

AuSCR Research Task Group approved projects

Title	An investigator-initiated, prospective, feasibility study to describe the occurrence of Fabry disease in people, aged 18 to 55 years, living with stroke in the community in Australia																														
Principle investigator	Professor Craig Anderson																														
Institute	The George Institute for Global Health																														
Co-investigators	Associate Professor Dominique Cadilhac, Professor Vincent Thijs, Ms Brenda Grabsch, Ms Joyce Lim, Dr Alejandra Malavera																														
Submission date	2 February 2017																														
AuSCR role	Participant recruitment																														
Approved	23 May 2017																														
Status	Completed																														
Summary	Stroke in the young (18-55 years) represents about half of the total disability burden of stroke in Australia. Among the broader causes of stroke in the young, genetic disorders such as Fabry disease are of significant importance for investigation. One of the serious clinical manifestations of Fabry disease is stroke. The mechanisms underlying the pathogenesis of stroke in Fabry disease have not been clearly delineated. Currently in Australia, awareness of Fabry disease as a stroke risk factor among clinicians is low, and screening and detection are more ad hoc ('opportunistic') rather than systematic, so there is likely to be a high level of unmet need and the prevalence is uncertain. The primary aim of this study is to determine the feasibility of screening for Fabry disease among people aged 18-55 years, living in the community after stroke and registered in the Australian Stroke Clinical Registry (AuSCR).																														
Outcomes	<p>A total of 78 potential participants replied to the 326 invitations posted, with 59 consented to participate.</p> <table border="1"> <thead> <tr> <th>Participant flow</th> <th>Numbers</th> </tr> </thead> <tbody> <tr> <td>Initial mail response received</td> <td>78</td> </tr> <tr> <td> <ul style="list-style-type: none"> Notified by relative as deceased Declined to participate </td> <td>1 13</td> </tr> <tr> <td>Agreeable to participate</td> <td>64</td> </tr> <tr> <td> <ul style="list-style-type: none"> Signed consent not received </td> <td>13</td> </tr> <tr> <td>Data collection consented</td> <td>51</td> </tr> <tr> <td> <ul style="list-style-type: none"> Discontinue participation for blood collection </td> <td>1</td> </tr> <tr> <td>Blood collection performed and analysed</td> <td>50</td> </tr> </tbody> </table> <table border="1"> <thead> <tr> <th>Fabry disease features</th> <th>Frequency (n=78)</th> </tr> </thead> <tbody> <tr> <td>Acroparesthesias</td> <td>62%</td> </tr> <tr> <td>Angiokeratoma</td> <td>24%</td> </tr> <tr> <td>Joint swelling</td> <td>15%</td> </tr> <tr> <td>Gastrointestinal problems ('spending more time in toilet than others')</td> <td>21%</td> </tr> <tr> <td>Hearing loss</td> <td>18%</td> </tr> <tr> <td>Heat intolerance</td> <td>8%</td> </tr> </tbody> </table> <p>Of the 51% participants who returned their signed consent with the above characteristics, 50 participants agreed to have a blood collection. The great majority (98%) returned a negative result for Fabry disease; only 1 (2%) female participant who was noted to be a pathogenic variant confirmed as a Fabry Disease carrier status.</p>	Participant flow	Numbers	Initial mail response received	78	<ul style="list-style-type: none"> Notified by relative as deceased Declined to participate 	1 13	Agreeable to participate	64	<ul style="list-style-type: none"> Signed consent not received 	13	Data collection consented	51	<ul style="list-style-type: none"> Discontinue participation for blood collection 	1	Blood collection performed and analysed	50	Fabry disease features	Frequency (n=78)	Acroparesthesias	62%	Angiokeratoma	24%	Joint swelling	15%	Gastrointestinal problems ('spending more time in toilet than others')	21%	Hearing loss	18%	Heat intolerance	8%
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Publications	<p>Presented:</p> <ol style="list-style-type: none"> J. Lim, Leveraging infrastructure for screening (Fabry Disease), National Stroke and Data Quality Improvement Workshop, 2 May 2018, Melbourne A. Malavera <i>et al.</i> Feasibility of screening for Fabry Disease in stroke : linked data from the Australia Stroke Clinical Registry (AuSCR).Stroke Society of Australasia Conference, 7-10 Aug 2018, Sydney <p>Report: 1. Australian Fabry Disease Study (AusFabry) Report to SHIRE Australia, 31 Aug 2018.</p>																														