



**Australian Government**  

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**Repatriation Medical Authority**

**REPATRIATION MEDICAL AUTHORITY**

**STATEMENT OF REASONS**

**S196B(6) *VETERANS' ENTITLEMENTS ACT 1986***

**DECISION NOT TO MAKE A STATEMENT OF PRINCIPLES CONCERNING**

**X-LINKED MYOPATHY WITH EXCESSIVE AUTOPHAGY**

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## **PART I INTRODUCTION**

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1. In accordance with subsection 196B(6) of the *Veterans' Entitlements Act 1986* (the VEA) and following an investigation which was notified in the *Commonwealth of Australia Gazette* on 4 November 2014, the Repatriation Medical Authority (the Authority) has decided not to make Statements of Principles concerning x-linked myopathy with excessive autophagy,
2. The Authority is of the view that there are no factors concerning x-linked myopathy with excessive autophagy which can be related to service rendered by a person as outlined in subsection 196B(14) of the VEA.

## **PART II BACKGROUND TO THE INVESTIGATION**

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3. A request dated 5 August 2014 was received from a person eligible to make a claim under the *Military Rehabilitation and Compensation Act 2004* (the MRCA), seeking an investigation of x-linked myopathy with excessive autophagy to find out whether Statements of Principles may be determined concerning that condition. As no Statements of Principles have been determined for x-linked myopathy with excessive autophagy, the Authority decided to carry out an investigation pursuant to subsection 196B(4) of the VEA to find out whether Statements of Principles may be determined in respect of x-linked myopathy with excessive autophagy.
4. A Notice of Investigation was signed by the Authority Chairperson on 17 October 2014 and was gazetted in accordance with section 196G of the VEA in the *Commonwealth of Australia Gazette* on 4 November 2014. Submissions were invited from persons and organisations eligible to do so. The closing date for submissions as noted in the Gazette Notice was 23 January 2015.
5. The investigation was carried out to find out whether there is information available about how x-linked myopathy with excessive autophagy may be suffered or contracted, or death from x-linked myopathy with excessive autophagy may occur, and the extent to which x-linked myopathy with excessive autophagy or death from x-linked myopathy with excessive autophagy may be war-caused, defence-caused, a service injury, a service disease or a service death.

## **PART III SUBMISSIONS RECEIVED BY THE AUTHORITY PURSUANT TO SECTION 196F**

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6. Following notification of its investigation, the Authority received a further submission from the applicant on 6 November 2014.

#### **PART IV EVIDENCE/INFORMATION AVAILABLE TO THE REPATRIATION MEDICAL AUTHORITY**

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7. In support of the request, the applicant provided:
  - 7.1. a covering letter in which the applicant states that previous claims have not succeeded due to lack of information about x-linked myopathy with excessive autophagy. The applicant is of the view that symptoms of the disease can be aggravated by exercises such as climbing ladders or stairs, squatting and other leg exercises. No specific sound medical-scientific evidence was provided in the request.
  - 7.2. a timeline of the applicant's medical condition in an additional submission on 6 November 2014. This document showed how the applicant developed progressive lower limb muscle weakness over a period of seven years from enlistment in 2005.
8. Literature searches were conducted using the Ovid search engine from 1996 to November Week 2 2014, limited to English language. The search terms were: x-linked myopathy with excessive autophagy.mp. Articles were selected based on relevance, study quality, reliability and journal authority. The above search was supplemented by internet searches and manual searches of reference lists.
9. A briefing paper and associated documents were prepared for presentation to the Authority by a Medical Researcher of the Secretariat in consultation with a member of the Authority.
10. The medical-scientific publications referred to in the briefing paper are set out in the bibliography attached hereto.

#### **PART V SOUND MEDICAL-SCIENTIFIC EVIDENCE**

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11. The Statements of Principles are determined on the basis of the available "sound medical-scientific evidence" as defined in section 5AB(2) of the VEA which states:

*"Information about a particular kind of injury, disease or death is taken to be **sound medical-scientific evidence** if:*

  - (a) the information:*
    - (i) is consistent with material relating to medical science that has been published in a medical or scientific publication and has been, in the opinion of the Repatriation Medical Authority, subjected to a peer review process; or*
    - (ii) in accordance with generally accepted medical practice, would serve as the basis for the diagnosis and management of a medical condition; and*
  - (b) in the case of information about how that kind of injury, disease or death may be caused - meets the applicable criteria for assessing causation currently applied in the field of epidemiology."*

12. Subsection 196B(14) of the VEA states the following about the relationship between a factor concerning a particular condition and service:

*"A factor causing, or contributing to, an injury, disease or death is **related to service** rendered by a person if:*

- (a) it resulted from an occurrence that happened while the person was rendering that service; or*
- (b) it arose out of, or was attributable to, that service; or*
- (c) it resulted from an accident that occurred while the person was travelling, while rendering that service but otherwise than in the course of duty, on a journey:
  - (i) to a place for the purpose of performing duty; or*
  - (ii) away from a place of duty upon having ceased to perform duty; or**
- (d) it was contributed to in a material degree by, or was aggravated by, that service; or*
- (e) in the case of a factor causing, or contributing to, an injury—it resulted from an accident that would not have occurred:
  - (i) but for the rendering of that service by the person; or*
  - (ii) but for changes in the person's environment consequent upon his or her having rendered that service; or**
- (f) in the case of a factor causing, or contributing to, a disease—it would not have occurred:
  - (i) but for the rendering of that service by the person; or*
  - (ii) but for changes in the person's environment consequent upon his or her having rendered that service; or**
- (g) in the case of a factor causing, or contributing to, the death of a person—it was due to an accident that would not have occurred, or to a disease that would not have been contracted:
  - (i) but for the rendering of that service by the person; or*
  - (ii) but for changes in the person's environment consequent upon his or her having rendered that service."**

## **PART VI REASONS FOR THE DECISION**

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13. X-linked myopathy with excessive autophagy is a genetic disorder of lysosomal function, heritable as an X-linked recessive trait, characterized by autophagic vacuolar myopathy, onset during childhood and slowly progressive atrophy and weakness of proximal skeletal muscles without cardiac or central nervous system involvement.
14. Weakness of the proximal muscles of the lower extremities progresses slowly to involve other muscle groups, generally tending toward loss of independent ambulation after age 50. Weakness is may be demonstrated by difficulty climbing stairs and running. The rate of progression is variable; some patients do not lose their ability to walk and some patients are wheelchair bound by about age 60. Increasing exercise intolerance is an expected feature of this progressive disease. There is no evidence to suggest that any particular environmental risk factors cause a more rapid progression of muscle weakness.
15. Having considered and evaluated the sound medical-scientific evidence, the Authority concluded that there is no evidence of any specific factors relating to the

clinical onset or clinical worsening of x-linked myopathy with excessive autophagy and there is as yet, no specific treatment recommended for this condition. This being the case there is no factor at present which could relate this disease to service by a person within the terms of subsection 196B(14) of the VEA.

## **PART VII DECISION**

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16. At its meeting on 11 February 2015 the Authority decided that as there is no sound medical-scientific evidence on which it could rely to determine a Statement of Principles in respect of x-linked myopathy with excessive autophagy either under subsection 196B(2) or (3) of the VEA, it did not propose to make a Statement of Principles for that disease.



**PROFESSOR NICHOLAS SAUNDERS AO**

Chairperson

Repatriation Medical Authority

27 February 2015

## PART VIII BIBLIOGRAPHY

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