically affected; square: man; circle: woman; arrow: proband; cross: deceased; asterisk: year of birth.

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Published online 29 December 2015 in Wiley Online Library (wileyonlinelibrary.com). DOI 10.1002/mus.24971

ANTI- C N1A ANTIBODIES IN SOUTH AUSTRALIAN PATIENTS WITH INCLUSION BODY MYOSITIS

Only recently, with the identification of antibodies to a 43-kDa muscle autoantigen,[1] subsequently identified as cytoplasmic 5′-nucleotidase 1A (cN1A) in sera from patients with inclusion body myositis (IBM),[2,3] has a role for B-cell autoimmunity in IBM been demonstrated. IgG antibodies to cN1A have $>90\%$ specificity and 34%–70% sensitivity in IBM.[1–3] Detection of all 3 isotypes (IgG, IgM, and IgA) by enzyme-linked immunoassay (ELISA) has increased sensitivity to 76%.[4] Hence, these antibodies may be useful biomarkers[5] for IBM.

We determined the prevalence and serological associations of anti-cN1A in South Australian patients with a definitive histological diagnosis of IBM, made by accepted histological criteria.[6–8] All 58 patients fulfilled the histopathological requirements, and 55 of 58 fulfilled the clinicopathological requirements of the European Neuromuscular Centre (ENMC) criteria,[9] which have superior performance in IBM diagnosis compared with other diagnostic criteria.[10] The clinical distribution of muscle involvement is shown in Table 1. Autoantibody production in inflammatory myopathies is at least partially genetically determined,[11] and myositis-specific autoantibodies (MSA) and myositis-associated autoantibodies (MAA) are associated with DR3 and DR4 in South Australian patients with inflammatory myositis.[12] Hence, we sought to further determine whether production of anti-cN1A is also determined by class II alleles.

Antibodies to cN1A were detected by ELISA, as described elsewhere[4] in 24 of 69 (34.8%) patients with IBM. Autoantibodies of the IgM isotype were the most frequent ($n=17$), followed by IgG ($n=13$) and IgA ($n=5$). All 3 isotypes were present in 1 patient; 9 sera showed antibodies of 2 isotypes (IgM and IgA, $n=2$; IgM and IgG, $n=5$; IgA and IgG, $n=2$). There was no gender difference between IBM patients with anti-cN1A (15 of 24 women) compared with those without antibodies (27 of 45 women). Although some MSA have been associated with malignancy,[13] there was no difference in

Table 1. Clinical features of patients with inclusion body myositis (IBM)

	Number present/total
Hip flexor weakness	55/58 (95%)
Knee extensor weakness	55/58 (95%)
Finger flexor weakness	27/58 (47%)
Increased serum creatine kinase	54/61 (89%)
Clinicopathologically defined IBM per ENMC criteria	55/58 (95%)
Histopathological requirements of ENMC criteria met	58/58 (100%)
Definite IBM per Griggs criteria	69/69 (100%)

ENMC, European Neuromuscular Centre.

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prevalence of malignancy in patients with anti-cN1A (3 of 20) compared with those without (10 of 39; $P = 0.51$).

Antibodies to MSA/MAA were present in a minority (8 of 56) of patients with IBM (6 anti-Ro52, 1 anti–PMSCL-100, and 1 anti-PL7) and were significantly less prevalent than anti-cN1A ($P = 0.01$). Only 3 of 24 patients with anti-cN1A (2 IgM and IgG, 1 isolated IgM) had anti-Ro52 antibodies, but there was no concurrent detection of other MSA/MAA.

We were unable to demonstrate that production of anti-cN1A is associated with specific class II HLA alleles. The high background prevalence of DR3 in this cohort due to the strong association of IBM with this allele[14–16] may mask evidence of any such relationship, but notably no relationship with other DR alleles was identified. The low frequency of anti-cN1A in this IBM cohort restricted the sample size in which to study possible genetic associations. The lower prevalence of anti-cN1A in this cohort compared with the 76% sensitivity reported earlier[4] may reflect fluctuating antibody titers or response of antibodies to immunosuppressive therapy (prescribed to 70% of our cohort).

We conclude that anti-cN1A is not associated with gender or malignancy in IBM and appears to not be genetically determined by DR alleles.

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Published online 24 November 2015 in Wiley Online Library (wileyonlinelibrary.com). DOI 10.1002/mus.24989

LOW BACK PAIN WITH RADICULAR SYMPTOMS AS A PRESENTATION OF HEREDITARY NEUROPATHY WITH LIABILITY TO PRESSURE PALSIES: THE DIAGNOSTIC CHALLENGE OF AN ATYPICAL PRESENTATION

The classic presentation of hereditary neuropathy with liability to pressure palsies (HNPP) is described as recurrent common entrapment mononeuropathies with autosomal dominant inheritance of a deletion of the peripheral myelin protein 22 (*PMP22*) gene.[1–3] However, descriptions of phenotypic heterogeneity of HNPP have included Charcot-Marie-Tooth-like[4,5] and chronic inflammatory demyelinating polyneuropathy-like disorders.[6] Recently, other atypical presentations such as cramps[7] and musculoskeletal pain have been described.[8] Family history may be unknown in many cases.[9] Therefore, in an atypical presentation without family history, HNPP may be misdiagnosed. We describe a case of HNPP presenting with low back pain and initially negative family history.

A 26-year-old man presented to our clinic with a 5-year history of low back pain following a back injury. He had radicular symptoms in a roughly left L5/S1 distribution. He also had chronic neck pain and paresthesias in distal fingers. Initially, family history was unknown. Physical exam showed sensory deficits in all fingertips, the left lateral calf and foot without focal weakness. Reflexes were 1+ throughout. His symptoms were suspicious for radiculopathy, but electrodiagnostic findings did not support this diagnosis. Nerve conduction studies demonstrated instead a pattern of multiple common compression mononeuropathies in all limbs. Electromyography was normal. The initial electrodiagnostic impression was that findings suggested hereditary sensorimotor polyneuropathy such as HNPP. MRI of the lumbar spine and work-up for metabolic and autoimmune etiologies was unremarkable. However, given a lack of family history and a presentation not consistent with the traditional recurrent common entrapment neuropathies, HNPP was

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