

ASMD Enrolment

Data Point	Response Options
Patient Id	Autogenerated
Date enrolled	Date
Type of entry	Prospective / Retrospective
Consent	Yes / No
Reason (if no consent)	Text
Consent date	Date
Consent Form Type	Adult / Parent and Guardian / Consultee
Consent Form Version	Text
Has consent been given to share anonymised data with approved research projects academic, charity and other not for profit institutions?	Yes / No
Has consent been given to share anonymised data with approved commercial organisations involved in medicinal products for the treatment of Niemann- Pick Diseases?	Yes / No
Has consent been given to link the Clinician Reported Database record with their Patient Reported Dataset record?	Yes / No
Has consent been given to share data with research projects outside Europe?	Yes / No
Date diagnosis was confirmed	Date
Diagnosis age	Numerical
ASMD phenotype	A / B / Intermediate / Unclear / Unknown
Lost to follow up	Yes / No / Unknown
Date last seen in clinic	Date
Deceased	Yes / No / Unknown
Date of death	Date

ASMD Baseline

Data Point	Response Options
Date of assessment	Date
Age at assessment	Numerical

Acid Sphingomyelinase activity

Enzyme test performed	Yes / No / Unknown
Enzyme test method :	14C-SM / 4MU / HNP / MS/MS / Unknown
Leukocytes	Yes / No / Unknown
Leukocytes date	Date
Leukocytes lab	Fundación Niemann Pick / Seccio Errors Congenits- Institut de Bioquímica Clínica / University Hospitals Birmingham / Salford Royal NHS Foundation Trust / Birmingham Children's Hospital / Central Manchester University Hospitals / University College London / Royal Free Hampstead NHS Trust / Cambridge University Hospitals / General University Hospital in Prague / Charles University in Prague / Other / Unknown
Please specify if other	Free text
Leukocytes result	Numerical
Leukocytes result Not Available	Check box
Reference ranges for leukocytes test	Check box
Lower limit - Leukocyte	Numerical
Lower limit - Leukocyte Not Available	Check box
Upper limit - Leukocyte	Numerical
Upper limit - Leukocyte Not Available	Check box
Fibroblasts	Yes / No / Unknown
Fibroblasts date	Date

Fibroblasts Lab	Fundación Niemann Pick / Seccio Errors Congenits- Institut de Bioquímica Clínica / University Hospitals Birmingham / Salford Royal NHS Foundation Trust / Birmingham Children's Hospital / Central Manchester University Hospitals / University College London / Royal Free Hampstead NHS Trust / Cambridge University Hospitals / General University Hospital in Prague / Charles University in Prague / Other / Unknown
Please specify	Free text
Fibroblast result (nmol/h/mg)	Numerical
Fibroblast result (nmol/h/mg) Not Available	Check box
Reference ranges for fibroblast test	Check box
Lower limit - Fibroblast	Numerical
Lower limit - Fibroblast Not Available	Check box
Upper limit - Fibroblast	Numerical
Lower limit - Fibroblast Not Available	Check box
Dry blood spot	Yes / No / Unknown
Dry blood spot date	Date
Dry blood spot lab	Fundación Niemann Pick / Seccio Errors Congenits- Institut de Bioquímica Clínica / University Hospitals Birmingham / Salford Royal NHS Foundation Trust / Birmingham Children's Hospital / Central Manchester University Hospitals / University College London / Royal Free Hampstead NHS Trust / Cambridge University Hospitals / General University Hospital in Prague / Charles University in Prague / Other / Unknown
Please specify	Free text
Dry blood spot result (nmol/h/mg)	Numerical
Dry blood spot result (nmol/h/mg) Not Available	Check box
Reference ranges for Dry blood spot test	Check box
Lower limit - Dry blood spot	Numerical
Lower limit - Dry blood spot Not Available	Check box
Upper limit - Dry blood spot	Numerical
Upper limit - Dry blood spot Not Available	Check box
Leukocytes result	<10% / 10-20% / 20-30% / Unknown
Fibroblasts result	<10% / 10-20% / 20-30% / Unknown
Dry blood spot result	<10% / 10-20% / 20-30% / Unknown
Other enzyme tests	Amniocytes / Chorionic villi / Liver

Genetic Testing

DNA testing performed	Yes / No / Unknown
DNA testing performed date :	Date
Age test performed	Numerical
DNA testing performed lab :	Drop-down list
Please specify	Free text
Variant 1	Yes / No / Unknown
Specify Variant 1	Free text
Allele #1 protein change	Known / Unknown
Specify Allele #1 protein change	Free text
Variant 2	Yes / No / Unknown
Specify Variant 2	Free text
Allele #2 protein change	Known / Unknown
Specify Allele #2 protein change	textline
Further SMPD1 genetic variants	Yes / No / Unknown
Reference Sequence Accession Number	Free text
Have parents been studied	Yes / No / Unknown
Has father been studied ?	Yes / No / Unknown
Has mother been studied ?	Yes / No / Unknown

Patient chitotriosidase activity studied	Yes / No / Unknown
Patient chitotriosidase activity measurement (nmol/h/l)	Numerical
Reference ranges for Chitotriosidase test	Known / Unknown
Lower limit -Chitotriosidase	Numerical
Upper limit -Chitotriosidase	Numerical

Biobanking

Sample stored ?	Yes / No / Unknown
Samples (Can select multiple)	Blood / DBS / DNA / Fibroblasts / Plasma
Name of insitution where sample is stored	Drop-down list
Name of insitution	Free text

Current Health Status

Height (cm)	Numerical
Height (m) Unknown	Check box
Weight	Numerical
Weight unknown	Check box

Physical Examination

Skin	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text
Skin Age of onset	Numerical
Head	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text
Head Age of onset	Numerical
Neck	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text
Neck Age of onset	Numerical
Lymphatic System	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text
Lymphatic System Age of onset	Numerical
Abdomen	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text
Abdomen Age of onset	Numerical
Central Nervous System	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text
Central Nervous System Age of onset	Numerical
Respiratory System	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text
Respiratory System System Age of onset	Numerical
Cardiovascular/Peripheral Vascular System	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text
Cardiovascular/Peripheral Vascular System Age of onset	Numerical
Musculoskeletal	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text
Musculoskeletal Age of onset	Numerical
Other Body System	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text
Other Body System Age of onset	Numerical

Family History

Family history of "Niemann-Pick disease"	Yes / No / Unknown
Any siblings diagnosed with ASMD?	Yes / No / Unknown

Parents consanguinity	Yes / No / Unknown
Parents country of origin known?	Yes / No / Unknown
Father's family country of origin	European / Asian / African / Arab / Latino-American / Unknown
Mother's family country of origin	European / Asian / African / Arab / Latino-American / Unknown
Complication during pregnancy and delivery	Yes / No / Unknown
Specify complication	Free text
Gestational age	Preterm (<37 weeks) / Term (37-42 weeks) / Post-term (>42 weeks) / Unknown
Ancestry	Arab / Jewish / Turkish / Other / Unknown
Ancestry other	Free text
Ancestry Jewish, specify	Askenazi / Sephardic / Unspecified

Pregnancy and Sexual Development History

Did the patient experience delayed puberty?	Yes / No / Unknown
Tanner Stage	Stage 1 / Stage 2 / Stage 3 / Stage 4 / Stage 5
Has patient ever been pregnant?	Yes / No / Patient is male / Unknown
Has the patient fathered any biological children?	Yes / No / Patient is female / Unknown

Visceral Symptoms

Splenomegaly	Yes / No / Unknown
Splenomegaly - age of onset	float
Hepatomegaly	Yes / No / Unknown
Hepatomegaly - age of onset	float
Bleeding tendency	Yes / No / Unknown
Bleeding tendency age	float
Bleeding tendency signs	codedvalue

Liver Disease History

History of liver disease ?	Yes / No / Unknown
Liver Disease	Inflammation / Fibrosis / Cirrhosis / Steatosis / Carcinoma / Other / None
Other Liver Disease	Free text
Severity	Mild / Moderate / Severe / Unknown
Diagnosis Modality	CT / Fibroscan / Elastography / Biopsy / Normal
Start Date	Date
Ongoing	Check box
End Date	Date
Liver failure ?	Yes / No / Unknown
Liver failure - age of onset	float
Persistent jaundice longer than 1 month?	Yes / No / Unknown
Persistent jaundice Start Date	date
Persistent jaundice Ongoing	Yes / No / Unknown
Persistent jaundice Stop Date	date

Renal Impairment History

Renal impairment?	Yes / No / Unknown
Renal impairment type	Reduced Glomerular Filtration / Renal Stones / Other / None
Other Renal Impairment	Free text
Start Date	Date
Ongoing	Check box
End Date	Date

Pulmonary and Cardiovascular Disease

Was Chest X-ray done?	Yes / No / Unknown
Date of Chest X-ray	Date
Chest X-Ray findings	0 = No interstitial lung disease / 1 = Mild (affecting 1% to 25% of the lung volume) / 2 = Moderate (affecting 26% to 50% of the lung volume) / 3 = Severe (affecting 51% to 100% of the lung volume) / Unknown
Was Chest CT done?	Yes / No / Unknown
Date of Chest CT	Date

Chest CT findings	0 = No interstitial lung disease / 1 = Mild (affecting 1% to 25% of the lung volume) / 2 = Moderate (affecting 26% to 50% of the lung volume) / 3 = Severe (affecting 51% to 100% of the lung volume) / Unknown
Ventilatory support	Yes / No / Unknown
Ventilatory support - age started	Numerical
Ventilatory support - age started Unknown	Check box
Non-invasive ventilatory support duration	After exercise / At night / Constant
Non-invasive ventilatory support type	CPAP / BiPAP / Supplementary Oxygen
Cardiovascular disease	Yes / No / Unknown
Cardiovascular Disease	Coronary Artery / Ischemic / Rhythm Abnormality / Heart Failure / Valvular / Other / None
Other	Free text
Start Date	Date
Ongoing	Check box
End Date	Date
Neurological and psychiatric Symptoms	
Development milestones appropriate to age	Yes / No / Unknown
Head control	Normal / Delayed / Lost / Never Achieved / Unknown
Head control problem - age of onset	Numerical
Sitting	Normal / Delayed / Lost / Never Achieved / Unknown
Sitting problem - age of onset	Numerical
Standing-up	Normal / Delayed / Lost / Never Achieved / Unknown
Standing-up problem - age of onset	Numerical
Neurological abnormalities	Yes / No / Unknown
Specify Neurological abnormalities	Ataxia / Dysphagia / Extrapyrimald signs / Gait abnormality / Hyperreflexia / Hypertonia / Hypotonia / Learning Disability / Loss of deep tendon reflexes / Peripheral neuropathy / Seizure / Spasticity / Other
Other Neurological Abnormality	Free text
Start Date	Date
Ongoing	Check box
End Date	Date
Psychiatric manifestations	Yes / No / Unknown
Specify psychiatric manifestations	Depression / Impulsiveness / Psychomotor agitation / Psychosis / Sleeping disorder / Other
Other Psychiatric manifestation	Free text
Start Date	Date
Ongoing	Check box
End Date	Date
Social Interaction	Normal / Poor / Absent / Unknown
Fundoscopic examination	Normal / Abnormal / Unknown / Not Done
Other Symptoms	
Chronic pain	Yes / No / Unknown
Specify chronic pain	Abdomen / Generalised / Headache / Joint / Limb / Other
Please specify Other	Free text
Chronic fatigue	Yes / No / Unknown
Early satiety	Yes / No / Unknown
Diarrhoea	Yes / No / Unknown
Frequent infection (children > 6-8/year ; adult > 2-4/year)	Yes / No / Unknown
Types of infection	Ear / Sinus / Pulmonary / Other
Other	textline
Fractures (low trauma)	Yes / No / Unknown
Feeding difficulties (age < 2)	Yes / No / Unknown
Any other medical illness	Yes / No / Unknown

Specify other medical illness	Cancer / Gammopathies / Gastrointestinal tract / Respiratory / Other
Please specify Other	textline
Delayed growth	Yes / No / Unknown
Gender (auto-populated and hidden to support menarche logic)	Autogenerated
Approximate age at menarche (girls) known	Yes / No / Unknown
Age at menarche	Numerical

Surgical Procedures

Splenectomy	Yes / No / Unknown
Splenectomy surgery age	Numerical
Splenectomy Age Unknown	Check box
Type of Splenectomy	Total / Partial / Unknown
Valvular surgery	Yes / No / Unknown
Valvular surgery Age	Numerical
Valvular surgery Age Unknown	Check box
Hernia surgery	Yes / No / Unknown
Hernia surgery Age	Numerical
Hernia surgery Age Unknown	Check box
Gastric button surgery	Yes / No / Unknown
Gastric button surgery age	Numerical
Gastric button surgery age Unknown	Check box
Other surgery	Yes / No / Unknown
Specify other surgery	Free text
Age at other surgery	Numerical

Transplant History

Transplant	Yes / No / Unknown
Bone Marrow transplant	Yes / No / Unknown
Bone Marrow transplant age	Numerical
Bone Marrow transplant age Unknown	Check box
Liver transplant	Yes / No / Unknown
Liver transplant age	Numerical
Liver transplant age Unknown	Check box

Laboratory Data

Anaemia	Yes / No / Unknown
Anaemia age	Numerical
Anemia Age Unknown	Check box
Thrombocytopenia	Yes / No / Unknown
Thrombocytopenia age	Numerical
Thrombocytopenia age Unknown	Check box
Total cholesterol	Drop-down list
Total cholesterol abnormality age	Numerical
Total cholesterol abnormality age Unknown	Check box
HDL-cholesterol	Drop-down list
HDL-Cholesterol Abnormality Age	Numerical
HDL-Cholesterol Abnormality Age Unknown	Check box
Triglyceride	Drop-down list
Triglyceride abnormality age	Numerical
Triglyceride abnormality age Unknown	Check box
7-Ketocholesterol measured?	Yes / No / Unknown
7-Ketocholesterol age at test	Numerical
7-Ketocholesterol age at test Unknown	Check box
7-Ketocholesterol result (ng/ml)	Numerical
Cholestane-triol measured?	Yes / No / Unknown
Cholestane-triol age at test	Numerical
Cholestane-triol age at test Unknown	Check box
Cholestane-triol result (ng/ml)	Numerical

LysoSM measured?	Yes / No / Unknown
LysoSM age at test	Numerical
LysoSM age at test Unknown	Check box
LysoSM result (ng/ml)	Numerical
LysoSM-509 measured?	Yes / No / Unknown
LysoSM-509 age at test	Numerical
LysoSM-509 age at test Unknown	Check box
LysoSM-509 result (ng/ml)	Numerical

ASMD Encounter

Data Point	Response Options
Visit Details	
Encounter date	Date
Clinic date	Date
Height (cm)	Numerical
Height Percentile (%)	Numerical
Weight (kg)	Numerical
Weight percentile (%)	Numerical
Arterial pressure (mmHg)	Free Text
BMI	Numerical
Significant change since last seen	Yes / No / Unknown
Specify change	Improvement / Stable / Deteriorated
Has the patient had any non-Nieman Pick significant disease or illness	Check Box
Please update the Medical History and Co- Morbidities form to capture the non-Nieman Pick significant disease or illness	Check Box

Physical Examination

Skin	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text
Skin Age of onset	Numerical
Skin Age of onset - Unknown	Check Box
Head	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text
Head Age of onset	Numerical
Head Age of onset - Unknown	Check Box
Neck	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text
Neck Age of onset	Numerical
Neck Age of onset - Unknown	Check Box
Lymphatic System	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text
Lymphatic System Age of onset	Numerical
Lymphatic System Age of onset - Unknown	Check Box
Abdomen	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text
Abdomen Age of onset	Numerical
Abdomen Age of onset - Unknown	Check Box
Central Nervous System	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text
Central Nervous System Age of onset	Numerical
Central Nervous System Age of onset - Unknown	Check Box
Respiratory System	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text

Respiratory System System Age of onset	Numerical
Respiratory System System Age of onset - Unknown	Check Box
Cardiovascular/Peripheral Vascular System	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text
Cardiovascular/Peripheral Vascular System Age of onset	Numerical
Cardiovascular/Peripheral Vascular System Age of onset - Unknown	Check Box
Musculoskeletal	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text
Musculoskeletal Age of onset	Numerical
Musculoskeletal Age of onset - Unknown	Check Box
Other Body System	Normal / Abnormal - Not Clinically Significant / Abnormal - Clinically Significant / Not Done
Describe clinically significant finding	Free text
Other Body System Age of onset	Numerical
Other Body System Age of onset - Unknown	Check Box

Visceral Features

Splenomegaly	Yes / No / Unknown
Spleen measurement	Normal / Abnormal / Unknown / Not Done
Spleen clinical examination	Normal / Enlarged / Reduced / Unknown
Spleen MRI	Yes / No / Unknown
Spleen MRI volume (cm3) (most recent data)	Numerical
Spleen CT	Yes / No / Unknown
Spleen CT volume (cm3) (most recent data)	Numerical
Spleen echography	Yes / No / Unknown
Spleen size from echography (cm)	Numerical
Hepatomegaly	Yes / No / Unknown
Liver measurement	Normal / Abnormal / Unknown / Not Done
Liver clinical examination	Normal / Enlarged / Reduced / Unknown
Liver MRI	Yes / No / Unknown
Liver MRI actual measurement (cm)	Numerical
Liver CT	Yes / No / Unknown
Liver CT volume (cm*3) (most recent data)	Numerical
Liver Echography	Yes / No / Unknown
Liver size from echography (cm)	Numerical
Liver Disease	Yes / No / Unknown
Liver failure or portal hypertension	Yes / No / Unknown
Liver Disease	Inflammation / Fibrosis / Cirrhosis / Steatosis / Carcinoma / Other / None
Other Liver Disease	Free text
Severity	Mild / Moderate / Severe / Unknown
Diagnosis Modality	CT / Fibroscan / Elastography / Biopsy / Normal
Start Date	Date
Ongoing	Check box
End Date	Date
Bleeding tendency	Yes / No / Unknown
Bleeding tendency signs	Easy Bruising / Epistaxis / Excessive Bleeding
Renal impairment?	Yes / No / Unknown
Renal impairment type	Reduced Glomerular Filtration / Renal Stones / Other / None
Other Renal Impairment	Free text
Start Date	Date
Ongoing	Check box
End Date	Date

Pulmonary and Cardiovascular Examination

Increased shortness of breath	Yes / No / Unknown
-------------------------------	--------------------

Shortness of breath scale	Resting / After Exertion
Dyspnea on exertion	Yes / No / Unknown
Ventilatory support	Yes / No / Unknown
Non-invasive ventilatory support type	CPAP / BiPAP / Supplemental Oxygen
Non-invasive ventilatory support duration	After Exercise / At Night / Constant
Was Chest X-ray done?	Yes / No / Unknown
Date of Chest X-ray	Date
Chest X-Ray findings	0 = No interstitial lung disease / 1 = Mild (affecting 1% to 25% of the lung volume) / 2 = Moderate (affecting 26% to 50% of the lung volume) / 3 = Severe (affecting 51% to 100% of the lung volume) / Unknown
Ground glass opacity	Mild / Moderate / Severe
Was Chest CT done?	Yes / No / Unknown
Date of Chest CT	Date
Chest CT findings	0 = No interstitial lung disease / 1 = Mild (affecting 1% to 25% of the lung volume) / 2 = Moderate (affecting 26% to 50% of the lung volume) / 3 = Severe (affecting 51% to 100% of the lung volume) / Unknown
Ground glass opacity	Mild / Moderate / Severe
Lung Function tests	Yes / No / Unknown
FEV1(litre)	Numerical
FEV1 predicted (%)	Numerical
FVC (litre)	Numerical
FVC predicted (%)	Numerical
FEV1/FVC (%)	Numerical
DLCO (mmol/min/kPa)	Numerical
Cardiovascular disease	Yes / No / Unknown
Specify Cardiovascular Disease	Coronary Artery / Ischemic / Rhythm Abnormality / Heart Failure / Valvular / Other / None
Please specify Other	Free Text
ECG result	Normal / Abnormal / Unknown / Not Done
Specify ECG result	Free Text
Echocardio results of last test done	Normal / Abnormal / Unknown / Not Done
Specify Echocardio result	Free Text
Heart CT status	Normal / Abnormal / Unknown / Not Done
Heart CT	Free Text
Heart MRI status	Normal / Abnormal / Unknown / Not Done
Heart MRI	Free Text

Transplant Procedures

Transplant	Yes / No / Unknown
Bone Marrow transplant	Yes / No / Unknown
Bone Marrow transplant age	Numerical
Bone Marrow transplant age Unknown	Check Box
Liver transplant	Yes / No / Unknown
Liver transplant age	Numerical
Liver transplant age Unknown	Check Box

Laboratory Assessments

Hb (g/L)	Numerical
Platelets (10*9/L)	Numerical
Leukocytes (10*9/L)	Numerical
INR	Numerical
PT	Numerical
PTT (partial thromboplastin time)	Numerical
AST (u/l)	Numerical
ALAT (u/l)	Numerical
gamma-GT (u/l)	Numerical
Protein (g/l)	Numerical

Albumin (g/l)	Numerical
Total cholesterol (mmol/l)	Numerical
HDL-cholesterol (mmol/l)	Numerical
Triglycerides (mmol/l)	Numerical
Creatinine (μmol/L)	Numerical
Blood Urea Nitrogen (mmol/l)	Numerical
NT pro-BNP (ng/L)	Numerical
Lysosphingomyeline (ng/ml)	Numerical
CCL18	Normal / Abnormal / Unknown / Not Done
Patient chitotriosidase activity studied	Yes / No / Unknown
Patient chitotriosidase activity measurement (nmol/ml/hr)	Numerical
7-Ketocholesterol measured?	Yes / No / Unknown
7-Ketocholesterol result (ng/ml)	Numerical
Cholestane-triol measured?	Yes / No / Unknown
Cholestane-triol result (ng/ml)	Numerical
LysoSM measured?	Yes / No / Unknown
LysoSM result (ng/ml)	Numerical
LysoSM-509 measured?	Yes / No / Unknown
LysoSM-509 result (ng/ml)	Numerical
Pulmonary HRCT status	Normal / Abnormal / Unknown / Not Done
Pulmonary HRCT	Free text
Pulmonary HRCT unit	Free text
Heart CT status	Normal / Abnormal / Unknown / Not Done
Heart CT	Free text
Heart MRI status	Normal / Abnormal / Unknown / Not Done
Heart MRI	Free text
DEXA scan	Yes / No / Unknown
Z-score (including unit)	Numerical
T-score (including unit)	Numerical

ASMD Medical History and Comorbidites

Data Point	Response Options
Date Reported	Date
Does the patient have any non-Niemann Pick disease related medical history and/or co-morbidities?	Check Box
Description	Free Text
Description ICD-10 Code	ICD-10 List
Start Date	Date
Ongoing?	Check Box
End date	Date
Do you suspect a relationship to any of the medications the patient is currently taking?	Check Box
Do you suspect a relationship to Nieman-Pick Specific Medication?	Not Related / Unlikely Related / Possibly Related / Definitely Related / Not Applicable
Please complete an Adverse Drug Reaction form for this Nieman-Pick medication related medical history/comorbidity event	Check Box

ASMD Medications

Data Point	Response Options
Date Reported	Date
Is the patient receiving any medication?	Check Box
Medication name	Free Text
Indication description	Free Text
ICD-10 Code selection options	Niemann-Pick Disease ICD-10 codes / Other Disease Indication ICD-10 codes
Niemann Pick Disease ICD-10 codes	E75.240 type A / E75.241 type B
Other Disease Indication ICD-10 Code	ICD-10 List
Start Date	Date

Ongoing?	Check Box
End date	Date
Dose	Numerical
Unit	Drop-Down List
Please specify	Free Text
Route	Drop-Down List
Please specify	Free Text
Frequency	Drop-Down List
Please specify	Free Text
Was treatment discontinued?	Check Box
Reason for treatment discontinuation	Adverse Drug Reaction / Lack of Efficacy / Patient Choice / Unaffordability / Clinician Decision / Change in Treatment Goals / Other
Adverse Drug Reaction ID linked to treatment discontinuation	Free Text
Please specify	Free Text

Olipudase Alfa only

Date Reported	Date
Is patient receiving olipudase alfa?	Check Box
Start Date	Date
Ongoing	Check Box
End Date	Date
Dose	Numerical
Unit	Drop-Down List
Other unit	Free Text
Administration Setting	Clinic / At Home
Primary therapy	Change / Ongoing / Discontinued
Describe change	Change in Therapy / Missed Dose / Reduced Dose / Increased Dose
Reason for change	Safety / Efficacy / Other
Other reason	Free Text
Number of missed doses	Numerical
Re-escalation?	Check Box

ASMD Adverse Drug Reactions

Data Point	Response Options
Date reported	Date
Has an Adverse Drug Reaction occurred?	Check Box
Adverse Drug Reaction Description	Free Text
Start Date	Date
Ongoing?	Check Box
End Date	Date
Severity	Mild / Moderate / Severe
Do you suspect a relationship to any of the medications the patient is currently taking?	Yes / No
Do you suspect a relationship to Nieman Pick Specific Medication?	Not Related / Unlikely Related / Possibly Related / Probably Related / Defintely Related / Not Applicable
Seriousness	Not Serious / Death / Life-Threatening / Requires Inpatient Hospitalisation or Prolongation of Existing Hospitalisation / Resulted in Persistent or Significant Incapacity or Disability / Congenital Abnormality or Birth Defect / Other Medically Important Event
Specify other medically important event	Free Text
Initial hospitalisation or Prolongation of current hospitalisation episode?	Initial / Prolongation
Admission date	Date
Discharge date	Date
Outcome	Recovered/Resolved / Recovering/Resolving / Not Recovered/Not Resolved / Recovered/Resolved with Sequelae / Fatal / Unknown
Has any medication been discontinued as a result of ADR ?	Yes / No / Unknown

Did the ADR abate after dose interruption/discontinuation?	Yes / No / Unknown
--	--------------------

ASMD Pregnancy

Data Point	Response Options
Encounter Date	Date
Has Pregnancy occurred?	Yes / No / Unknown
Date Reported	Date
Date of Last Menstrual Period	Date
Outcome of Pregnancy	Live Birth / Fetal Death / Termination / Spontaneous Abortion
Health Status of live birth	Free Text

ASMD End of Data Collection

Data Point	Response Options
End of participation date	Date
Reason for withdrawl of consent	Deceased / Lost to Follow Up / Other
Please specify	Free Text
Date of Death	Date
Lost to follow up date	Date
Status at end of study	Alive / Deceased / Unknown
Cause of Death	Free Text