

NPC Enrolment

Label	Response	Mandatory
Consent	Yes/No/Not required	Mandatory
Reason	Free text	Based on previous response
Consent date	dd/mmm/yyyy	Based on previous response
Patient Id	Autogenerated	Mandatory
Date enrolled	dd/mmm/yyyy	Mandatory
Date Data Collection Starts	dd/mmm/yyyy	Expected
Type of entry	Prospective/Retrospective	Mandatory
Lost to follow up	Yes/No/Unknown	Mandatory
Lost to follow up date	dd/mmm/yyyy	Based on previous response
NPC subtype	NPC1/NPC2/Unknown	Mandatory
Diagnosis age	Autogenerated or Year,Month,Day	Mandatory
Diagnosis date	dd/mmm/yyyy	Mandatory
Clinical Form	Neonatal rapidly fatal (0 years)/Early infantile neurological (<2 years)/Late infantile neurological (2-6 years)/Juvenile neurological (6-15 years)/Adult neurological (>15years)/Unknown neurological/Visceral symptoms only	Mandatory
Deceased	Yes/No/Unknown	Mandatory
Date of death	dd/mmm/yyyy	Based on previous response

NPC Baseline

Label	Response	Mandatory
Biochemical and Genetic testing		
DNA testing performed	Yes/No/Unknown	Mandatory
NP-C1 mutation identified?	Yes/No/Unknown	Based on previous response
Mutation #1 NP-C1	Free text	Based on previous response
Mutation #2 NP-C1	Free text	Based on previous response
Further NPC1 genetic variants identified ?	Yes/No/Unknown	Based on previous response
NP-C2 mutation identified?	Yes/No/Unknown	Based on previous response
Mutation #1 NP-C2	Free text	Based on previous response
Mutation #2 NP-C2	Free text	Based on previous response
Further NPC2 genetic variants identified ?	Yes/No/Unknown	Based on previous response
Age test performed	Year/Month/Day	Based on previous response
DNA testing performed lab :	Lab list	Based on previous response
Please specify	Free text	prohibited
Have parents been studied	Yes/No/Unknown	Based on previous response
Father studied	Yes/No/Unknown	Based on previous response
Mother studied	Yes/No/Unknown	prohibited
Filipin-test performed ?	Yes/No/Unknown	Mandatory
Results of Filipin-test	Classic/Variant/Uncertain/Normal	Based on previous response
Date test performed	dd/mmm/yyyy or Unknown	Based on previous response
Name of laboratory the test performed	Lab list	Based on previous response
Please specify	Free text	Mandatory
7-Ketocholesterol tested	Yes/No/Unknown	Based on previous response
7-Ketocholesterol result	Normal/Borderline/Elevated/Unknown	Based on previous response
7-Ketocholesterol date	dd/mmm/yyyy or Unknown	Based on previous response
7-Ketocholesterol lab	Lab list	Mandatory
Please specify	Free text	Based on previous response
Cholestane-triol tested ?	Yes/No/Unknown	Based on previous response
Cholestane-triol results	Normal/Borderline/Elevated/Unknown	Based on previous response
Cholestane-triol date	dd/mmm/yyyy or Unknown	Based on previous response
Cholestane-triol lab	Lab list	Based on previous response

Please specify	Free text	Based on previous response
Sample stored ?	Yes/No/Unknown	Mandatory
Samples (Can select multiple)	Blood/DBS/DNA/Fibroblasts/Plasma	Based on previous response
Family History		
Family history of "Niemann-Pick disease"	Yes/No/Unknown	Mandatory
Parents consanguinity	Yes/No/Unknown	Mandatory
Father's family country of origin	European/Asian/African/Arab/Latino-America/Unknown	expected
Mother's family country of origin	Yes/No/Unknown	expected
Complication during pregnancy and delivery	Yes/No/Unknown	expected
Specify complication	Free text	expected
Gestational age	Preterm:<37 weeks/Term:37-42 weeks/Post-term:>42 weeks/Unknown	Based on previous response
Clinical History		
Visceral		
Suffered from neonatal (< 28d) Jaundice	Yes/No/Unknown	Mandatory
Neonatal Jaundice period	<=14 days/>14 days/Unknown	Based on previous response
Ever have Hepatomegaly/Splenomegaly in neonatal period	Yes/No/Unknown	Mandatory
Showed signs of hepatic failure in neonatal period	Yes/No/Unknown	Mandatory
Splenomegaly	Yes/No/Unknown	Mandatory
Splenomegaly (age (in years) first observed)	Year/Month/Day	Based on previous response
Hepatomegaly	Yes/No/Unknown	Mandatory
Hepatomegaly (age (in years) first observed)	Year/Month/Day	Based on previous response
Pulmonary dysfunction	Yes/No/Unknown	Based on previous response
Pulmonary dysfunction age (in years) at first observed	Year/Month/Day	prohibited
Neurological		
Development delay (Child)	Yes/No/Unknown	Mandatory
Development delay age (in years) first observed (Child)	Year/Month/Day	Based on previous response
Development delay (Motor)	Yes/No/Unknown	Based on previous response
Development delay (Motor) (Sitting)	Yes/No/Unknown	Based on previous response
Development delay (Motor) (Sitting) age (years) first observed	Number	Based on previous response
Development delay (Motor) (Standing)	Yes/No/Unknown	Based on previous response
Development delay (Motor) (Standing) age (years) first observed	Number	Based on previous response
Development delay (Motor) (Walking)	Yes/No/Unknown	Based on previous response
Development delay (Motor) (Walking) age (years) first observed	Number	Based on previous response
Development delay (Speech)	Yes/No/Unknown	Based on previous response
Development delay severity (Speech)	Slight/Moderate/Severe/Regression	Based on previous response
Development delay (Cognitive)	Yes/No/Unknown	Based on previous response

Development delay severity (Cognitive)	Slight/Moderate/Severe/Regression	Based on previous response
Development delay (Social)	Yes/No/Unknown	Based on previous response
Development delay severity (Social)	Slight/Moderate/Severe/Regression	Based on previous response
Clumsiness/impaired coordination	Yes/No/Unknown	Mandatory
Clumsiness/impaired coordination age (in years) first observed	Year/Month/Day	Based on previous response
Ataxia	Yes/No/Unknown	Mandatory
Ataxia age (yrs) first observed	Year/Month/Day	Based on previous response
Ataxia severity	Mild/Moderate/Severe	Based on previous response
Cataplexy	Yes/No/Unknown	Mandatory
Cataplexy age (years) first observed	Year/Month/Day	Based on previous response
Seizure	Yes/No/Unknown	Mandatory
Seizure age (years) first observed	Year/Month/Day	Based on previous response
Dysarthria	Yes/No/Unknown	Mandatory
Dysarthria age (years) first observed	Year/Month/Day	Based on previous response
Dysphagia	Yes/No/Unknown	Mandatory
Dysphagia age (years) first observed	Year/Month/Day	Based on previous response
Dystonia	Yes/No/Unknown	Mandatory
Dystonia age (years) first observed	Year/Month/Day	Based on previous response
Abnormal muscle tone	Yes/No/Unknown	Mandatory
Abnormal muscle tone age (years) first observed	Year/Month/Day	Based on previous response
Specify abnormal muscle tone	Hypertonia/Hypotonia	Based on previous response
Abnormal Vertical Saccades	Yes/No/Unknown	Mandatory
Abnormal Vertical Saccades age (years) first observed	Year/Month/Day	Based on previous response
Specify abnormal vertical saccades	Slow/Absent	Based on previous response
Vertical gaze palsy	Yes/No/Unknown	Mandatory
Vertical gaze palsy age	Year/Month/Day	Based on previous response
Hearing impairment	Yes/No/Unknown	Mandatory
Hearing Impairment age (years) first observed	Year/Month/Day	Based on previous response
Cognitive and psychiatric		
Cognitive impairment	Yes/No/Unknown	Mandatory
Cognitive impairment age (yrs) first observed	Year/Month/Day	Based on previous response
Cognitive impairment type	Attention/Executive/Language/Memory	Based on previous response
Psychiatric manifestations	Yes/No/Unknown	Mandatory
Psychiatric manifestations age (yrs) first observed	Year/Month/Day	Based on previous response
Psychiatric manifestations types	Depression/Impulsiveness (blow up of temper, tendency towards aggressive behaviour)/Psychomotor agitation/Psychosis (including hallucinations, delusions)/Sleeping disorder/Other	Based on previous response
Please specify	Free text	prohibited
Other cognitive and psychiatric symptoms ?	Yes/No/Unknown	Based on previous response

Specify other cognitive and pyschiatric symptoms	Free text	Based on previous response
Other clinical signs and symptoms		
Any other medical illness ?	Yes/No/Unknown	Mandatory
Specify other medical illness	Free text	Based on previous response
Treatments		
Symptomatic therapies	Antacids/Antidepressant/Antiepileptic /Antipsychotics/Antireflux/Antispastic /Dopaminergic/Mood stabilizer/Other/Unknown	expected
Please specify	Free text	Based on previous response
Name	Miglustat/Other	Mandatory
Please specify	Free text	Mandatory
Start date	dd/mm/yyyy	Based on previous response
Ongoing	Yes or No	Mandatory
End date	dd/mm/yyyy	Mandatory
Dose (mg)	50/75/100/200/500/Other	Based on previous response
Please specify	Free text	Mandatory
Frequency	Once a day/ Twice a day/ Three times a day	Based on previous response
Has an adverse event occurred to patient	Yes or No	Based on previous response
Adverse event	Diarrhoea/Low platelets/Tremour/Weight loss/Other	Based on previous response
Please specify	Free text	Expected
Clinical Trial Medication - Name	Free text	Mandatory
Start date	dd/mm/yyyy	Mandatory
Ongoing	Yes or No	Based on previous response
End date	dd/mm/yyyy	Mandatory
Dose	Free text	Mandatory
Frequency	Free text	Based on previous response
Has an adverse event occurred to patient	Yes or No	Mandatory
Adverse event	Free text	Expected
Disability Scale		
Ambulation	Normal (0)/Clumsiness (1)/Autonomus ataxic gait (2)/Outdoor assisted ambulation (3)/Indoor assisted ambuation (4)/Wheel-chair bound (5)	Mandatory
Manipulation	Normal (0)/ Tremor (1)/Slight dysmetria/dystonia (allows for autonomous manipulation) (2)/Mild dysmetria/dystonia (requires help for several tasks but is able to feed self) (3)/ Severe dysmetria/dystonia (requires assistance in all activities) (4)	Mandatory
Speech	Normal (0)/ Delayed acquisition (1)/Mild dysarthria (understandable) (2)/ Severe dysarthria (only comprehendable to some family members (3)/ Non-verbal communication (4)/ Absence of communications (5)	Mandatory
Swallowing	Normal (0)/ Abnormal chewing (1)/ Occasional dysphagia (2)/ Daily dysphagia (3)/ Nasogastric/gastric button feeding (4)	Mandatory

Eye movements	Normal (0)/ Slow ocular pursuit (1)/ Vertical ophthalmoplegia (2)/ Complete ophthalmoplegia (3)	Mandatory
Seizure	No (0)/ Occasional seizures (1)/ Seizures with AED (2)/ Seizures resistant to AED (3)	Mandatory
Total	autocalculated	Expected
Additional Investigation		
Neuroimaging assessment done	Yes/No/Unknown	Mandatory
MRI result	Normal/Abnormal/Unknown/Not done	Based on previous response
Date of assessment (MRI)	dd/mm/yyyy	Based on previous response
MRI assessment finding	Cerebellar atrophy/Cortical atrophy/Ventricular dilation/White matter changes/Other	Based on previous response
Please specify	Free text	Based on previous response
CT result	Yes/No/Unknown	Mandatory
Date of assessment (CT)	dd/mm/yyyy	Based on previous response
CT assessment finding	Cerebellar atrophy/Cortical atrophy/Ventricular dilation/White matter changes/Other	Based on previous response
Please specify	Free text	Based on previous response
Oxysterols	Yes/No/Unknown	prohibited
Oxysterols details	Cholestane-3b,5a,6b-triol/7- ketcholesterol	prohibited

NPC Encounter

Label	Response	Mandatory
Visit date	dd/mm/yyyy or Unknown	expected
Height (cm)	Number	expected
Height Percentile (%)	Number	expected
Weight (kg)	Number	expected
Weight percentile (%)	Number	expected
BMI	autocalculated	Based on previous response
Significant change since last seen	Yes/No/Unknown	prohibited
Specify change	Improvement/Stable/Deteriorated	prohibited
Visceral		
Splenomegaly	Yes/No/Unknown	Mandatory
Hepatomegaly	Yes/No/Unknown	Mandatory
Pulmonary dysfunction	Yes/No/Unknown	Mandatory
Jaundice	Yes/No/Unknown	Mandatory
Neurological		
Development delay (Child)	Yes/No/Unknown	Mandatory
Development delay (Motor)	Yes/No/Unknown	Based on previous response
Development delay (Motor) (Sitting)	Yes/No/Unknown	Based on previous response
Development delay (Motor) (Standing)	Yes/No/Unknown	Based on previous response
Development delay (Motor) (Walking)	Yes/No/Unknown	Based on previous response
Development delay (Speech)	Yes/No/Unknown	Based on previous response
Development delay severity (Speech)	Slight/Moderate/Severe/Regression	Based on previous response
Development delay (Cognitive)	Yes/No/Unknown	Based on previous response
Development delay severity (Cognitive)	Slight/Moderate/Severe/Regression	Based on previous response
Development delay (Social)	Yes/No/Unknown	Based on previous response
Development delay severity (Social)	Slight/Moderate/Severe/Regression	Based on previous response

Clumsiness/impaired coordination	Yes/No/Unknown	Mandatory
Ataxia	Yes/No/Unknown	Mandatory
Ataxia severity	Mild/Moderate/Severe	Based on previous response
Cataplexy	Yes/No/Unknown	Mandatory
Seizure	Yes/No/Unknown	Mandatory
Dysarthria	Yes/No/Unknown	Mandatory
Dysphagia	Yes/No/Unknown	Mandatory
Dystonia	Yes/No/Unknown	Mandatory
Abnormal muscle tone	Yes/No/Unknown	Mandatory
Specify abnormal muscle tone	Hypertonia/Hypotonia	Based on previous response
Vertical gaze palsy	Yes/No/Unknown	Mandatory
Hearing impairment	Yes/No/Unknown	Mandatory
Abnormal Saccades	Yes/No/Unknown	Based on previous response
Specify abnormal saccades	Horizontal/Vertical/Complete	Expected
Cognitive and Psychiatric		
Cognitive impairment	Yes/No/Unknown	Mandatory
Cognitive impairment type	Attention/Executive/Language/Memory	Based on previous response
Psychiatric manifestations	Yes/No/Unknown	Mandatory
Psychiatric manifestations types	Depression/Impulsiveness (blow up of temper, tendency towards aggressive behaviour)/Psychomotor agitation/Psychosis (including hallucinations, delusions)/Sleeping disorder/Other	Based on previous response
Please specify	Free text	Mandatory
Other developmental delay	Yes/No/Unknown	Based on previous response
Specify other developmental delay	Free text	prohibited
Disability Scale		
Ambulation	Normal (0)/Clumsiness (1)/Autonomous ataxic gait (2)/Outdoor assisted ambulation (3)/Indoor assisted ambulation (4)/Wheel-chair bound (5)	Mandatory
Manipulation	Normal (0)/ Tremor (1)/Slight dysmetria/dystonia (allows for autonomous manipulation) (2)/Mild dysmetria/dystonia (requires help for several tasks but is able to feed self) (3)/ Severe dysmetria/dystonia (requires assistance in all activities) (4)	Mandatory
Speech	Normal (0)/ Delayed acquisition (1)/Mild dysarthria (understandable) (2)/ Severe dysarthria (only comprehensible to some family members) (3)/ Non-verbal communication (4)/ Absence of communications (5)	Mandatory
Swallowing	Normal (0)/ Abnormal chewing (1)/ Occasional dysphagia (2)/ Daily dysphagia (3)/ Nasogastric/gastric button feeding (4)	Mandatory
Eye movements	Normal (0)/ Slow ocular pursuit (1)/ Vertical ophthalmoplegia (2)/ Complete ophthalmoplegia (3)	Mandatory
Seizure	No (0)/ Occasional seizures (1)/ Seizures with AED (2)/ Seizures resistant to AED (3)	Mandatory

Total	autocalculated	Expected
Other clinical signs		
Any other medical illness ?	Yes/No/Unknown	Mandatory
Specify other medical illness	Free text	Based on previous response
Treatments		
Symptomatic therapies	Antacids/Antidepressant/Antiepileptic /Antipsychotics/Antireflux/Antispastic /Dopaminergic/Mood stabilizer/Other/Unknown	expected
Please specify	Free text	Based on previous response
Clinical Trial Medication - Name	Free text	Mandatory
Start date	dd/mmm/yyyy	Mandatory
Ongoing	Yes or No	Based on previous response
End date	dd/mmm/yyyy	Mandatory
Dose	Free text	Mandatory
Frequency	Free text	expected
Has an adverse event occurred to patient	Yes or No	Based on previous response
Adverse event	Free text	Expected
Disease Modifying Therapies - Name	Miglustat/Other	Mandatory
Please specify	Free text	Mandatory
Start date	dd/mmm/yyyy	Based on previous response
Ongoing	Yes or No	Mandatory
End date	dd/mmm/yyyy	Mandatory
Dose	50/75/100/200/500/Other	expected
Please specify	Free text	Based on previous response
Frequency	Once a day/ Twice a day/ Three times a day	Based on previous response
Has an adverse event occurred to patient	Yes or No	Based on previous response
Adverse event	Diarrhoea/Low platelets/Tremour/Weight loss/Other	Based on previous response
Please specify	Free text	Expected
Additional Investigations		
Neuroimaging assessment done	Yes/No	Mandatory
Has MRI	Yes/No/Unknown	Based on previous response
MRI result	Yes/No/Unknown	Based on previous response
Date of assessment (MRI)	dd/mmm/yyyy	Based on previous response
MRI assessment finding	Cerebellar atrophy/Cortical atrophy/Ventricular dilation/White mater changes/Other	Based on previous response
Please specify	Free text	Based on previous response
Has CT	Yes/No/Unknown	Based on previous response
CT result	Yes/No/Unknown	Mandatory
Date of assessment (CT)	dd/mmm/yyyy	Based on previous response
CT assessment finding	Cerebellar atrophy/Cortical atrophy/Ventricular dilation/White mater changes/Other	Based on previous response
Please specify	Free text	Based on previous response
Oxysterols	Yes/No/Unknown	Based on previous response
Oxysterols details	Cholestane-3b,5a,6b-triol/7-ketocholesterol	Based on previous response

NPC Medical History and Comorbidities

Data Point	Response Options	Answer required
Date Reported	Date	Required

Does the patient have any non-Niemann Pick disease related medical history and/or co-morbidities?	Check Box	Required
Description	Free Text	Dependent on previous response
Description ICD-10 Code	ICD-10 List	Dependent on previous response
Start Date	Date	Dependent on previous response
Ongoing?	Check Box	Dependent on previous response
End date	Date	Dependent on previous response
Do you suspect a relationship to any of the medications the patient is currently taking?	Check Box	Dependent on previous response
Do you suspect a relationship to Nieman-Pick Specific Medication?	Not Related / Unlikely Related / Possibly Related / Definitely Related / Not Applicable	Dependent on previous response
Please complete an Adverse Drug Reaction form for this Nieman-Pick medication related medical history/comorbidity event	Check Box	Dependent on previous response
NPC Medications		
Data Point	Response Options	Answer required
Date Reported	Date	Required
Is the patient receiving any medication?	Check Box	Required
Medication name	Free Text	Dependent on previous response
Indication description	Free Text	Dependent on previous response
ICD-10 Code selection options	Niemann-Pick Disease ICD-10 codes / Other Disease Indication ICD-10 codes	Dependent on previous response
Niemann Pick Disease ICD-10 codes	E75.240 type A / E75.241 type B	Dependent on previous response
Other Disease Indication ICD-10 Code	ICD-10 List	Dependent on previous response
Start Date	Date	Dependent on previous response
Ongoing?	Check Box	Dependent on previous response
End date	Date	Dependent on previous response
Dose	Numerical	Dependent on previous response
Unit	Drop-Down List	Dependent on previous response
Please specify	Free Text	Dependent on previous response
Route	Drop-Down List	Dependent on previous response
Please specify	Free Text	Dependent on previous response
Frequency	Drop-Down List	Dependent on previous response
Please specify	Free Text	Dependent on previous response
Was treatment discontinued?	Check Box	Dependent on previous response
Reason for treatment discontinuation	Adverse Drug Reaction / Lack of Efficacy / Patient Choice / Unaffordability / Clinician Decision / Change in Treatment Goals / Other	Dependent on previous response
Adverse Drug Reaction ID linked to treatment discontinuation	Free Text	Dependent on previous response
Please specify	Free Text	Dependent on previous response

NPC Adverse Drug Reactions		
Data Point	Response Options	Answer required
Date reported	Date	Required
Has an Adverse Drug Reaction occurred?	Check Box	Required

Adverse Drug Reaction Description	Free Text	Dependent on previous response
Start Date	Date	Dependent on previous response
Ongoing?	Check Box	Dependent on previous response
End Date	Date	Dependent on previous response
Severity	Mild / Moderate / Severe	Dependent on previous response
Do you suspect a relationship to any of the medications the patient is currently taking?	Yes / No	Dependent on previous response
Do you suspect a relationship to Nieman Pick Specific Medication?	Not Related / Unlikely Related / Possibly Related / Probably Related / Definitely Related / Not Applicable	Dependent on previous response
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	Dependent on previous response
Specify other medically important event	Free Text	Dependent on previous response
Initial hospitalisation or Prolongation of current hospitalisation episode?	Initial / Prolongation	Dependent on previous response
Admission date	Date	Dependent on previous response
Discharge date	Date	Dependent on previous response
Outcome	Recovered/Resolved / Recovering/Resolving / Not Recovered/Not Resolved / Recovered/Resolved with Sequelae / Fatal / Unknown	Dependent on previous response
Has any medication been discontinued as a result of ADR ?	Yes / No / Unknown	Dependent on previous response
Did the ADR abate after dose interruption/discontinuation?	Yes / No / Unknown	Dependent on previous response
NPC Pregnancy		
Data Point	Response Options	Answer required
Encounter Date	Date	Required
Has Pregnancy occurred?	Yes / No / Unknown	Required
Date Reported	Date	Dependent on previous response
Date of Last Menstrual Period	Date	Dependent on previous response
Outcome of Pregnancy	Live Birth / Fetal Death / Termination / Spontaneous Abortion	Dependent on previous response
Health Status of live birth	Free Text	Dependent on previous response
NPC End of Data Collection		
Data Point	Response Options	Answer required
End of participation date	Date	Required
Reason for withdrawal of consent	Deceased / Lost to Follow Up / Other	Dependent on previous response
Please specify	Free Text	Dependent on previous response
Date of Death	Date	Dependent on previous response
Lost to follow up date	Date	Dependent on previous response
Status at end of study	Alive / Deceased / Unknown	Dependent on previous response
Cause of Death	Free Text	Dependent on previous response