**Appendix Table C90. Outcome assessment: Comparator, inherited metabolic diseases**

| **Study (Investigator, country, year)** | **Record Number** | **Group (N)** | **Primary Outcomes** | **Secondary Outcomes** | **F/U Frequency/Duration** | **Comment** |
| --- | --- | --- | --- | --- | --- | --- |
| Arvio M, Finland, 2001 | 14180 | 12 |   |   |   |   |
| Autti T, Finland, 1999 | 15540 | 6 non-HSCT7 non-diseased |   |   |   |   |
| Banjar H, Saudi Arabia, 1998 | 17920 | 3 | pulmonary involvement | skeletal changes |   |   |
| Chan LL, Malaysia, 2002 | 11330 | 1 | organomegaly growth |   |   |   |
| El-Beshlawy A, Egypt, 2006 | 5750 | 11 | skeletal changes |   |   |   |
| Erikson A, Sweden, 1995 | 21630 | 3 | neuropsychologic scores glucosylceramide levels (lower is better) | growth |   |   |
| Goker-Alpan O, US, 2008 | 1790 | 30 | neuropsychometric assessments |   |   |   |
| Maegawa GHB, Canada, 2009 | 56590B | 2 | neurological assessments neuropsychological tests, 2 types depending on severity of impairment |   | neurological assessments at baseline and every 3 mos neuropsychological tests at baseline and every 6 mos |   |
| Maegawa GHB, Canada, 2009 | 56590A | 3 | neurological assessments neuropsychological tests, 2 types depending on severity of impairment |   | neurological assessments at baseline and every 3 mos neuropsychological tests at baseline and every 6 mos |   |
| Malm G, Sweden, 2004 | 8490 | 2 |   |   |   |   |
| Muenzer J, US, 2006 | 57160 | 96 | 6-minute walk test forced vital capacity |   | baseline, 18 wks, 36 wks, 53 wks |   |
| Muenzer J, US, 2007 | 57070 | 12 | change in urinary glucosaminoglycans | liver and spleen size 6-minute walk test pulmonary function joint mobility heart size and function sleep study | baseline, wk 13, wk 24, wk 25, wk 51 |   |
| Paciorkowski AR, US, 2008 | 2980 | 1 | gait analysis neurologic exams growth parameters |   | gait every 6 mos neurologic exams every 3 mos |   |
| Patterson MC, US, 2007 | 56970 | 12 | change in horizontal saccadic eye movement-alpha (HSEM-alpha) | mini-mental status examination ambulatory index difficulty in swallowing |   | HSEM-alpha is an indicator of disease severity |
| Patterson MC, US, 2010 | 56500 | 10 | horizontal saccadic eye movement (correlates well with disease progression) | neurological assessments swallowing ambulation |   |   |
| Pineda M, Spain, 2009 | 56560 | 66 | 4 disease disability scales: ambulation, manipulation, language, swallowing (the lower the score, the better) |   | at diagnosis, at start of treatment, last clinical contact | Psychiatric impairment was not part of the disability scales because most psychiatric impairment in this disease starts in adolescence or adulthood. |
| Schiffman R, Netherlands, 2008 | 56750 | 30 | change in vertical saccadic eye movement velocity (VSEM) | neurological assessments pulmonary function liver and spleen volume hematological assessments safety evaluations |   | VSEM chosen end point because supranuclear gaze palsy is the only universal neurological symptom of Gaucher Type 3 |
| Schiffmann R, Netherlands, 1997 | 58150 | 5 | neurocognitive scores lumbar puncture (3 of 5 pts) |   | lumbar puncture every 3-6 mos for 3 yrs |   |
| Seto T, Japan, 2001 | 13460A | 7 | MRI findings in MPS pts |   |   |   |
| Seto T, Japan, 2001 | 13460B | 3 | MRI findings in MPS pts |   |   |   |
| Sivakumar P, England, 1999 | 16200 | 1 | neuropsychologic scores neurodevelopmental milestones |   |   |   |
| Takahashi, Japan, 2001 | 14030 | 2 | magnetic resonance spectroscopy measurement of mucopolysaccharides in the central nervous system |   |   |   |