**Supplementary Material 3**

**Literature Search and Preliminary Results**

We used several strategies to identify eligible peer-reviewed literature. First, we identified relevant studies, in English language only, through systematic searches of CENTRAL, CINAHL, EMBASE, MEDLINE and PsycINFO, PapersFirst, ProQuest Dissertations, and Web of Science from the inception of each database up to January 2015. An experienced medical librarian collaborated in the development of the search strategies for each electronic database. With the help of the same librarian, we divided the search results into two sets: peer-reviewed publications and potentially eligible literature. Potentially eligible literature included conference or meeting abstracts, dissertations, or registered trials that were identified through the literature searches. Table 1 details the MEDLINE search strategy, and Table 2 details the number of results from each database.

To determine if complete reports of the potentially eligible literature had been published, we performed a keyword search in Google Scholar, EMBASE, and MEDLINE using the main phrases in the title combined with the authors’ names. If a full report was not identified, we used the authors’ names and the address affiliation information in the citation record to search in Google for the first and last authors’ email addresses, and, if located, we contacted authors to ask for a copy of their full report. In addition, we screened the bibliographies of all eligible studies to identify other relevant studies not captured by the database searches.

**Table 1. Ovid MEDLINE(R) search strategy:**

|  |  |
| --- | --- |
| 1 | "Quality of Life"/ |
| 2 | "outcome assessment (health care)"/ or treatment outcome/ or treatment failure/ |
| 3 | exp pain/ |
| 4 | exp disease attributes/ or exp "signs and symptoms"/ |
| 5 | or/1-4 |
| 6 | health status indicators/ or "severity of illness index"/ or sickness impact profile/ or interviews as topic/ or questionnaires/ or self report/ |
| 7 | Pain Measurement/ |
| 8 | patient satisfaction/ or patient preference/ |
| 9 | or/6-8 |
| 10 | (quality of life or life qualit??? or hrqol or hrql).mp. |
| 11 | (assessment? outcome? or measure? outcome? or outcome? studies or outcome? study or outcome? assessment? or outcome? management or outcome? measure\* or outcome? research or patient? outcome? or research outcome? or studies outcome? or study outcome? or therap\* outcome? or treatment outcome? or treatment failure?).mp. |
| 12 | pain????.mp. |
| 13 | ((activity or sever\* or course) adj3 (disease or disabilit\* or symptom\*)).mp. |
| 14 | or/10-13 |
| 15 | 9 or 14 [database subject headings textwords] |
| 16 | (questionnaire? or instrument? or interview? or inventor\* or test??? or scale? or subscale? or survey? or index?? or indices or form? or score? or measurement?).mp. |
| 17 | (patient? rating? or subject\* report? or subject\* rating? or self report\* or self evaluation? or self appraisal? or self assess\* or self rating? or self rated).mp. |
| 18 | (patient? report\* or patient? observ\* or patient? satisf\*).mp. |
| 19 | anchor base??.mp. |
| 20 | or/16-19 |
| 21 | 15 and 20 [pros] |
| 22 | ("(type 1) gaucher??" or "(type i) gaucher??" or "gaucher ?? disease (gd1)" or "gaucher ?? disease (gdi)" or "gaucher ?? disease (type 1)" or "gaucher ?? disease (type i)" or "gaucher ?? disease type 1" or "gaucher ?? disease type i" or "gaucher?? (type 1)" or "gaucher?? (type i)" or "gaucher?? disease (gd1)" or "gaucher?? disease (gdi)" or "gaucher?? type 1" or "gaucher?? type i" or "non neuronopathic gaucher??" or "noncerebral juvenile gaucher??" or "type 1 (non neuronopathic) gaucher??" or "type 1 (nonneuronopathic) gaucher??" or "type 1 gaucher??" or "type 1 gd" or "type i form GDi)" or "type 1 form GD1)" or "type i (non neuronopathic) gaucher??" or "type i (nonneuronopathic) gaucher??" or "type i gaucher??" or "type i gd").tw. |
| 23 | 21 and 22 [pros gaucher type 1] |
| 24 | ("(type 3) gaucher??" or "(type iii) gaucher??" or "cerebral adult form of gaucher??" or "cerebral juvenile form of gaucher??" or "chronic neuronopathic gaucher??" or "gaucher?? disease (gd3)" or "gaucher?? disease (gdiii)" or "gaucher?? disease (type 3)" or "gaucher?? disease (type iii)" or "gaucher?? disease type 3 " or "gaucher?? disease type iii" or "gaucher?? disease, subacute neuronopathic type" or "gaucher?? type 3" or "gaucher?? type iii" or "neuropathic gaucher?? disease" or "subacute neuronopathic type of gaucher??" or "type 3 chronic neuronopathic) gaucher??" or "type 3 neuronopathic) gaucher??" or "type 3 form GD3)" or "type iii form GDiii)" or "type 3 gaucher??" or "type 3 gd" or "type iii (chronic neuronopathic) gaucher??" or "type iii neuronopathic) gaucher??" or "type iii form GD3)" or "type iii gaucher??" or "type iii gd").tw. |
| 25 | 21 and 24 [pros gaucher type 3] |
| 26 | fabry disease/ |
| 27 | ("alpha galactosidase a deficienc\*" or "angiokeratoma? corporis diffusum" or "angiokeratoma? diffuse" or "angiokeratoma? diffusum" or "ceramide trihexosidase deficienc\*" or "ceramide trihexosidosis" or "diffuse angiokeratoma?" or "fabry anderson disease" or "fabry dyslipidosis" or "fabry?? syndrome" or "fabry?? disease" or "gla deficiency" or "glycosphingolipid lipidosis" or "glycosphingolipidosis" or "glycosphingolipoidosis" or "hereditary dystopic lipidoses" or "hereditary dystopic lipidosis" or "mckusick 30150").tw. |
| 28 | 26 or 27 |
| 29 | 21 and 28 [pros Fabry] |
| 30 | Niemann-Pick Disease, Type B/ |
| 31 | ("b (nmpb)" or "niemann-pick a/b" or "(ndp) type b" or "b form? of niemann pick??" or "b ndp" or "b niemann pick??" or "b subtype? of niemann pick??" or "b type? of niemann pick??" or "form b of niemann pick??" or "ndp a/b" or "niemann pick (a and b)" or "niemann pick disease visceral" or "niemann pick?? (b)" or "niemann pick?? (form? b)" or "niemann pick?? (subtype? b)" or "niemann pick?? (type? b)" or "niemann pick?? a/b" or "niemann pick?? disease (b)" or "niemann pick?? disease (b form?)" or "niemann pick?? disease (b subtype?)" or "niemann pick?? disease (b type?)" or "niemann pick?? disease adult non neuronopathic" or "niemann pick?? disease adult nonneuronopathic" or "niemann pick?? disease nonneuronopathic" or "niemann pick?? disease subtype? b" or "niemann pick?? disease type a/b" or "niemann pick?? disease type b" or "niemann pick?? syndrome, type? b" or "niemann pick?? type a/b" or "niemann pick?? type? b" or "non neuronopathic niemann pick??" or "nonneurologic (type? b) forms of niemann pick??" or "nonneuronopathic niemann pick??" or "np type b" or "npa/b" or "npd type b" or "npd-a/b" or "subtype b of niemann pick??" or "type b of niemann pick??").tw. |
| 32 | 30 or 31 |
| 33 | 21 and 32 [pros Niemann-Pick B] |
| 34 | glycogen storage disease type ii/ |
| 35 | ("acid maltase deficienc\*" or "alpha 14 glucosidase deficienc\*" or "alpha-1,4-glucosidase acid deficienc\*" or "alpha-1,4-glucosidase deficienc\*" or "alpha-glucosidase deficienc\* acid" or "cardiomuscular glycogenosis" or "deficienc\* of alpha-glucosidase" or "diffuse glycogenosis" or "gaa deficienc\*" or "generali?ed glycogenoses" or "generalized glycogenosis" or "glucan 1,4 alpha glucosidase deficiency syndrome" or "glycogen storage disease ii" or "glycogen storage disease type 2" or "glycogen storage disease type ii" or "glycogen storage disease,generalized" or "glycogenoses 2" or "glycogenoses generalized" or "glycogenoses ii" or "glycogenoses type 2" or "glycogenoses type ii" or "glycogenosis 2" or "glycogenosis due to acid maltase deficienc\*" or "glycogenosis generalized" or "glycogenosis ii" or "glycogenosis type 2" or "glycogenosis type ii" or "gsd due to acid maltase deficienc\*" or "gsd ii" or "gsd type 2" or "gsd2" or "lysosomal alpha 14 glucosidase deficiency disease" or "mckusick 23230" or pompe or "pompes disease" or "pompe's disease" or "type 2 glycogenoses" or "type 2 glycogenosis" or "type ii glycogenoses" or "type ii glycogenosis").tw. |
| 36 | 34 or 35 |
| 37 | 21 and 36 [pros Pompe] |
| 38 | Mucopolysaccharidosis II/ |
| 39 | ("Hunter?? disease" or "Hunter?? glossitis" or "Hunter?? hurler ?? " or "Hunter?? syndrome" or "Hurler?? Hunter?? " or "iduronate 2 sulfatase deficienc\*" or "iduronate sulfatase deficienc\*" or "mckusick 30990" or "mps 2" or "mps 2s" or "mps ii" or "mps iis" or "mps type 2" or "mps type 2s" or "mps type ii" or "mps type iis" or "mucopolysaccharidoses 2" or "mucopolysaccharidoses 2s" or "mucopolysaccharidoses ii" or "mucopolysaccharidoses iis" or "mucopolysaccharidoses type 2" or "mucopolysaccharidoses type 2s" or "mucopolysaccharidoses type ii" or "mucopolysaccharidoses type iis" or "mucopolysaccharidosis 2" or "mucopolysaccharidosis 2s" or "mucopolysaccharidosis ii" or "mucopolysaccharidosis iis" or "mucopolysaccharidosis type 2" or "mucopolysaccharidosis type 2s" or "mucopolysaccharidosis type ii" or "mucopolysaccharidosis type iis" or "sulfoiduronate sulfatase deficienc\*" or "type 2 mucopolysaccharidoses" or "type 2 mucopolysaccharidosis" or "type 2s mucopolysaccharidoses" or "type 2s mucopolysaccharidosis" or "type ii mucopolysaccharidoses" or "type ii mucopolysaccharidosis" or "type iis mucopolysaccharidoses" or "type iis mucopolysaccharidosis").tw. |
| 40 | 38 or 39 |
| 41 | 21 and 40 [pros Mucopolysaccharidoses - MPS II Hunter Disease] |
| 42 | Mucopolysaccharidosis I/ |
| 43 | ("mucopolysaccharidosis type 1" or "alpha l iduronidase deficienc\*" or "chondroosteodysplasia" or "chondroosteodystrophy" or "chondroosteoplasia" or "deficienc\* alpha l iduronidase" or "disease hurler" or "disease hurler's" or "dysostosis multiplex" or "gargoylism" or "helmholtz harrington syndrome" or "hurler disease" or "hurler scheie syndrome" or "hurler syndrome" or "hurlers disease" or "hurler's disease" or "hurlers syndrome" or "hurler's syndrome" or "hurler scheie syndrome" or "iduronidase deficiency syndrome" or "lipochondrodystroph\*" or "mckusick 25280" or "mucopolysaccharidosis 1" or "mucopolysaccharidosis 5" or "mucopolysaccharidosis i" or "mucopolysaccharidosis is" or "mucopolysaccharidosis i-sor mucopolysaccharidosis type i" or "mucopolysaccharidosis type ih" or "mucopolysaccharidosis type ih/ss" or "mucopolysaccharidosis type ihs" or "mucopolysaccharidosis type is" or "mucopolysaccharidosis v" or "pfaundler hurler syndrome" or "scheie syndrome" or "syndrome hurler's" or "syndrome hurler scheie" or "syndrome scheie" or "type ih mucopolysaccharidosis" or "type ih/ss mucopolysaccharidosis" or "type ihs mucopolysaccharidosis" or "type is mucopolysaccharidosis").tw. |
| 44 | 42 or 43 |
| 45 | 21 and 44 [pros Mucopolysaccharidoses I Hurler Disease] |
| 46 | 23 or 25 or 29 or 33 or 37 or 41 or 45 [pros 7 rare diseases] |

**Table 2. Results for each database searched**

|  |  |
| --- | --- |
| **Databases Searched (Platform)** | **TOTAL** |
| MEDLINE In-Process & Other Non-Indexed Citations <July 3, 2013> (Ovid) | 47 |
| MEDLINE(R) 1946 to Present with Daily Update <July 3, 2013> (Ovid) | 725 |
| Embase Classic+Embase <1947 to 2013 Week 25> (Ovid) | 1112 |
| PsycINFO <1806 to June Week 4 2013> (Ovid) | 141 |
| Cochrane Central Register of Controlled Trials <May 2013> (Ovid) | 25 |
| CINAHL <July 3, 2013> (EBSCO) | 282 |
| (Web of Science) <July 3, 2013>  Science Citation Index E1xpanded (SCI-EXPANDED) -1900-present  Social Sciences Citation Index (SSCI)-1956-present  Conference Proceedings Citation Index- Science(CPCI-S) 1990-present  Conference Proceedings Citation Index- Social Science & Humanities (CPCI-SSH)-1990-present | 1392 |
| PapersFirst <July 3, 2013> (ProQuest) | 8 |
| Dissertations & Theses <July 3, 2013> (ProQuest) | 12 |
| **TOTAL** | **3744** |