Extract By Encounter Type

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Additional Review Questions	NSODERF934			
Affected family member, other	NBS0032-01	The relationship of the affected family member to the newborn screen positive infant (DERF) or the affected infant (SDRF)		
Age of Symptom Onset	NBS0139	Age of symptom onset		
Analyte Gene	NDERFANALYG	The gene that was interrogated as part of the infant / child's diagnostic investigations	IL2RG (X-Linked SCID)	
			ADA (Adenosine Deaminase Deficiency)	
			IL7R (AR SCID)	
			JAK3 (AR SCID)	
			PNP (Purine nucleoside phosphorylase deficiency)	
			RAG1 (AR SCID)	
			RAG2 (AR SCID)	
			CD3D (AR SCID)	
			CD3E (AR SCID)	
			CD247 (CD3-zeta defect)	
			DCLRE1C (Omenn syndrome)	
			AK2 (SCID with leukopenia)	
			NHEJ1 (SCID)	
			LIG4 (SCID)	
			RAC2 (neutrophil deficiency syndrome)	
			PTPRC (PTPRC-related SCID)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Analyte Gene	NDERFANALYG	The gene that was interrogated as part of the infant / child's diagnostic investigations	ZAP70 (ZAP70-related SCID)	
			RMRP (Cartilage-Hair hypoplasia)	
			ACADM (Medium-chain acyl-CoA dehydrogenase)	
			ACADVL (Very long chain acyl-CoA dehydrogenase)	
			ACAT1 (acetoacetyl 1-Co A-thiolase)	
			AHCY (S-adenosylhomocysteine hydrolase)	
			ASL (Argininosuccinate lyase)	
			ASS1 (Argininosuccinate synthetase)	
			AUH (AU RNA binding protein/enoyl-Coenzyme A hydratase)	
			BCKDHA (Branched chain keto acid dehydrogenase E1, alpha polypeptide)	
			BCKDHB (Branched chain keto acid dehydrogenase E1, beta polypeptide)	
			BTD (Biotinidase)	
			CBS (Cystathionine beta synthase)	
			CFTR (Cystic fibrosis transmembrane conductance regulator, ATP-binding cassette (sub-family C, member 7) ABCC7)	
			CPT1A (Carnitine Palmitoyltransferase I, liver)	
			CPT2 (Carnitine Palmitoyltransferase 2)	
			CTH (Cystathionase (cystathionine gamma-lyase))	
			CYP21A2 (Cytochrome P450, subfamily XXIA (steroid 21-hydroxylase, congenital adrenal hyperplasia), polypeptide 2)	
			DBT (Dihydrolipoamide branched chain transacylase E2)	
			DLD (Dihydrolipoamide Dehydrogenase)	
			DNAJC19 (Dnaj (hsp40) homologue, subfamily c, member 19)	
			DUOX2 (Dual oxidase 2)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Analyte Gene	NDERFANALYG	The gene that was interrogated as part of the infant / child's diagnostic investigations	ETFA (Electron-transfer-flavoprotein, alpha polypeptide)	
			ETFB (Electron-transfer-flavoprotein, beta polypeptide)	
			ETFDH (Electron-transferring-flavoprotein dehydrogenase)	
			FAH (fumarylacetoacetate hydrolase (fumarylacetoacetase))	
			GALT (Galactose-1-phosphate uridyl transferase)	
			GCDH (Glutaryl-CoA Dehydrogenase)	
			GCH1 (GTP Cyclohydrolase I)	
			GNMT (Glycine N-methyltransferase)	
			HADHA (Long-chain hydroxyacyl-CoA dehydrogenase)	
			HADHA (Mitochondrial trifunctional protein, alpha subunit)	
			HADHB (Mitochondrial trifunctional protein, beta subunit)	
			HBB (hemoglobin, beta)	
			HLCS (Holocarboxylase synthetase)	
			HMGCL (3-hydroxymethyl-3-methylglutaryl-CoA lyase)	
			HPD (4-hydroxyphenylpyruvate dioxygenase)	
			HSD17B10 (hydroxysteroid (17-beta) dehydrogenase 10)	
			IVD (Isovaleryl-CoA dehydrogenase)	
			IYD (Iodotyrosine deiodinase)	
			MAT1A (Methionine adenosyltransferase I, alpha)	
			MCCC1 (Methylcrotonoyl-coenzyme A carboxylase 1 (alpha))	
			MCCC2 (Methylcrotonoyl-coenzyme A carboxylase 2 (beta))	
			MMAA (Methylmalonic aciduria (cobalamin deficiency) type A)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Analyte Gene	NDERFANALYG	The gene that was interrogated as part of the infant / child's diagnostic investigations	MMAB (Methylmalonic aciduria (Cobalamin deficiency) type B)	
			MMACHC (Methylmalonic aciduria (cobalamin deficiency) cblC type)	
			MMADHC (methylmalonic aciduria (cobalamin deficiency) cblD type, with homocystinuria)	
			MUT (Methylmalonyl CoA mutase)	
			OPA3 (Optic atrophy 3 (autosomal recessive, with chorea and spastic paraplegia))	
			PAH (Phenylalanine hydroxylase)	
			PAX8 (Paired box homeotic gene 8)	
			PCBD1 (Pterin-4-alpha-carbinolamine dehydratase 1)	
			PCCA (propionyl CoA carboxylase, alpha polypeptide)	
			PCCB (propionyl CoA carboxylase, beta polypeptide)	
			PTS (6-pyruvoyltetrahydropterin synthase)	
			QDPR (Quinoid dihydropteridine reductase)	
			SLC22A5 (Solute carrier family 22 (organic cation transporter), member 5 (OCTN2))	
			SLC25A13 (Solute carrier family 25, member 13 (Citrin))	
			SPR (sepiapterin reductase)	
			TAT (Tyrosine aminotransferase)	
			TPO (Thyroid peroxidase)	
			TSHR (Thyroid stimulating hormone receptor)	
			ACADSB	
			HBG1	
			HBG2	
			MLYCD	
			PRKDC	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Analyte Gene	NDERFANALYG	The gene that was interrogated as part of the infant / child's diagnostic investigations	SLC25A20	
			HADHA	
			CD320	
			Other, specify:	
Ascertainment of newborn screening missed cases	D0131	The way by which an individual affected with an NSO targeted disease came to medical attention.	Symptoms, specify:	
			Family history / Cascade testing	
			Post mortem / coroner	
			Case finding	
			Other, specify:	
Ascertainment other	D0131-02	The reason(s) that lead to the diagnosis of a newborn screening target disease in an infant who was not ascertained by newborn screening excluding family history, cascade testing, symptoms and post-mortem investigations.		
Ascertainment symptoms	D0131-01	The symptom(s) that lead to the diagnosis of a newborn screening target disease in an infant who was not ascertained by newborn screening		
Biopterin Response Comment	NBS0145	Biopterin Response Comment		
Birth Hospital	Y02091	The name of the hospital where the birth occurred if Birth Location =hospital. Includes all Ontario hospitals with and without birthing units and some out of province and out of country hospitals.		
Birth Location	N0005	The location where the birth occurred (hospital, home, other)	Hospital	
			Home	
			Nursing Station	Primary health care facilities on First Nation reserves.
			Other Ontario location	
			Outside of Ontario	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Carrier Testing Not Offered Reason	NBS0044	Reason why the parents were not offered carrier testing	Previously done	
			Baby adopted or CAS	
			Other, specify reason	
Carrier Testing Not Offered Reason - Other	NBS0045	Specifics for why the parents were not offered carrier testing		
Carrier Testing Offered	NBS0043	Specify whether the baby's parents were offered carrier testing and the subsequent action	Yes, testing performed	
			Yes, will consider	
			Yes, declined	
			Not offered	
Cause of death	NBS0111-03	The cause of death for the infant who had a positive newborn screen (DERF) or the infant affected with an NSO target disorder not ascertained by newborn screening (SDRF)		
Centre for Follow-Up Services	MMMSS0091	Name of the clinical centre that provided follow-up counselling services following the disclosure of the maternal serum screen result.		
Comments in regards to definitive diagnosis	NBS0109	Comments regarding the newborn's definitive diagnosis made after a positive newborn screen (DERF) or regarding an infant affected with an NSO target disorder not ascertained by newborn screening (SDRF)		
Comments regarding ascertainment of missed cases (for data element D0131)	NSOSTFU2010	Comments regarding ascertainment of infants with an NSO target disease who were not ascertained by newborn screening (for data element D0131)		
Comments regarding etiology of transient CH	NSOSTFU2009	Comments regarding etiology of transient CH		
Comments regarding family members of screen positive infant (for data element NBS0030, NBS0031, NBS0108, NBS0032)	NSOSTFU2008	Comments regarding family members of the screen positive infant(DERF) or the infant affected with an NSO target disease not ascertained by newborn screening (SDRF)		
Comments regarding maternal steroids	NSOSTFU2007	Comments regarding maternal steroids		

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Comments regarding the clinical status of the screen positive patient (for data element NBS0111)	NSOSTFU2001	Comments regarding the clinical status of the screen positive patient. If unwell is selected, please indicate the reason(s) why the infant was unwell at retrieval.		
Comments regarding whether the infant was diagnosed prior to retrieval (for data element NBS0110)	NSOSTFU2002	Comments regarding whether the infant was diagnosed prior to retrieval (for data element NBS0110)		
Comments regarding whether the infant was gaining weight at the time of retrieval	NSOSTFU2004	Comments regarding whether the infant was gaining weight at the time of retrieval		
Comments regarding whether the infant was hospitalized due to the screen positive disease at the time of retrieval	NSOSTFU2005	Comments regarding whether the infant was hospitalized at the time of retrieval. If the infant was hospitalized at the time of retrieval, please specify reason(s) here.		
Comments regarding whether the infant was symptomatic of the disease prior to retrieval	NSOSTFU2003	Comments regarding whether the infant had symptoms attributable to the disease for which they screened positive. If the infant did have symptoms, please specify symptoms here.		
Comments regarding whether the infant was virilised	NSOSTFU2006	Comments regarding whether the infant was virilised		
Date DERF completed	NBS0128	The date on which the DERF or SDRF was confirmed to be submitted to BORN by the regional treatment centre. This data element is derived based on the date the DERF / SDRF is confirmed to be submitted to BORN.		
Date of death	NBS0111-02	The date the infant / child died.		
Date of Definitive Diagnosis	NBS0014	Date of definitive diagnosis for the infant / child. This may differ from the date that the diagnosis was communicated to the family / health care provider.		
Date of Diagnostic evaluation	NSOFUDIAGDT	Date of Diagnostic evaluation		
Date of Disposition	NBS0039	Date disposition decision made		

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Date of First Contact with Family / MD in regards to positive Newborn Screen	NBS0017	As reported by the treatment centre, the date of that provider or family made aware of positive newborn screen result		
Date of infant retrieval for diagnostic evaluation after positive Newborn Screen	NBS0016	As reported by regional treatment centre, the date when the screen positive baby was located and diagnostic evaluation begun.		
Date of NSO target disease symptom onset	NBS0013	Date of symptom onset for NSO target disease		
Date Retrieval Confirmation Form Submitted to NSO	NDERFFRM1	The date that the Retrieval Confirmation Information was saved in the BORN system		
Definitive diagnosis after diagnostic evaluation and follow up	NBS0018	Diagnosis made in the infant / child (or mother) following clinical and / or diagnostic evaluations	Not Affected	
			Not affected - Carrier	
			Presumed carrier	
			21-Hydroxylase Deficiency: Non-classical	
			21-Hydroxylase Deficiency: Salt Wasting	
			21-Hydroxylase Deficiency: Simple Virilizing	
			2-methyl-3-hydroxybutyryl CoA dehydrogenase deficiency	
			3-MCC deficiency	
			3-Methylglutaconic aciduria Type 1 (3-methylglutaconyl-CoA hydratase deficiency)	
			3-Methylglutaconic aciduria Type 2 (Barth Syndrome)	
			3-Methylglutaconic aciduria Type 3 (Costeff optic atrophy)	
			3-Methylglutaconic aciduria Type 4	
			3-Methylglutaconic aciduria Type 5 (Dilated cardiomyopathy with ataxia)	
			Beta-kethothiolase deficiency - Variant	
			HMGCoA Lyase Deficiency - Variant	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Definitive diagnosis after diagnostic evaluation and follow up	NBS0018	Diagnosis made in the infant / child (or mother) following clinical and / or diagnostic evaluations	3-MCC deficiency - Variant	
			2-methyl-3hydroxybutyryl CoA dehydrogenase deficiency - Variant	
			Holocarboxylase synthase deficiency - Variant	
			3-Methylglutaconic aciduria Type 1 (3-methylglutaconyl-CoA hyratase deficiency) - Variant	
			3-Methylglutaconic aciduria Type 2 (Bath Syndrome) - Variant	
			3-Methylglutaconic aciduria Type 3 (Costeff optic atrophy) - Variant	
			3-Methylglutaconic aciduria Type 4 - Variant	
			3-Methylglutaconic aciduria Type 5 (Dialted cardiomyopathy w/ ataxia) - Variant	
			Maternal mitochondrial dysfunction	
			CPT2 deficiency - Variant	
			ASA lyase deficiency	
			Beta-ketothiolase deficiency	
			Biotinidase deficiency	
			Carnitine transporter defect	
			Carnitine transporter defect - Variant	
			Maternal MCADD	
			Maternal carnitine deficiency	
			CBS deficiency	
			Classical PKU (Phe >= 1200)	
			CH Athyreosis	
			CH Apparent Athyreosis	
			Moderate PKU (Phe 900-1199)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Definitive diagnosis after diagnostic evaluation and follow up	NBS0018	Diagnosis made in the infant / child (or mother) following clinical and / or diagnostic evaluations	Mild PKU (Phe 600-899)	
			CH Dyshormonogensis	
			CH Ectopic	
			CH Hypoplastic	
			Mild hyperphenylalaninemia (gray zone) (Phe 360-599)	
			Mild hyperphenylalaninemia (benign) (Phe 120-359)	
			Other known cause of TSH elevation, specify:	
			Iodine exposure	
			DHPR Deficiency	
			GTP Cyclohydrolase Deficiency	
			Maternal Disease: Maternal PTU/Graves Disease	
			Prematurity	
			PCD Deficiency	
			Maternal Disease: Thyroid blocking antibodies	
			SR Deficiency	
			PTPS Deficiency	
			DHPR - Variant	
			GTP Cyclohydrolase Deficiency - Variant	
			PCD Deficiency - Variant	
			SR Deficiency - Variant	
			PTPS Deficiency - Variant	
			Maternal PKU	
			CH Undetermined: Presumed Dysgenesis or Dyshormonogenesis	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Definitive diagnosis after diagnostic evaluation and follow up	NBS0018	Diagnosis made in the infant / child (or mother) following clinical and / or diagnostic evaluations	Transient TSH elevation	
			CH Idiopathic: Overt (TSH ≥ 30)	
			CH Idiopathic: Overt (TSH < 30)	
			Central Hypothyroidism	
			CH Idiopathic: Compensated (TSH ≥ 30)	
			CH Idiopathic: Compensated (TSH < 30)	
			Citrullinemia (ASA synthetase deficiency)	
			Citrullinemia Type II (Citrin deficiency)	
			Classical Galactosemia	
			Cobalamin A	
			Cobalamin B	
			Methylmalonyl CoA Mutase deficiency - Variant	
			Methylmalonyl CoA Mutase deficiency (B12 responsive) - Variant	
			Propionic acidemia - Variant	
			Cobalamin A - Variant	
			Cobalamin B - Variant	
			Cobalamin C - Variant	
			Cobalamin D - Variant	
			Cobalamin C	
			Cobalamin D	
			CPT1 deficiency	
			CPT1 deficiency - Inuit Variant	
			CPT2 deficiency	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Definitive diagnosis after diagnostic evaluation and follow up	NBS0018	Diagnosis made in the infant / child (or mother) following clinical and / or diagnostic evaluations	Cystathioninemia	
			Cystic Fibrosis	
			E3 deficiency	
			E3 Deficiency - Variant	
			Glutaric Aciduria Type I low excretor	
			Glutaric Aciduria Type I	
			Glutaric Aciduria Type II	
			Glycine N-Methyltransferase Deficiency	
			HMGCoA Lyase Deficiency	
			Holocarboxylase synthase deficiency	
			Isovaleric acidemia	
			LCHAD deficiency	
			Maternal 3-MCC deficiency	
			Maternal biotin deficiency	
			Maternal carnitine transporter defect	
			Maternal other (Specify:)	
			Maternal Vitamin B12 deficiency	
			Methionine Adenosyltransferase Deficiency	
			Methylmalonyl CoA mutase deficiency	
			Methylmalonyl CoA mutase deficiency (B12 responsive)	
			Mitochondrial trifunctional protein deficiency	
			LCHAD deficiency - Variant	
			Mitochondrial trifunctional protein deficiency - Variant	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Definitive diagnosis after diagnostic evaluation and follow up	NBS0018	Diagnosis made in the infant / child (or mother) following clinical and / or diagnostic evaluations	CACT - Variant	
			Glutaric Aciduria Type II - Variant	
			MSUD	
			Partial Biotinidase Deficiency	
			Profound Biotinidase Deficiency	
			Propionic acidemia	
			S-Adenosylhomocysteine Hydrolase	
			SAM transferase deficency	
			S-Beta thalassemia	
			SC disease	
			Secondary carnitine deficiency	
			SS disease	
			Transient Tyrosinemia	
			Type 1 Tyrosinemia (FAH deficiency)	
			Type 2 Tyrosinemia (TAT deficiency)	
			Type 3 Tyrosinemia	
			Type 1 Tyrosinemia (FAH deficiency) - Variant	
			Type 2 Tyrosinemia (TAT deficiency) - Variant	
			Type 3 Tyrosinemia - Variant	
			VLCAD deficiency - Variant	
			VLCAD deficiency - Classical	
			SCID	
			SCID \ Typical SCID	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Definitive diagnosis after diagnostic evaluation and follow up	NBS0018	Diagnosis made in the infant / child (or mother) following clinical and / or diagnostic evaluations	SCID \ Leaky SCID	
			SCID \ Variant SCID	
			Syndrome with T-cell impairment	
			Syndrome with T-cell impairment \ 22q11.2 deletion syndrome	
			Syndrome with T-cell impairment \ CHARGE syndrome	
			Syndrome with T-cell impairment \ Jacobsen syndrome	
			Syndrome with T-cell impairment \ RAC2 defect	
			Syndrome with T-cell impairment \ DOCK8 deficiency	
			Syndrome with T-cell impairment \ Ataxia telangiectasia	
			Syndrome with T-cell impairment \ Cartilage-hair hypoplasia	
			Syndrome with T-cell impairment \ VACTERL association	
			Syndrome with T-cell impairment \ Barth syndrome	
			Syndrome with T-cell impairment \ TAR syndrome	
			Syndrome with T-cell impairment \ Down syndrome / Trisomy 21	
			Syndrome with T-cell impairment \ Ectrodactyly ectodermal dysplasia syndrome	
			Syndrome with T-cell impairment \ Other, specify:	
			Secondary T-cell lymphopenia	
			Secondary T-cell lymphopenia \ Intestinal lymphangiectasia	
			Secondary T-cell lymphopenia \ Anasarca	
			Secondary T-cell lymphopenia \ Gastroschisis	
			Secondary T-cell lymphopenia \ Third-spacing	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Definitive diagnosis after diagnostic evaluation and follow up	NBS0018	Diagnosis made in the infant / child (or mother) following clinical and / or diagnostic evaluations	Secondary T-cell lymphopenia \ Gastrointestinal atresia	
			Secondary T-cell lymphopenia \ Cardiac surgery +/- thymectomy	
			Secondary T-cell lymphopenia \ Congenital heart defects	
			Secondary T-cell lymphopenia \ Neonatal leukemia	
			Secondary T-cell lymphopenia \ Chylothorax	
			Secondary T-cell lymphopenia \ Chyloperitoneum	
			Secondary T-cell lymphopenia \ Hypoplastic left heart syndrome	
			Secondary T-cell lymphopenia \ Multiple congenital anomalies / NOS	
			Secondary T-cell lymphopenia \ Degenerative neuromuscular disease / NOS	
			Secondary T-cell lymphopenia \ Presumed metabolic disorders / NOS	
			Secondary T-cell lymphopenia \ Unmarkable lymphocytes / NOS	
			Secondary T-cell lymphopenia \ Other, specify:	
			Other, specify:	
			11-Hydroxlyase deficiency	
			3-beta dehydrogenase deficiency	
			11-Hydroxlyase deficiency - Variant	
			3-beta dehydrogenase deficiency - Variant	
			Alpha globin variant(s)	
			Beta and alpha globin variant(s)	
			Beta globin variant(s)	
			Beta thalassemia (ex: beta0)	
			Beta+ thalassemia	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Definitive diagnosis after diagnostic evaluation and follow up	NBS0018	Diagnosis made in the infant / child (or mother) following clinical and / or diagnostic evaluations	C Carrier	
			CACT	
			C-beta thalassemia	
			C-beta+ thalassemia	
			CC disease	
			CF Indeterminate: borderline sweat (+/- genotype)	
			CF Indeterminate: genotype/normal sweat	
			Cobalamin E	
			Cobalamin F	
			Cobalamin G	
			CPTII (myopathic)	
			CPTII (neonatal/infantile)	
			D Carrier	
			DD Variant	
			E Carrier	
			E-beta thalassemia	
			E-beta+ thalassemia	
			EE	
			Gamma globin variant	
			GD Variant	
			Hb H disease	
			HbS/HPFH Ghana deletion	
			HPFH	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Definitive diagnosis after diagnostic evaluation and follow up	NBS0018	Diagnosis made in the infant / child (or mother) following clinical and / or diagnostic evaluations	IVA variant	
			Liver Disease	
			MCAD deficiency - Classical	
			MCAD deficiency - Variant	
			MSUD variant	
			Persistent lab abnormalities	
			Pyridoxine responsive CBS deficiency	
			CBS Deficiency - Variant	
			Pyridoxine responsive CBS deficiency - Variant	
			Cystathioninemia - Variant	
			Glycine N-Methyltransferase Deficiency - Variant	
			Methionine Adenosyltransferase Deficiency - Variant	
			S-Adenoslhomocysteine Hydrolase - Variant	
			SAM transferase deficiency - Variant	
			Citrullinemia (ASA synthetase deficiency) - Variant	
			ASA lysase deficiency - Variant	
			Citrullinemia Type II (Citrin deficiency) - Variant	
			S Carrier	
			SD disease	
			SE disease	
			TCII deficiency	
			Variant hemoglobin - no alpha/beta gene variants detected	
			Vitamin B12 deficiency	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Definitive diagnosis CH FIXED, other	NBS0018-03	The specific reason that an infant with a positive newborn screen for congenital hypothyroidism was diagnosed with FIXED CH that is not athyrotic, ectopic, idiopathic, subclinical or related to dyshormonogenesis		
Definitive diagnosis CH TRANSIENT, other	NBS0018-04	The specific reason that an infant with a positive newborn screen for congenital hypothyroidism was diagnosed with TRANSIENT CH that is not athyrotic, ectopic, idiopathic, subclinical or related to dyshormonogenesis		
Definitive diagnosis, other	NBS0018-02	The diagnosis made in the infant / child (or mother) after investigations following a positive newborn screen when a definitive diagnosis is not available from the pick list.		
DERF completed by	NBS0127	The name of the regional treatment centre health care professional who completed the newborn screen positive infant's DERF / SDRF	ALABDOULSALAM, DR. TAREQ (Children's Hospital of Winnipeg)	
			ANDRIGHETTI, HEATHER (Hamilton Health Sciences)	
			ATHALE, DR. UMA (Hamilton Health Sciences Centre)	
			BABIC, DR. BOJANA (Hamilton Health Sciences Centre)	
			BELANGER, BRIGITTE (Children's Hospital of Eastern Ontario)	
			BELEGRIS, CATHY ((London Health Sciences Centre - Pediatric Endocrinology)	
			BENOIT, CAROLINE (Children’s Hospital of Eastern Ontario)	
			BERES, LORRIE - GC (Hamilton Health Sciences Centre)	
			BOILEAU, CORALEE (CHILDREN'S HOSPITAL OF EASTERN ONTARIO)	
			BOLAND, DR. MARGARET PATRICIA (Children's Hospital of Eastern Ontario)	
			BOUWMAN, THEA - RN (Children's Hospital of Eastern Ontario)	
			BRAGER, RAE (Hamilton Health Sciences)	
			BRICK, LAUREN - GC (Hamilton Health Sciences Centre)	
			CHAKRABORTY, DR. PRANESH (Children's Hospital of Eastern Ontario)	
			CLARSON, DR. CHERIL LINDA (London Health Sciences Centre - Endocrinology)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
DERF completed by	NBS0127	The name of the regional treatment centre health care professional who completed the newborn screen positive infant's DERF / SDRF	COLAICOVO, SAMANTHA (London Health Sciences Centre - Genetics)	
			COOK, CINDY (Children’s Hospital of Eastern Ontario)	
			COSTANTINI, SHARON (Children’s Hospital of Eastern Ontario)	
			COWING, GINA (McMaster Children's Hospital)	
			CRUZ, Vivian (Hospital for Sick Children)	
			DALE, BREANNE (Hamilton Health Sciences Centre)	
			DAVIES, CHRISTINE - GC (Children's Hospital of Eastern Ontario)	
			DEAN, HEATHER	
			DIRAIMO, JENNIFER (London Health Sciences Centre - Genetics)	
			DOLA, ERIN	
			EFFA, ANGELA	
			FRASER, Meghan (Hospital for Sick Children)	
			GALL, KIM - GC (Children's Hospital of Eastern Ontario)	
			GALLEGO, DR. P (London Health Sciences Centre - Endocrinology)	
			GERAGHTY, DR. MICHAEL (Children's Hospital of Eastern Ontario)	
			GIGNAC, MARIEVE (Children's Hospital Of Eastern Ontario)	
			GOOBIE, DR. SHARAN LYNN (London Health Sciences Centre - Genetics)	
			GRASEMANN, HARTMUT (Hospital for Sick Children)	
			HADJIYANNAKIS, DR. ANASTASIA (Children's Hospital of Eastern Ontario)	
			HAMILTON, DR. JILL KRYSTI (Hospital for Sick Children)	
			Hartley, Jessica	
			HEWSON, STACY (Hospital for Sick Children)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
DERF completed by	NBS0127	The name of the regional treatment centre health care professional who completed the newborn screen positive infant's DERF / SDRF	HENDERSON, NATALIE	
			HIGGINS, LAUREN	
			HO, STEPHANIE (London Health Sciences Centre - Genetics)	
			JARDINE, DR. Lawrence (London Health Sciences Centre - Genetics)	
			JOHNSON, KIRSTI (Hospital for Sick Children)	
			JUSKEY, LIA (RN)	
			KIRBY, DR. MELANIE-ANN (Hospital for Sick Children)	
			KIROUAC, Nicole	
			KLAASSEN, DR. ROBERT JOHN (Children's Hospital of Eastern Ontario)	
			KOVESI, DR. THOMAS ANDREW (Children's Hospital of Eastern Ontario)	
			KOZENKO, DR. MARIYA (Hamilton Health Sciences Centre)	
			KRONICK, DR. JONATHAN (Metabolics)	
			LANGDON, KRISTEN (London Health Sciences Centre)	
			ROADHOUSE, CHELSEA (Hamilton Health Sciences Centre)	
			LAWRENCE, DR. SARAH ELIZABETH (Children's Hospital of Eastern Ontario)	
			WATTS-DICKENS, ABBY (Hospital for Sick Children)	
			LEPPINGTON, SARAH (Children's Hospital of Western Ontario)	
			LESCADRE, TAMMY ((London Health Sciences Centre - Pediatric Endocrinology)	
			LI, DR. CHUMEI (Hamilton Health Sciences Centre)	
			LOCKYER, LINDSAY (The Hospital for Sick Children)	
			LOOMER-VANDERSLUIS, MADELINE (Hamilton Health Sciences Centre)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
DERF completed by	NBS0127	The name of the regional treatment centre health care professional who completed the newborn screen positive infant's DERF / SDRF	MACKENZIE, DR. JENNIFER JANE (Kingston General Hospital)	
			MAHMUD, DR. FARID HUSSAIN (Hospital for Sick Children)	
			MAHMUTOGLU, DR. SAADET (Metabolics)	
			MARCADIER, JANET - GC (Children's Hospital of Eastern Ontario)	
			WATTS-DICKENS, ABBY (Hospital for Sick Children)	
			MAYO, Jane	
			McASSEY, DR. KAREN LYNN (Hamilton Health Sciences Centre)	
			MCKAYRN, ANNA	
			MCGREGOR, ANGELA (Kingston)	
			McSHANE, HOLLY (RN - Hotel Dieu Hospital)	
			MHANNI, DR AZIZ (WRHA Genetics & Metabolism Program)	
			MORAR, OANA (London Health Sciences Centre - Genetics)	
			NAPIER, MELANIE (London Health Sciences Centre - Genetics)	
			NOWACZYK, DR. MALGORZATA JOANNA (Hamilton Health Sciences Centre)	
			ODAME, DR. ISAAC (Hospital for Sick Children)	
			MUCHANTEF, KIM (Children’s Hospital of Eastern Ontario)	
			PALMERT, DR. MARK RANEY (Hospital for Sick Children)	
			PARADISO, CHRISTINA - GC (Hamilton Health Sciences Centre)	
			PASTERKAMP, DR. HANS (CHILDREN'S HOSPITAL OF WINNIPEG)	
			PEDDER, DR. LINDA (Hamilton Health Sciences Centre)	
			PERLMAN, DR. KUSIEL (Hospital for Sick Children)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
DERF completed by	NBS0127	The name of the regional treatment centre health care professional who completed the newborn screen positive infant's DERF / SDRF	PIRES, LINDA (Canadian Centre for Primary Immunodeficiency)	
			POTTER, DR. MURRAY ALEXANDER (Hamilton Health Sciences Centre)	
			PRASAD, DR. CHITRA (London Health Sciences Centre - Genetics)	
			PRICE, DR. APRIL KATHERINE (London Health Sciences Centre - Genetics)	
			RAIMAN, DR. JULIAN ANDREW JONATHON (Hospital for Sick Children)	
			RATJEN, DR. FELIX ALEXANDER (Hospital for Sick Children)	
			REID, BRENDA (Canadian Centre for Primary Immunodeficiency)	
			ROCKMAN-GREENBERG, DR. CHERYL (WRHA Genetics & Metabolism Program)	
			RYBANSKY, SUSAN (London Health Sciences Centre - Genetics)	
			SALEH, DR.DAVID (Kingston General Hospital)	
			SAMAAN, DR. CONSTANTINE (Hamilton Health Sciences Centre)	
			ROBART, SARAH (Hospital for Sick Children)	
			SCHULZE, DR. ANDREAS (Hospital for Sick Children)	
			SELLERS, ELIZABETH	
			SHAVER, JENNIFER (Kingston General Hospital)	
			SILVA, DR. MARIANA (Kingston General Hospital)	
			SIRIWARDENA, DR. KOMUDI PULSARA (Hospital for Sick Children)	
			SOCHETT, ETIENNE (Hospital for Sick Children)	
			SOLOMON, MELINDA (Hospital for Sick Children)	
			SOTTOSANTI, DR. M (London Health Sciences Centre - Endocrinology)	
			STEIN, DR. ROBERT IAN (London Health Sciences Centre - Endocrinology)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
DERF completed by	NBS0127	The name of the regional treatment centre health care professional who completed the newborn screen positive infant's DERF / SDRF	SVELA, YULIA (Hospital for Sick Children)	
			TAM, KAREN (Hospital for Sick Children)	
			TAZBAZ, TANYA - RN (Hospital for Sick Children)	
			Thibert, Anna(WRHA Respirology)	
			URBACH, DR. STACEY LISA (Hospital for Sick Children)	
			VAN WYLICK, DR. RICHARD CECIL (Kingston General Hospital)	
			VANDERMEULEN, DR. JOHN AUGUST (Hamilton Health Sciences Centre)	
			WAGLER, STEOHANIE - GC (Hamilton Health Sciences Centre)	
			WASSERMAN, DR. JONATHAN (Metabolics)	
			WHERRETT, DR. DIANE KATHERINE (Hospital for Sick Children)	
			WICKLOW, BRANDY	
			ZELENIETZ, SARI - GC (Children's Hospital of Eastern Ontario)	
			Zingerat, Sharri-Lynne (Sudbury CF Clinic)	
			TELLIER, ISABELLE (CHU St Justine)	
			LEVASSEUR, MICHELE (Children’s Hospital of Eastern Ontario)	
			BRUNEL-GUITTON, DR.CATHARINE (CHE Sainte Justine, Medical Genetics)	
			MAZUR, Katherine (Hamilton Health Sciences)	
			Mol, Jarmila RN (Hospital for Sick Children)	
			Brittney Johnstone (Hospital for Sick Children)	
			Sabrina Bhathal (Hospital for Sick Children)	
			ANDRIGHETTI, HEATHER (London Health Sciences Centre)	
			Other, specify	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
DERF Follow-Up Visit Required Flag	NDERFFVISR	DERF Follow-Up Visit Required Flag		
DERF HCP not informed reason	NDERFHCPINFR	DERF HCP not informed reason	Triaged	
			Awaiting call back from parents	
			Incorrect contact information	
			No contact information for primary health care provider	
			Family has moved	
			Phone number out of service	
			Awaiting call back from primary health care provider	
			Unable to contact health care provider	
			Infant is deceased	
			Transferred to another treatment centre	
			Incorrect health care provider listed	
			No voice mail to leave message	
			Registered mail undeliverable	
			CAS involved to help locate	
			Family / parents out of country	
			Appointment pending	
			Other	
			No health care provider indicated	
			Parents contacted directly	
			No contact information for family	
DERF is CCPID flag	NDERFCCPID	DERF is CCPID flag		

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
DERF is user added	NDERFUSRADD	DERF is user added		
DERF results appended details	NDERFRESAPPDET	Investigations specified to be faxed to NSO at the given fax number		
DERF results appended?	NDERFRESAPP	Indicates whether the treatment centre will fax results from the diagnostic investigations performed on the referred infant to NSO. If checked, please print a copy of the DERF / SDRF and fax DERF / SDRF and appropriate investigations to NSO at the given fax number		
DERF review date	NDERFREVD	DERF review date		
Determination ID	D0067	Unique number linked to test ID and result flag		
Diagnosis Made Outside of Regional Treatment Centre Flag	NBS0143	Indicates if the diagnoses was made outside of a Regional Treatment Centre. If so, please enter the SRDF Completed by and Responsible Physician boxes.		
Diagnosis prior to retrieval of patient	NBS0110	Indicates that the infant was diagnosed with the disease for which they screened positive prior to being retrieved for diagnostic evaluation		
Diagnostic testing pending or initiated flag	NSODDTINIT			
Disposition - Date treatment initiated (CCPID)	NBS0039-6	Disposition - Date treatment initiated (CCPID)		
Disposition - Date treatment initiated (RTC)	NBS0039-2	Disposition - Date treatment initiated (RTC)		
Disposition - Date treatment stopped (CCPID)	NBS0039-8	Disposition - Date treatment stopped (CCPID)		
Disposition - Date treatment stopped (RTC)	NBS0039-4	Disposition - Date treatment stopped		
Disposition - Was treatment ever initiated (CCPID)	NBS0039-5	Disposition - Was treatment ever initiated (CCPID)		
Disposition - Was treatment stopped (CCPID)	NBS0039-7	Disposition - Was treatment stopped (CCPID)		

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Disposition - Was treatment stopped (RTC)	NBS0039-3	Disposition - Was treatment stopped (RTC)		
Disposition after diagnostic evaluation and follow up	NBS0038	After clinical and / or diagnostic evaluations are completed, the decision to discharge, continue to follow without treatment or initiate treatment as indicated by regional treatment centre.	Continue to follow with no treatment	
			Discharge	
			Initiate Treatment	
			Refer to the Canadian Centre for Primary Immunodeficiency	
Disposition rationale	NBS0038-01	Comments regarding the disposition. If 'continue to follow without treatment' is selected, please specify reason(s) / rationale.		
Etiology of transient CH	NSOSTFU015	The etiology of the newborn's transient congenital hypothyroidism as determined after diagnostic investigations and evaluations following a positive newborn screen	Maternal PTU / Grave's treatment	
			Thyroid blocking antibodies	
			Prematurity	
			Other known cause, specify:	
			Unknown	
Etiology of transient CH, other	NSOSTFU015-01	An indication of the cause of the transient congenital hypothyroidism in the infant with a positive newborn for CH excluding maternal PTU/Grave's treatment, thyroid blocking antibodies and prematurity		
Family History of dyshormonogenesis	NSODERF756			
Gene Variant Presumed Homozygous flag	NDERFGNPRSHMY	Gene Variant Presumed Homozygous flag		
Genetic Testing Methodology	NSODERFGTM	Genetic testing methodology	Common Mutation	
			Panel	
			Sequencing \ Single Gene Sequencing	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Genetic Testing Methodology	NSODERFGTM	Genetic testing methodology	Sequencing \ Gene Panel Sequencing	
			Sequencing \ Whole Exome	
			Deletion/Duplication	
			Deletion/Duplication \ FISH	
			Deletion/Duplication \ Microarray	
			Deletion/Duplication \ MLPA	
			Deletion/Duplication \ QPCR	
Genetic Testing Type (Maternal / Paternal)	FAN1001	See "Molecular Genetic Testing" & "Karyotype Genetic Testing" tabs		
Health of screen positive infant at retrieval	NBS0111	What was the infant's health at the time they were located for diagnostic evaluation after a positive newborn screen	Well	
			Unwell, specify:	
			Deceased, specify date and cause	
Hemoglobin method after positive newborn screen	NSOSTFU016	The method by which an infant's hemoglobin pattern and the percentages of each hemoglobin detected are determined after a positive newborn screen		
How dyshormonogenesis diagnosis was made	NSODERF757			
If the DERF and review process complete?	NDERFCMPL	Indicates whether the DERF / SDRF and the NSO review process of the DERF / SDRF are complete. This field is to be completed by NSO users only.		
Infant Not Referred Followup	NDERFNRF	Indicates that the infant was not referred for followup		
Is further information needed from the Treatment Centre	NSODERF931			
Missed case NSO_accession_num	MDERFMCACC	The NSO accession number as recorded on the symptomatic diagnosis report form		
Molecular Result Interpretation	NSOSTFU012I	The interpretation of the results of the molecular investigations	Non-Pathogenic Polymorphism – Previously Reported	
			Pathogenic Mutation - Previously Reported	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Molecular Result Interpretation	NSOSTFU012I	The interpretation of the results of the molecular investigations	Presumed Non-Pathogenic Polymorphism – Previously Unreported	
			Presumed Pathogenic Mutation – Previously Unreported	
			Variant of Unknown Signficance – Previously Reported	
			Variant of Unknown Signficance – Previously Unreported	
Newborn Screen Positive Infant Hospitalized due to (screen positive disease name)	NBS0122	An indication of whether an infant with a positive newborn screen was hospitalized at the time of retrieval for diagnostic evaluation		
Newborn Screen Positive Infant Symptomatic of (screen positive disease name)	NBS0120	An indication of whether the infant had symptoms that could be attributed to the disease they were screen positive for at the time of retrieval		
Newborn Screen Positive Infant Virilised	NBS0125	An indication of whether the infant with a newborn screen positive for congenital adrenal hyperplasia was virilised at birth, noted at the time of retrieval for diagnostic evaluation		
Newborn screen positive infant's health care provider at diagnostic testing	NBS0114	Newborn screen positive infant's primary health care provider at the time of retrieval and / or diagnostic testing.		
Newborn screen positive infant's name if different at diagnostic testing	NBS0113	Newborn screen positive infant's name at diagnostic testing. Only captured if newborn's name is different than Newborn Name - Family Name (N0001)		
NICU Length of Stay	N0080	Calculated length of neonatal stay in NICU/SCN		
No follow up diagnostic testing initiated	NDERF02324	Indicates that no diagnostic testing was initiated in follow up to a infant / child's referral to a Regional Treatment Centre from Newborn Screening Ontario	After clinical evaluation by specialist physician completed, diagnostic testing deemed unnecessary	
			Parents declined – previous child had a positive screen and normal diagnostic testing	
			Parents declined – current child had a negative screen in addition to a positive screen	
			Parents refused	
			Infant deceased	
			Other	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
No follow up diagnostic testing initiated Other	NDERF02324OTHER	An indication that the reason that no follow up diagnostic testing was initiated is not captured by the values presented in the pick list		
NSO Accession Number	NBS0033	Unique NSO identifier assigned at the time of NSO Accessioning.		
NSO Cough or Wheeze (CF)	NBS0124	An indication of whether the infant with a newborn screen positive for cystic fibrosis had a cough or wheeze at the time of retrieval for diagnostic evaluation		
NSO DERF CH Imaging by Ultrasound	NSODERFCHUSIMG	Imaging results of the thyroid gland	No thyroid tissue seen (Athyrotic)	
			Reduced (Hypoplastic)	
			Enlarged (Goiter)	
			Normal	
			Not reported	
NSO DERF CH Thyroid Scan	NSODERFCHTS	Results of the nuclear medicine examination using a radioactive iodine tracer to determine thyroid location and function	Increased uptake	
			Decreased uptake	
			Normal	
			No uptake	
			Not reported	
NSO DERF Form Type	NDERFFRMTYPE	Indicates the disease specific Diagnostic Evaluation Report Form	Biotinidase Deficiency	
			C5OH	
			Congenital Adrenal Hyperplasia (CAH)	
			Cystic Fibrosis (CF)	
			Congenital Hypothyroidism (CH)	
			Citrullinemias	
			CPT I	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Form Type	NDERFFRMTYPE	Indicates the disease specific Diagnostic Evaluation Report Form	CPT II	
			CUD	
			GA1	
			Galactosemia	
			Homocystinuria	
			Hemoglobinopathies	
			Isovaleric Acidemia (IVA)	
			LCHAD	
			MCADD	
			Maple Syrup Urine Disease (MSUD)	
			PA/MMA	
			Phenylketonuria (PKU)	
			Severe Combined Immunodeficiency (SCID)	
			Tyrosinemias	
			VLCAD	
			Generic Form	
NSO DERF Gene Allele 1	NSOFUGNALLELE1	NSO DERF Gene Allele 1	c.1001A>G (p.Lys334Arg; p.K334R)	
			c.1009_1011del (p.Leu337del)	
			7T	
			9T	
			5T	
			c.1A>G (p.Met1Val; p.M1V)	
			c.2002C>T (p.Arg668Cys; p.R668C)	
			c.199-10T>G	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 1	NSOFUGNALLELE1	NSO DERF Gene Allele 1	c.19G>A (p.Asp7Asn; Hb Dunn)	
			c.1868G>T (p.Gly623Val; p.G623V)	
			c.194T>C (p.Ile65Thr; p.I65T)	
			c.1843C>T (p.Arg615Ter)	
			c.1844G>A (p.Arg615Gln; p.R615Q)	
			c.1741G>T (p.Val581Phe; p.V581F)	
			c.1793A>G (p.His598Arg; p.H598R)	
			c.1717-1G>T	
			c.1727G>C (p.Gly576Ala; p.G576A)	
			c.-170G>A (Sardinian HPFH)	
			c.1717-1G>A	
			c.17_del	
			c.1700G>A (p.Arg567Gln; p.R567Q)	
			c.1678+1G>A	
			c.17_18del	
			c.163_167delTTTTTinsAA	
			c.165del (c.165delT)	
			c.1616A>G (p.Gln539Arg; p.Q539R)	
			c.1627delG	
			c.1603C>G (p.Leu535Val; p.L535V)	
			c.1612C>T (p.Arg538Cys; p.R538C)	
			c.157G>A (p.Asp53Asn; Hb Osu Christiansborg)	
			c.1584G>A (p.Glu528Glu; p.E528E)	
			c.1528G>C (p.Glu510Gln; p.E510Q)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 1	NSOFUGNALLELE1	NSO DERF Gene Allele 1	c.157C>T (p.Arg53Cys; p.R53C)	
			c.1500_1502del (p.Leu502del)	
			c.151C>T (p.His51Tyr; p.H51Y)	
			c.1475C>T (p.Thr492Ile; p.T492I)	
			c.1489C>T (p.Pro497Ser; p.P497S)	
			c.1451A>G (p.His484Arg; p.H484R)	
			c.1475A>C (p.Asp492Ala; p.D492A)	
			c.143T>C (p.Leu48Ser; p.L48S)	
			c.1451_1452delGGinsC (p.Arg484Profs)	
			c.1408A>G (p.Met470Val; M470V)	
			c.142G>C (p.Asp48His; Hb Hasharon)	
			c.138C>T	
			c.-138C>T	
			c.-137C>G	
			c.1388G>A (p.Gly463Glu; p.G463E)	
			c.1375dupC (p.Arg459Profs)	
			c.1376G>A (p.Arg459Gln; p.R459Q)	
			c.1363delC (p.Leu455Phefs)	
			c.1367G>A (p.Arg456His; p.R456H)	
			c.1360G>A (p.Asp454Asn; p.D454N)	
			c.1361A>G (p.Tyr454Cys; p.Y454C)	
			c.133G>A (p.Gly45Arg; p.G45R)	
			c.1348T>G (p.Trp450Gly; p.W450G)	
			c.-132-329A>G	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 1	NSOFUGNALLELE1	NSO DERF Gene Allele 1	c.1324G>T (p.Val442Phe; p.V442F)	
			c.1315+1G>A	
			c.1322G>A (p.Gly441Asp; p.G441D)	
			c.1253G>A (p.Ser418Asn; p.S418N)	
			c.1312T>A (p.Tyr438Asn; p.Y438N)	
			c.1241A>G (p.Tyr414Cys; p.Y414C)	
			c.1247T>C (p.Ile416Thr; p.I416T)	
			c.1213A>G (p.Met405Val; p.M405V)	
			c.1222C>T (p.Arg408Trp; p.R408W)	
			c.-11G>T	
			c.1208C>T (p.Ala403Val; p.A403V)	
			c.1182+1G>A	
			c.1193C>A (p.Ala398Asp; p.A398D)	
			c.1171C>T (p.Pro391Ser; p.P391S)	
			c.1173C>A (p.Phe391Leu; p.F391L)	
			c.1168G>A (p.Gly390Arg; p.G390R)	
			c.1169A>G (p.Glu390Gly; p.E390G)	
			c.1135G>A (p.Ala379Thr; p.A379T)	
			c.1148T>A (p.Phe383Tyr; p.F383Y)	
			c.1091-2_1092del4ins18	
			c.1115C>A (p.Ala372Asp; p.A372D)	
			c.1066-11G>A	
			c.1068T>A	
			c.105_109del (p.Arg37Alafs*20)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 1	NSOFUGNALLELE1	NSO DERF Gene Allele 1	c.1052delC (p.Thr351Lysfs*12; p.T351Kfs*12)	
			c.1046C>T (p.Ala349Val; p.A349V)	
			c.1049 C>A (p.Ser350Tyr; p.S350Y)	
			c.1040G>C (p.Arg347Pro; p.R347P)	
			c.1045C>T (p.Arg349Ter; p.R349X)	
			c.1016C>T (p.Ser339Leu; p.S339L)	
			c.1027G>C (p.Gly343Arg; p.G343R)	
			c.100G>A (p.Gly34Ser; p.G34S)	
			c.1010A>C (p.Tyr337Ser; p.Y337S)	
			c.604T>C (p.Phe202Leu; p.F202L)	
			c.609G>A (p.Trp203Ter)	
			c.577C>T (p.Arg193Trp; p.R193W)	
			c.579+1G>T (711+1G>T)	
			c.572T>C (p.Met191Thr; p.M191T)	
			c.575T>G (p.Phe192Cys; p.F192C)	
			c.56_57delGCinsAA (p.Arg19Gln; p.R19Q)	
			c.568C>T (p.His190Tyr; p.H190Y)	
			c.556C>T (p.His186Tyr; p.H186Y)	
			c.557G>A (p.Cys186Tyr; p.C186Y)	
			c.547C>T (p.Arg183Trp; p.R183W)	
			c.556C>T (p.Arg186Trp; p.R186W)	
			c.51dupT (p.Ala5CysfsX5)	
			c.535G>T (p.Gly179Trp; p.G179W)	
			c.518T>A (p.Ile173Asn; p.I173N)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 1	NSOFUGNALLELE1	NSO DERF Gene Allele 1	c.51C>G (p.Phe17Leu; p.F17L)	
			c.50G>A (p.Gly17Asp; Hb J-Baltimore)	
			c.511G>A (p.Ala171Thr; p.A171T)	
			c.473G>A (p.Arg158Gln; p.R158Q)	
			c.507delG (p.Glu169AspfsX11)	
			c.456G>A (p.Trp152Ter)	
			c.470G>A (p.Arg157His; p.R157H)	
			c.443G>A (p.Arg148Lys; p.R148K)	
			c.443T>C (p.Ile148Asn; p.I148N)	
			c.443A>G (p.Gln148Arg; p.Q148R)	
			c.443C>T (p.Thr148Ile; p.T148I)	
			c.44 T>C (p.Val5Ala; p.V5A)	
			c.442delA (p.Ile148Leufs)	
			c.4389G>A (p.Gln1463Gln; p.Q1463Q)	
			c.43G>T (p.Gly15Trp; p.G15W)	
			c.436C>T (p.Arg146Trp; pR146W)	
			c.437delT (p.Leu146Trpfs)	
			c.428A>G (p.Asp143Gly; p.D143G)	
			c.42C>A	
			c.-41A>C	
			c.4272C>T (p.Tyr1424Tyr; p.Y1424Y)	
			c.394C>T (p.Arg132Stop; p.R132X)	
			c.397G>T (p.Val133Leu; p.V133L)	
			c.3889dupT(p.Ser1297Phefs*5)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 1	NSOFUGNALLELE1	NSO DERF Gene Allele 1	c.3905T>C (p.Met1302Thr; p.M1302T)	
			c.387+91dupA	
			c.3879delA	
			c.384-20A>G	
			c.3846G>A (p.Trp1282Ter; W1282X)	
			c.382_384delTAC (p.Tyr128del)	
			c.383_385del (p.Val128del)	
			c.3794G>T (p.Gly1265Val; p.G1265V)	
			c.380C>T (p.Pro127Leu; p.P127L)	
			c.372T>A (p.Asp142Glu; p.D142E)	
			c.377G>A (p.Gly126Asp; p.G126D)	
			c.362C>T (p.Thr121Ile; p.T121I)	
			c.366T>G (p.Asn122Lys; p.D122K)	
			c.349C>T (p.Arg117Cys; p.R117C)	
			c.34G>A (p.Ala12Thr; p.A12T)	
			c.332_339delGAGACTAC (p.Gly111Valfs)	
			c.3486_3487delAG	
			c.3222T>A (p.Phe1074Leu; p.F1074L)	
			c.331C>T (p.Arg111Ter)	
			c.3196C>T (p.Arg1066Cys; p.R1066C)	
			c.3197G>A (p.Arg1066His; p.R1066H)	
			c.315+1G>A	
			c.316-2A>G	
			c.293-13C>G	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 1	NSOFUGNALLELE1	NSO DERF Gene Allele 1	c.3067_3072delTAGTG (p.Ile1023Serfs)	
			c.287G>T (p.Gly96Val; p.G96V)	
			c.290A>T (p. Asp97Val; p.D97V)	
			c.281 A>G (p.Asp94Gly; p.D94G)	
			c.283G>A (p.Asp95Asn; Hb F Columbus-GA)	
			c.262_264delGAG (p.Glu88del; p.E88del)	
			c.276G>T (p.Glu92Asp; p.E92D)	
			c.2562T>G (p.Thr854Thr; 2694T>G)	
			c.2604A>G	
			c.2490+1G>A (2622+1G>A)	
			c.253G>T (p.Gly85Cys; p.G85C)	
			c.233T>C (p.Ile78Thr; p.I78T)	
			c.236G>A (p.Arg79His; p.R79H)	
			c.227T>C (p.Ile76Thr; Hb F Waynesboro)	
			c.227T>C (p.Leu76Pro; Hb Atlanta)	
			c.224G>A (p.Arg75Gln; R75Q)	
			c.2260G>A (p.Val754Met; V754M)	
			c.216+2T>C	
			c.223C>T (p.Arg75Ter)	
			c.209C>T (p.Ser70Leu; p.S70L)	
			c.2128A>T (p.Lys710Ter)	
			c.200C>T (p.Thr67Met; p.T67M)	
			c.2015_2052delAAinsG (p.Lys684SerfsX38; 2183AA>G)	
			c.887_888delCT (p.Pro296Argfs)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 1	NSOFUGNALLELE1	NSO DERF Gene Allele 1	c.887G>A (p.Arg296Gln; p.R296Q)	
			c.877G>A (p.Ala293Thr; p.A293T)	
			c.881G>A (p.Gly294Glu; p.G294E)	
			c.860G>A (p.Gly287Asp; p.G287D)	
			c.865G>A (p.Gly289Arg; p.G289R)	
			c.855G>T (p.Lys285Asn; p.K285N)	
			c.8591A>C (p.His2864Pro; p.H2864P)	
			c.782G>A (p.Arg261Gln; R261Q)	
			c.787G>A (p.Val263Met; p.V263M)	
			c.775C>T (p.Arg259Trp; p.R259W)	
			c.779C>T (p.Thr260Met; p.T260M)	
			c.753-2A>C	
			c.757G>A (p.Asp253Asn; p.D253N)	
			c.744-33GATT[7]+[7]	
			c.753-23_753-22del	
			c.728G>A (p.Arg243Gln; p.R243Q)	
			c.734T>C (p.Val245Ala; p.V245A)	
			c.719T>A (p.Met240Lys; p.M240K)	
			c.722G>A (p.Arg241His; p.R241H)	
			c.710T>A (p.Ile237Asn; p.I237N)	
			c.713T>A (p.Val238Glu; p.V238E)	
			c.701G>A (p.Arg234Gln; p.R234Q)	
			c.709G>A (p.Ala237Thr; p.A237T)	
			c.688-2A>G	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 1	NSOFUGNALLELE1	NSO DERF Gene Allele 1	c.701C>T (p.Thr234Ile; p.T234I)	
			c.682G>T (p.Asp228Tyr; D228Y)	
			c.683A>G (p.Asn228Ser; p.N228S)	
			c.658_660delAAG (p.Lys220del)	
			c.680A>T (p.Asp227Val; p.D227V)	
			c.652C>T (p.Leu218Leu; p.L218L)	
			c.656G>A (p.Arg219His; p.R219H)	
			c.645C>T (p.Leu215Leu; p.L215L)	
			c.64C>T (p.Pro22Ser; p.P22S)	
			c.641A>G (p.Asn214Ser; p.N214S)	
			c.641T>A (p.Ile214Lys; p.I214K)	
			c.638T>C (p.Leu213Pro; p.L213P)	
			c.641A>C (p.Asn214Thr; p.N214T)	
			c.631C>T (p.Arg211Cys; p.R211C)	
			c.632G>A (p.Arg211His; p.R211H)	
			c.622C>T (p.Arg208Ter)	
			c.629A>G (p.Tyr210Cys; p.Y210C)	
			c.956C>T (p.Ser319Phe; p.S319F)	
			c.958T>G (p.Leu320Val; p.L320V)	
			c.950T>C (p.Val317Ala; p.V317A)	
			c.955C>T (p.Gln319Ter)	
			c.946-4C>G	
			c.948delT (1078delT)	
			c.939A>G (p.Ile313Met; p.I313M)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 1	NSOFUGNALLELE1	NSO DERF Gene Allele 1	c.945+1G>C	
			c.93-21G>C	
			c.937C>T (p.Arg313Trp; p.R313W)	
			c.92C>T (p.Pro31Leu; p.P31L)	
			c.92G>C (p.Arg31Thr; Hb Kairouan)	
			c.923dupT (p.Leu308Phefs)	
			c.925G>A (p.Gly309Arg; p.G309R)	
			c.916A>G (p.Ile306Val; p.I306V)	
			c.92+1G>T	
			c.896T>G (p.Phe299Cys; p.F299C)	
			c.898G>T (p.Ala300Ser; p.A300S)	
			c.896A>T (p.Lys299Met; p.K299M)	
			c.896C>T (p.Ala299Val; p.A299V)	
			c.895C>T (p.Arg299Cys; p.R299C)	
			c.896_898delAGA (p.Lys299del)	
			SNP rs140871321	
			SNP rs150422765 (intron variant)	
			SNP rs11316888 (intron variant)	
			SNP rs12022011 (intron variant)	
			c.991T>C (p.Cys331Arg; p.C331R)	
			c.996+17G>A	
			c.977T>C (p.Val326Ala; p.V326A)	
			c.990dupT (p.Glu331Terfs)	
			c.970C>T (p.Arg324Ter)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 1	NSOFUGNALLELE1	NSO DERF Gene Allele 1	c.971G>T (p.Gly324Val; p.G324V)	
			SNP rs41274867 (benign)	
			SNP rs60400822 (benign)	
			SNP rs3817641 (benign)	
			SNP rs41274865 (benign)	
			SNP rs17671352 (benign)	
			SNP rs2277202 (benign)	
			SNP rs6145976 (benign)	
			SNP rs77680021	
			SNP rs8064573	
			c.847_850delCTGG	
			619 bp deletion (g.71609_72227del619)	
			c.1000C>T (p.Arg334Trp; R334W)	
			c.1040G>A (p.Arg347His; R347H)	
			c.1062+5G>A (IVS12+5 G>A)	
			c.-119_-116delGTCA (5'UTR-119delGTCA)	
			c.1193C>T (p.P398L)	
			c.126_129delCTTT (p.Phe42fs)	
			c.1316G>A; (p.Gly439Asp; p.G439D)	
			c.1327_1330dup (p.Ile444Argfs)	
			c.1330G>C (p.D444H)	
			c.1364C>A (p.Ala455Glu; A455E)	
			c.1367T>C (p.Val456Ala)	
			c.1368A>C (p.Q456H)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 1	NSOFUGNALLELE1	NSO DERF Gene Allele 1	c.1413T>C (p.C471C)	
			c.1436C>T (p.P479L; p.Pro479Leu)	
			c.1463G>A (p.R488H)	
			c.1466C>A (p.Ser489X; S489X)	
			c.1508_1512delCCATG	
			c.1519_1521delATC (p.Ile507del; I507del; DI507)	
			c.1521_1523delCTT (p.Phe508del; F508del; dF508)	
			c.1523T>G (p.Phe508Cys; F508C)	
			c.1558G>T (p.Val520Phe; V520F)	
			c.1595C>T (p.T532M)	
			c.1616C>A	
			c.1624G>T (p.Gly542X; G542X)	
			c.1630G>A (p.Gly544Ser)	
			c.1646G>A (p.Ser549Asn; S549N)	
			c.1652G>A (p.Gly551Asp; G551D)	
			c.1657C>T (p.Arg553X; R553X)	
			c.1675G>A (p.Ala559Thr; A559T)	
			c.1679G>C (p.Arg560Thr; R560T)	
			c.1705T>G (p.Tyr569Asp; Y569D)	
			c.1766+1G>A (1898+1G>A)	
			c.178G>T (p.Glu60X; E60X)	
			c.190A>G (p.Met64Lys)	
			c.192G>T (p.Gln64His; Q64H)	
			c.199 T>C (p.Tyr67His; p.Y67H))	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 1	NSOFUGNALLELE1	NSO DERF Gene Allele 1	c.19G>A (HbC; p.Glu7Lys)	
			c.200C>T (p.Pro67Leu; P67L)	
			c.20A>T (HbS; p.Glu7Val)	
			c.216+1G>A	
			c.2162T>A (p.Val721Glu)	
			c.220G>A (p.Asp74Asn; Hb G-Accra; Korle-Bu variant of Hb)	
			c.223G>C (p.Asp74Gly; HbQ-Thailand)	
			c.2246G>T (p.R749L; p.Arg749Leu)	
			c.233C>A (p.Pro78His; HbToulon)	
			c.237C>A (p.Asn79Lys; HbStanleyville-II)	
			c.254G>A (p.Gly85Glu; G85E)	
			c.262_263delTT (p.Leu88IlefsX22; 394delTT)	
			c.263T>A (p.Leu88X)	
			c.2657+2_2657+3insA (2789+2insA)	
			c.2657+5G>A (2789+5G>A)	
			c.271dupA (p.Arg91Lysfs)	
			c.2734T>A (p.Ser912Thr)	
			c.278-41del	
			c.27dupG (c.27_28insG; p.Ser10Valfs*14)	
			c.2988+1G>A (3120+1G>A)	
			c.305G>A (p.Arg117His; R117H)	
			c.307A>C (p.Ser103Arg; HbManitoba I)	
			c.3080T>C (p.Ile1027Thr; I1027T)	
			c.3276C>A (p.Tyr1092X; Y1092X)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 1	NSOFUGNALLELE1	NSO DERF Gene Allele 1	c.3276C>G (p.Tyr1092X; Y1092X)	
			c.3454G>C (p.Asp1152His; D1152H)	
			c.3484C>T (p.Arg1162X; R1162X)	
			c.364G>C (p.Glu121Gln; HbD)	
			c.364G>C (p.Glu121Gly; HbD-Punjab; Hb D-Los Angelese)	
			c.3659delC (p.Thr1220LysfsX8)	
			c.367G>A (p.Gly123Arg)	
			c.3717+12191C>T (3849+10kbC>T)	
			c.3731G>A (p.Gly1244Glu; G1244E)	
			c.3773_3774insT (3905insT)	
			c.3808G>A (p.Asp1270Asn; D1270N)	
			c.3909C>G (p.Asn1303Lys; N1303K)	
			c.3937C>T (p.Gln1313X; Q1313X)	
			c.395C>G (p.P132R)	
			c.424G>T (p.A142S)	
			c.427T>C (p.Ter143Glnext32; HbCS; HbConstant Spring)	
			c.46G>C (p.Gly15Arg; HbOttawa)	
			c.470G>A (p.R157H)	
			c.489+1G>T (621+1G>T)	
			c.511G>A (p.A171T)	
			c.513C>G (p.Asp171Glu; p.D171E)	
			c.51delC (p.Lys18Argfs)	
			c.52A>T (p.Lys18Ter)	
			c.554-1G>T (IVS6-1 G>T)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 1	NSOFUGNALLELE1	NSO DERF Gene Allele 1	c.563A>G (p.Gln188Arg; p.Q188R)	
			c.583G>A (p.G195R)	
			c.607-6T>G (IVS7-6 T>G)	
			c.616C>T (p.R206C)	
			c.617T>G (p.Leu206Trp; L206W)	
			c.623-2_623-1del	
			c.744-33GATT[6]+[6] (TTAG repeats)	
			c.744-33GATT[6]+[7] (TTAG repeats)	
			c.776C>T (p.Ala259Val)	
			c.782C>T (p.Pro261Leu; P261L)	
			c.799 G>A (p.Gly267Arg; p.G267R)	
			c.-79A>G (-29A>G)	
			c.79G>A (p.Glu27Lys; HbE)	
			c.842G>C	
			c.848T>C (p.V283A)	
			c.869+11C>T (1001+11C>T)	
			c.889_891del (p.Glu297del; p.E297del))	
			c.91+5G>T (IVS1+5G>T; IVS2+5G>T)	
			c.91G>A (p.Glu31Gln; HbG-Honolulu)	
			c.92+5G>C (IVS1-5 G>C)	
			c.940A>G (p.Asn314Asp; N314D)	
			C.941C>T (p.Ala314Val)	
			c.98_104delGCGGCTGinsTCC (p.Cys33Phefs; c.98_104del7ins3)	
			c.985A>G (p.Lys329Glu; p.K329E)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 1	NSOFUGNALLELE1	NSO DERF Gene Allele 1	Complete deletion	
			deletion exon 2	
			deletion exons 4-11	
			--FIL (g.11684_43534del31851)	
			--MED (g.24664_41064del16401)	
			--SEA (g.26264_45564del19301)	
			--South African (g.209051_232787delins157)	
			--THAI (g.10664_44164del33501)	
			-α3.7 (3.7-kb rightward deletion)	
			-α4.2 (4.2-kb leftward deletion)	
			Other, please specify:	
NSO DERF Gene Allele 1 Other	NSOFUGNALL1OTHE	NSO DERF Gene Allele 1 Other		
NSO DERF Gene Allele 2	NSOFUGNALLELE2		9T	
			7T	
			c.1001A>G (p.Lys334Arg; p.K334R)	
			c.1009_1011del (p.Leu337del)	
			5T	
			c.1016C>T (p.Ser339Leu; p.S339L)	
			c.1010A>C (p.Tyr337Ser; p.Y337S)	
			c.100G>A (p.Gly34Ser; p.G34S)	
			c.1027G>C (p.Gly343Arg; p.G343R)	
			c.1045C>T (p.Arg349Ter; p.R349X)	
			c.1040G>C (p.Arg347Pro; p.R347P)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 2	NSOFUGNALLELE2		c.1046C>T (p.Ala349Val; p.A349V)	
			c.1049 C>A (p.Ser350Tyr; p.S350Y)	
			c.1066-11G>A	
			c.1052delC (p.Thr351Lysfs*12; p.T351Kfs*12)	
			c.105_109del (p.Arg37Alafs*20)	
			c.1068T>A	
			c.1115C>A (p.Ala372Asp; p.A372D)	
			c.1091-2_1092del4ins18	
			c.1135G>A (p.Ala379Thr; p.A379T)	
			c.1148T>A (p.Phe383Tyr; p.F383Y)	
			c.1171C>T (p.Pro391Ser; p.P391S)	
			c.1169A>G (p.Glu390Gly; p.E390G)	
			c.1168G>A (p.Gly390Arg; p.G390R)	
			c.1173C>A (p.Phe391Leu; p.F391L)	
			c.1193C>A (p.Ala398Asp; p.A398D)	
			c.1182+1G>A	
			c.-11G>T	
			c.1208C>T (p.Ala403Val; p.A403V)	
			c.1241A>G (p.Tyr414Cys; p.Y414C)	
			c.1222C>T (p.Arg408Trp; p.R408W)	
			c.1213A>G (p.Met405Val; p.M405V)	
			c.1247T>C (p.Ile416Thr; p.I416T)	
			c.1312T>A (p.Tyr438Asn; p.Y438N)	
			c.1253G>A (p.Ser418Asn; p.S418N)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 2	NSOFUGNALLELE2		c.1315+1G>A	
			c.1322G>A (p.Gly441Asp; p.G441D)	
			c.133G>A (p.Gly45Arg; p.G45R)	
			c.1324G>T (p.Val442Phe; p.V442F)	
			c.-132-329A>G	
			c.1348T>G (p.Trp450Gly; p.W450G)	
			c.1361A>G (p.Tyr454Cys; p.Y454C)	
			c.1360G>A (p.Asp454Asn; p.D454N)	
			c.1363delC (p.Leu455Phefs)	
			c.1367G>A (p.Arg456His; p.R456H)	
			c.-137C>G	
			c.1376G>A (p.Arg459Gln; p.R459Q)	
			c.1375dupC (p.Arg459Profs)	
			c.1388G>A (p.Gly463Glu; p.G463E)	
			c.-138C>T	
			c.138C>T	
			c.1408A>G (p.Met470Val; M470V)	
			c.142G>C (p.Asp48His; Hb Hasharon)	
			c.1451A>G (p.His484Arg; p.H484R)	
			c.1451_1452delGGinsC (p.Arg484Profs)	
			c.143T>C (p.Leu48Ser; p.L48S)	
			c.1475A>C (p.Asp492Ala; p.D492A)	
			c.1489C>T (p.Pro497Ser; p.P497S)	
			c.1475C>T (p.Thr492Ile; p.T492I)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 2	NSOFUGNALLELE2		c.1500_1502del (p.Leu502del)	
			c.151C>T (p.His51Tyr; p.H51Y)	
			c.157G>A (p.Asp53Asn; Hb Osu Christiansborg)	
			c.157C>T (p.Arg53Cys; p.R53C)	
			c.1528G>C (p.Glu510Gln; p.E510Q)	
			c.1584G>A (p.Glu528Glu; p.E528E)	
			c.1612C>T (p.Arg538Cys; p.R538C)	
			c.1603C>G (p.Leu535Val; p.L535V)	
			c.1616A>G (p.Gln539Arg; p.Q539R)	
			c.1627delG	
			c.1678+1G>A	
			c.165del (c.165delT)	
			c.163_167delTTTTTinsAA	
			c.17_18del	
			c.1700G>A (p.Arg567Gln; p.R567Q)	
			c.17_del	
			c.-170G>A (Sardinian HPFH)	
			c.1717-1G>A	
			c.1741G>T (p.Val581Phe; p.V581F)	
			c.1727G>C (p.Gly576Ala; p.G576A)	
			c.1717-1G>T	
			c.1793A>G (p.His598Arg; p.H598R)	
			c.1844G>A (p.Arg615Gln; p.R615Q)	
			c.1843C>T (p.Arg615Ter)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 2	NSOFUGNALLELE2		c.1868G>T (p.Gly623Val; p.G623V)	
			c.194T>C (p.Ile65Thr; p.I65T)	
			c.1A>G (p.Met1Val; p.M1V)	
			c.19G>A (p.Asp7Asn; Hb Dunn)	
			c.199-10T>G	
			c.2002C>T (p.Arg668Cys; p.R668C)	
			c.2015_2052delAAinsG (p.Lys684SerfsX38; 2183AA>G)	
			c.200C>T (p.Thr67Met; p.T67M)	
			SNP rs77680021	
			c.847_850delCTGG	
			SNP rs8064573	
			SNP rs3817641 (benign)	
			SNP rs41274865 (benign)	
			SNP rs6145976 (benign)	
			SNP rs60400822 (benign)	
			SNP rs41274867 (benign)	
			c.971G>T (p.Gly324Val; p.G324V)	
			c.990dupT (p.Glu331Terfs)	
			c.977T>C (p.Val326Ala; p.V326A)	
			c.991T>C (p.Cys331Arg; p.C331R)	
			c.996+17G>A	
			SNP rs140871321	
			SNP rs12022011 (intron variant)	
			SNP rs11316888 (intron variant)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 2	NSOFUGNALLELE2		SNP rs150422765 (intron variant)	
			SNP rs2277202 (benign)	
			SNP rs17671352 (benign)	
			c.896A>T (p.Lys299Met; p.K299M)	
			c.896C>T (p.Ala299Val; p.A299V)	
			c.916A>G (p.Ile306Val; p.I306V)	
			c.898G>T (p.Ala300Ser; p.A300S)	
			c.896T>G (p.Phe299Cys; p.F299C)	
			c.92+1G>T	
			c.925G>A (p.Gly309Arg; p.G309R)	
			c.923dupT (p.Leu308Phefs)	
			c.92C>T (p.Pro31Leu; p.P31L)	
			c.92G>C (p.Arg31Thr; Hb Kairouan)	
			c.939A>G (p.Ile313Met; p.I313M)	
			c.937C>T (p.Arg313Trp; p.R313W)	
			c.93-21G>C	
			c.945+1G>C	
			c.948delT (1078delT)	
			c.946-4C>G	
			c.950T>C (p.Val317Ala; p.V317A)	
			c.955C>T (p.Gln319Ter)	
			c.970C>T (p.Arg324Ter)	
			c.958T>G (p.Leu320Val; p.L320V)	
			c.956C>T (p.Ser319Phe; p.S319F)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 2	NSOFUGNALLELE2		c.629A>G (p.Tyr210Cys; p.Y210C)	
			c.632G>A (p.Arg211His; p.R211H)	
			c.631C>T (p.Arg211Cys; p.R211C)	
			c.638T>C (p.Leu213Pro; p.L213P)	
			c.641A>C (p.Asn214Thr; p.N214T)	
			c.645C>T (p.Leu215Leu; p.L215L)	
			c.641T>A (p.Ile214Lys; p.I214K)	
			c.641A>G (p.Asn214Ser; p.N214S)	
			c.64C>T (p.Pro22Ser; p.P22S)	
			c.656G>A (p.Arg219His; p.R219H)	
			c.652C>T (p.Leu218Leu; p.L218L)	
			c.658_660delAAG (p.Lys220del)	
			c.680A>T (p.Asp227Val; p.D227V)	
			c.688-2A>G	
			c.683A>G (p.Asn228Ser; p.N228S)	
			c.682G>T (p.Asp228Tyr; D228Y)	
			c.701C>T (p.Thr234Ile; p.T234I)	
			c.709G>A (p.Ala237Thr; p.A237T)	
			c.701G>A (p.Arg234Gln; p.R234Q)	
			c.710T>A (p.Ile237Asn; p.I237N)	
			c.713T>A (p.Val238Glu; p.V238E)	
			c.728G>A (p.Arg243Gln; p.R243Q)	
			c.722G>A (p.Arg241His; p.R241H)	
			c.719T>A (p.Met240Lys; p.M240K)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 2	NSOFUGNALLELE2		c.734T>C (p.Val245Ala; p.V245A)	
			c.753-23_753-22del	
			c.744-33GATT[7]+[7]	
			c.753-2A>C	
			c.757G>A (p.Asp253Asn; p.D253N)	
			c.782G>A (p.Arg261Gln; R261Q)	
			c.779C>T (p.Thr260Met; p.T260M)	
			c.775C>T (p.Arg259Trp; p.R259W)	
			c.787G>A (p.Val263Met; p.V263M)	
			c.8591A>C (p.His2864Pro; p.H2864P)	
			c.855G>T (p.Lys285Asn; p.K285N)	
			c.860G>A (p.Gly287Asp; p.G287D)	
			c.865G>A (p.Gly289Arg; p.G289R)	
			c.887_888delCT (p.Pro296Argfs)	
			c.881G>A (p.Gly294Glu; p.G294E)	
			c.877G>A (p.Ala293Thr; p.A293T)	
			c.887G>A (p.Arg296Gln; p.R296Q)	
			c.896_898delAGA (p.Lys299del)	
			c.895C>T (p.Arg299Cys; p.R299C)	
			c.209C>T (p.Ser70Leu; p.S70L)	
			c.2128A>T (p.Lys710Ter)	
			c.224G>A (p.Arg75Gln; R75Q)	
			c.223C>T (p.Arg75Ter)	
			c.216+2T>C	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 2	NSOFUGNALLELE2		c.2260G>A (p.Val754Met; V754M)	
			c.227T>C (p.Leu76Pro; Hb Atlanta)	
			c.227T>C (p.Ile76Thr; Hb F Waynesboro)	
			c.233T>C (p.Ile78Thr; p.I78T)	
			c.236G>A (p.Arg79His; p.R79H)	
			c.2562T>G (p.Thr854Thr; 2694T>G)	
			c.253G>T (p.Gly85Cys; p.G85C)	
			c.2490+1G>A (2622+1G>A)	
			c.2604A>G	
			c.276G>T (p.Glu92Asp; p.E92D)	
			c.262_264delGAG (p.Glu88del; p.E88del)	
			c.281 A>G (p.Asp94Gly; p.D94G)	
			c.283G>A (p.Asp95Asn; Hb F Columbus-GA)	
			c.293-13C>G	
			c.290A>T (p. Asp97Val; p.D97V)	
			c.287G>T (p.Gly96Val; p.G96V)	
			c.3067_3072delTAGTG (p.Ile1023Serfs)	
			c.316-2A>G	
			c.315+1G>A	
			c.3196C>T (p.Arg1066Cys; p.R1066C)	
			c.3197G>A (p.Arg1066His; p.R1066H)	
			c.332_339delGAGACTAC (p.Gly111Valfs)	
			c.331C>T (p.Arg111Ter)	
			c.3222T>A (p.Phe1074Leu; p.F1074L)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 2	NSOFUGNALLELE2		c.3486_3487delAG	
			c.34G>A (p.Ala12Thr; p.A12T)	
			c.349C>T (p.Arg117Cys; p.R117C)	
			c.362C>T (p.Thr121Ile; p.T121I)	
			c.366T>G (p.Asn122Lys; p.D122K)	
			c.3794G>T (p.Gly1265Val; p.G1265V)	
			c.377G>A (p.Gly126Asp; p.G126D)	
			c.372T>A (p.Asp142Glu; p.D142E)	
			c.380C>T (p.Pro127Leu; p.P127L)	
			c.383_385del (p.Val128del)	
			c.382_384delTAC (p.Tyr128del)	
			c.384-20A>G	
			c.3846G>A (p.Trp1282Ter; W1282X)	
			c.3889dupT(p.Ser1297Phefs*5)	
			c.3879delA	
			c.387+91dupA	
			c.3905T>C (p.Met1302Thr; p.M1302T)	
			c.397G>T (p.Val133Leu; p.V133L)	
			c.394C>T (p.Arg132Stop; p.R132X)	
			c.-41A>C	
			c.4272C>T (p.Tyr1424Tyr; p.Y1424Y)	
			c.436C>T (p.Arg146Trp; pR146W)	
			c.42C>A	
			c.428A>G (p.Asp143Gly; p.D143G)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 2	NSOFUGNALLELE2		c.437delT (p.Leu146Trpfs)	
			c.43G>T (p.Gly15Trp; p.G15W)	
			c.4389G>A (p.Gln1463Gln; p.Q1463Q)	
			c.44 T>C (p.Val5Ala; p.V5A)	
			c.442delA (p.Ile148Leufs)	
			c.443G>A (p.Arg148Lys; p.R148K)	
			c.443C>T (p.Thr148Ile; p.T148I)	
			c.443A>G (p.Gln148Arg; p.Q148R)	
			c.443T>C (p.Ile148Asn; p.I148N)	
			c.470G>A (p.Arg157His; p.R157H)	
			c.456G>A (p.Trp152Ter)	
			c.473G>A (p.Arg158Gln; p.R158Q)	
			c.507delG (p.Glu169AspfsX11)	
			c.518T>A (p.Ile173Asn; p.I173N)	
			c.511G>A (p.Ala171Thr; p.A171T)	
			c.50G>A (p.Gly17Asp; Hb J-Baltimore)	
			c.51C>G (p.Phe17Leu; p.F17L)	
			c.535G>T (p.Gly179Trp; p.G179W)	
			c.51dupT (p.Ala5CysfsX5)	
			c.547C>T (p.Arg183Trp; p.R183W)	
			c.556C>T (p.Arg186Trp; p.R186W)	
			c.56_57delGCinsAA (p.Arg19Gln; p.R19Q)	
			c.557G>A (p.Cys186Tyr; p.C186Y)	
			c.556C>T (p.His186Tyr; p.H186Y)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 2	NSOFUGNALLELE2		c.568C>T (p.His190Tyr; p.H190Y)	
			c.575T>G (p.Phe192Cys; p.F192C)	
			c.572T>C (p.Met191Thr; p.M191T)	
			c.577C>T (p.Arg193Trp; p.R193W)	
			c.579+1G>T (711+1G>T)	
			c.622C>T (p.Arg208Ter)	
			c.609G>A (p.Trp203Ter)	
			c.604T>C (p.Phe202Leu; p.F202L)	
			619 bp deletion (g.71609_72227del619)	
			c.1000C>T (p.Arg334Trp; R334W)	
			c.1040G>A (p.Arg347His; R347H)	
			c.1062+5G>A (IVS12+5 G>A)	
			c.-119_-116delGTCA (5'UTR-119delGTCA)	
			c.1193C>T (p.P398L)	
			c.126_129delCTTT (p.Phe42fs)	
			c.1316G>A; (p.Gly439Asp; p.G439D)	
			c.1327_1330dup (p.Ile444Argfs)	
			c.1330G>C (p.D444H)	
			c.1364C>A (p.Ala455Glu; A455E)	
			c.1367T>C (p.Val456Ala)	
			c.1368A>C (p.Q456H)	
			c.1413T>C (p.C471C)	
			c.1436C>T (p.P479L; p.Pro479Leu)	
			c.1463G>A (p.R488H)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 2	NSOFUGNALLELE2		c.1466C>A (p.Ser489X; S489X)	
			c.1508_1512delCCATG	
			c.1519_1521delATC (p.Ile507del; I507del; DI507)	
			c.1521_1523delCTT (p.Phe508del; F508del; dF508)	
			c.1523T>G (p.Phe508Cys; F508C)	
			c.1558G>T (p.Val520Phe; V520F)	
			c.1595C>T (p.T532M)	
			c.1616C>A	
			c.1624G>T (p.Gly542X; G542X)	
			c.1630G>A (p.Gly544Ser)	
			c.1646G>A (p.Ser549Asn; S549N)	
			c.1652G>A (p.Gly551Asp; G551D)	
			c.1657C>T (p.Arg553X; R553X)	
			c.1675G>A (p.Ala559Thr; A559T)	
			c.1679G>C (p.Arg560Thr; R560T)	
			c.1705T>G (p.Tyr569Asp; Y569D)	
			c.1766+1G>A (1898+1G>A)	
			c.178G>T (p.Glu60X; E60X)	
			c.190A>G (p.Met64Lys)	
			c.192G>T (p.Gln64His; Q64H)	
			c.199 T>C (p.Tyr67His; p.Y67H))	
			c.19G>A (HbC; p.Glu7Lys)	
			c.200C>T (p.Pro67Leu; P67L)	
			c.20A>T (HbS; p.Glu7Val)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 2	NSOFUGNALLELE2		c.216+1G>A	
			c.2162T>A (p.Val721Glu)	
			c.220G>A (p.Asp74Asn; Hb G-Accra; Korle-Bu variant of Hb)	
			c.223G>C (p.Asp74Gly; HbQ-Thailand)	
			c.2246G>T (p.R749L; p.Arg749Leu)	
			c.233C>A (p.Pro78His; HbToulon)	
			c.237C>A (p.Asn79Lys; HbStanleyville-II)	
			c.254G>A (p.Gly85Glu; G85E)	
			c.262_263delTT (p.Leu88IlefsX22; 394delTT)	
			c.263T>A (p.Leu88X)	
			c.2657+2_2657+3insA (2789+2insA)	
			c.2657+5G>A (2789+5G>A)	
			c.271dupA (p.Arg91Lysfs)	
			c.2734T>A (p.Ser912Thr)	
			c.278-41del	
			c.27dupG (c.27_28insG; p.Ser10Valfs*14)	
			c.2988+1G>A (3120+1G>A)	
			c.305G>A (p.Arg117His; R117H)	
			c.307A>C (p.Ser103Arg; HbManitoba I)	
			c.3080T>C (p.Ile1027Thr; I1027T)	
			c.3276C>A (p.Tyr1092X; Y1092X)	
			c.3276C>G (p.Tyr1092X; Y1092X)	
			c.3454G>C (p.Asp1152His; D1152H)	
			c.3484C>T (p.Arg1162X; R1162X)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 2	NSOFUGNALLELE2		c.364G>C (p.Glu121Gln; HbD)	
			c.364G>C (p.Glu121Gly; HbD-Punjab; Hb D-Los Angelese)	
			c.3659delC (p.Thr1220LysfsX8)	
			c.367G>A (p.Gly123Arg)	
			c.3717+12191C>T (3849+10kbC>T)	
			c.3731G>A (p.Gly1244Glu; G1244E)	
			c.3773_3774insT (3905insT)	
			c.3808G>A (p.Asp1270Asn; D1270N)	
			c.3909C>G (p.Asn1303Lys; N1303K)	
			c.3937C>T (p.Gln1313X; Q1313X)	
			c.395C>G (p.P132R)	
			c.424G>T (p.A142S)	
			c.427T>C (p.Ter143Glnext32; HbCS; HbConstant Spring)	
			c.46G>C (p.Gly15Arg; HbOttawa)	
			c.470G>A (p.R157H)	
			c.489+1G>T (621+1G>T)	
			c.511G>A (p.A171T)	
			c.513C>G (p.Asp171Glu; p.D171E)	
			c.51delC (p.Lys18Argfs)	
			c.52A>T (p.Lys18Ter)	
			c.554-1G>T (IVS6-1 G>T)	
			c.563A>G (p.Gln188Arg; p.Q188R)	
			c.583G>A (p.G195R)	
			c.607-6T>G (IVS7-6 T>G)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 2	NSOFUGNALLELE2		c.616C>T (p.R206C)	
			c.617T>G (p.Leu206Trp; L206W)	
			c.623-2_623-1del	
			c.744-33GATT[6]+[6] (TTAG repeats)	
			c.744-33GATT[6]+[7] (TTAG repeats)	
			c.776C>T (p.Ala259Val)	
			c.782C>T (p.Pro261Leu; P261L)	
			c.799 G>A (p.Gly267Arg; p.G267R)	
			c.-79A>G (-29A>G)	
			c.79G>A (p.Glu27Lys; HbE)	
			c.842G>C	
			c.848T>C (p.V283A)	
			c.869+11C>T (1001+11C>T)	
			c.889_891del (p.Glu297del; p.E297del))	
			c.91+5G>T (IVS1+5G>T; IVS2+5G>T)	
			c.91G>A (p.Glu31Gln; HbG-Honolulu)	
			c.92+5G>C (IVS1-5 G>C)	
			c.940A>G (p.Asn314Asp; N314D)	
			C.941C>T (p.Ala314Val)	
			c.98_104delGCGGCTGinsTCC (p.Cys33Phefs; c.98_104del7ins3)	
			c.985A>G (p.Lys329Glu; p.K329E)	
			Complete deletion	
			deletion exon 2	
			deletion exons 4-11	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO DERF Gene Allele 2	NSOFUGNALLELE2		--FIL (g.11684_43534del31851)	
			--MED (g.24664_41064del16401)	
			--SEA (g.26264_45564del19301)	
			--South African (g.209051_232787delins157)	
			--THAI (g.10664_44164del33501)	
			-α3.7 (3.7-kb rightward deletion)	
			-α4.2 (4.2-kb leftward deletion)	
			Other, please specify:	
NSO DERF Gene Allele 2 Other	NSOFUGNALL2OTHE	NSO DERF Gene Allele 2 Other		
NSO DERF Ultrasound	NSODERFUS	Imaging results of the gonads	Testes	
			Ovaries	
NSO DERF Was the result a true positive	NDERFRESPOS11		Other	
			Transfer of Care	
NSO DERF Were mutations / polymorphism/deletions/ duplications detected?	NDERFMUTPOLY	NSO DERF Were mutations / polymorphism/deletions/duplications detected?		
NSO Diagnosis Made Prenatally Flag	NBS0142	Indicates if the diagnosis was made prenatally or postnatally.		
NSO Follow-Up Additional Review Questions	NSOFUADDQ	NSO Follow-Up Additional Review Questions		
NSO Gaining Weight	NBS0121	An indication of whether an infant with a positive newborn screen was gaining weight at the time of retrieval for diagnostic evaluation		
NSO Gaining Weight Comment	NSOSTFU003	Comments regarding whether an infant with a positive newborn screen was gaining weight at the time of retrieval for diagnostic evaluation		

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO Maternal Steroids	NBS0126	An indication of whether the mother of an infant with a newborn screen positive for congenital adrenal hyperplasia was given steroids at any time during her pregnancy, noted at the time of retrieval for diagnostic evaluation		
NSO Meconium Ileus (CF)	NBS0123	An indication of whether the infant with a newborn screen positive for cystic fibrosis was born with meconium ileus, noted at the time of retrieval for diagnostic evaluation		
NSO Missed Case Maternal Name and OHIP Note	NDERFMAROHIP	The name and Ontario Health Card Number (if available) of the mother whose infant is affected with a disease on the NSO panel and was not diagnosed via newborn screening. Enter Unknown, CAS or adoption if applicable		
NSO Review Comments	NSODERF935			
NSO Reviewed By	NSODERF932		Dr. Pranesh Chakraborty	
			Dr. Michael Geraghty	
			Other (specify)	
NSO Reviewed By Other	NSODERF933			
NSO STFU Follow-Up Diagnostic Analyte Ref Range Lower	NBS0136	The lower limit of the reference range for the diagnostic analyte or profile analytes		
NSO STFU Follow-Up Diagnostic Analyte Ref Range Upper	NBS0135	The upper limit of the reference range for the diagnostic analyte or profile analytes		
NSO STFU Follow-Up Diagnostic Analyte Unit of Measure	NBS0134	The unit of measure of the diagnostic analyte or profile analytes		
NSO STFU Follow-Up Profile Comment	NBS0138	Comments regarding the diagnostic investigations and / or evaluations		
NSO STFU Follow-Up Profile Date	NBS0137	The date on which the diagnostic evaluations and investigations were performed		
NSO STFU Follow-Up Profile Diagnostic Analyte Name	NSOSTFU012	The name(s) of the analyte(s) measured in the diagnostic evaluation of the infant / child	Phenylalanine, Plasma	
			Tyrosine, Plasma	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO STFU Follow-Up Profile Diagnostic Analyte Name	NSOSTFU012	The name(s) of the analyte(s) measured in the diagnostic evaluation of the infant / child	Succinylacetone, urine	
			DHPR assay	
			Succinylacetone, dried blood spot	
			Citrulline, Plasma	
			C2 acylcarnitine, Plasma	
			AFP	
			C14 acylcarnitine, Plasma	
			C14:1 acylcarnitine, Plasma	
			C16 acylcarnitine, Plasma	
			C16:1 acylcarnitine, Plasma	
			CBC	
			Glutaric Acid, Urine	
			3-OH Glutaric Acid, Urine	
			C5DC, Urine	
			C5DC acylcarnitine, Plasma	
			C18 acylcarnitine, Plasma	
			C6 acylcarnitine, Plasma	
			Gal-1-P Uridyl Transferase, RBC	
			C8 acylcarnitine, Plasma	
			Galactose, RBC	
			C10 acylcarnitine, Plasma	
			Galactose-1-Phosphate, RBC	
			Galactitol, Urine	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO STFU Follow-Up Profile Diagnostic Analyte Name	NSOSTFU012	The name(s) of the analyte(s) measured in the diagnostic evaluation of the infant / child	C18:2 acylcarnitine, Plasma	
			C14:1OH acylcarnitine, Plasma	
			C14OH acylcarnitine, Plasma	
			C16:1OH acylcarnitine, Plasma	
			C16OH acylcarnitine, Plasma	
			C18:1 acylcarnitine, Plasma	
			C18:1OH acylcarnitine, Plasma	
			C18OH acylcarnitine, Plasma	
			C5 acylcarnitine, Plasma	
			Total Carnitine, Plasma Newborn	
			ASA, Plasma	
			Free Carnitine, Plasma Newborn	
			Creatinine, Plasma Newborn	
			Total Carnitine, Urine Newborn	
			Free Carnitine, Urine Newborn	
			Creatinine, Urine Newborn	
			Leucine	
			Specify FE Carnitine or TRR% and value	
			Total Carnitine, Plasma Mother	
			Free Carnitine, Plasma Mother	
			Creatinine, Plasma Mother	
			CK	
			Total Carnitine, Urine Mother	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO STFU Follow-Up Profile Diagnostic Analyte Name	NSOSTFU012	The name(s) of the analyte(s) measured in the diagnostic evaluation of the infant / child	Free Carnitine, Urine Mother	
			Creatinine, Urine Mother	
			Arginine, Plasma	
			Valine	
			Ornithine, Plasma	
			Isoleucine	
			Alanine, Plasma	
			C5OH acylcarnitine, Plasma	
			C5:1 acylcarnitine, Plasma	
			C6DC acylcarnitine, Plasma	
			Allo-Isoleucine	
			Sweat Chloride	
			Fecal elastase	
			72 hour fecal fat	
			TSH	
			FT4	
			TRAb	
			Maternal Antibodies	
			Thyroid Scan/Ultrasound (size)	
			Glutamine, Plasma	
			Ammonium, Plasma	
			Orotic Acid, Urine	
			ALT	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO STFU Follow-Up Profile Diagnostic Analyte Name	NSOSTFU012	The name(s) of the analyte(s) measured in the diagnostic evaluation of the infant / child	C3 acylcarnitine, Plasma	
			C4DC acylcarnitine, Plasma	
			Total Homocysteine, Plasma Newborn	
			Serum B12, Infant	
			Methionine, Plasma	
			Molecular result	
			Bilirubin, Serum	
			AST	
			GGT	
			Mutation 1	
			Bilirubin Total	
			17-OH Progesterone	
			Mutation 2	
			Bilirubin Direct	
			Total Homocysteine, Maternal Plasma	
			MMA, Maternal plasma	
			Serum B12, Maternal	
			Na	
			K	
			INR	
			PTT	
			Chloride	
			DHEAS	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO STFU Follow-Up Profile Diagnostic Analyte Name	NSOSTFU012	The name(s) of the analyte(s) measured in the diagnostic evaluation of the infant / child	Ultrasound (CAH)	
			Cortisol	
			Renin	
			HCO3	Venous blood gases - HCO3
			pH	Venous blood gases pH
			pCO2	Venous blood gases pCO2
			Biotinidase, Plasma	
			Glucose	
			Other	
			%Hgb A2	
			%Hgb A	
			%Hgb F	
			%Hgb S	
			%Hgb C	
			%Hgb D	
			%Hgb E	
			%Hgb Other	
			CD3	
			CD4	
			CD8	
			CD19	
			CD20	
			CD16 / CD56 / CD3neg	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO STFU Follow-Up Profile Diagnostic Analyte Name	NSOSTFU012	The name(s) of the analyte(s) measured in the diagnostic evaluation of the infant / child	Glutaric Acid	
			Isobutyrylglycine	
			N-Butyrylglycine	
			2-Methylbutyrylglycine	
			Isovalerylglycine	
			N-Hexanoylglycine	
			N-Octanoylglycine	
			3-Phenylpropionylglycine	
			Suberylglycine	
			Trans-Cinnamoylglycine	
			Methylcrotonylglycine	
			Tetrahydrobiopterin	
			Neopterin:Biopterin	
			Neopterin	
			Biopterin	
			ACTH at 0 min	
			ACTH at 30 min	
			ACTH at 60 min	
			Glutamine	
			Sarcosine	
			Alpha Aminoadipic Acid	
			Glycine	
			Alanine	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO STFU Follow-Up Profile Diagnostic Analyte Name	NSOSTFU012	The name(s) of the analyte(s) measured in the diagnostic evaluation of the infant / child	Citrulline	
			Amino-n-Butyric Acid	
			Cystine	
			Methionine	
			Phosphoserine	
			Cystathionine	
			Tyrosine	
			Phenylalanine	
			Beta-Alanine	
			Amino-Iso-Butyric acid	
			Aminobutyric Acid	
			Homocystine	
			Ethanolamine	
			Taurine	
			Tryptophan	
			Hydroxylysine / allo-Hydroxylysine	
			Ornithine	
			Lysine	
			1-Methylhistidine	
			Histidine	
			3-Methylhistidine	
			Anserine	
			Phosphoethanolamine	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO STFU Follow-Up Profile Diagnostic Analyte Name	NSOSTFU012	The name(s) of the analyte(s) measured in the diagnostic evaluation of the infant / child	Carnosine	
			Arginine	
			Hydroxyproline	
			Proline	
			Asparagine	
			Argininosuccinic Acid	
			Aspartic Acid	
			Threonine	
			Serine	
			Glutamic Acid	
			Homocitrulline	
			WBC	
			RBC	
			Hgb	
			Hct	
			MCV	
			MCH	
			MCHC	
			RDW	
			Platelets	
			MPV	
			Neutro Bands	
			Neutro Polys	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO STFU Follow-Up Profile Diagnostic Analyte Name	NSOSTFU012	The name(s) of the analyte(s) measured in the diagnostic evaluation of the infant / child	Lymphocytes	
			Monocytes	
			Eosinophils	
			Basophils	
			Whole blood TREC	
			Repeat NSO TREC on dried blood spot	
			ADA activity	
			PNP activity	
			Adenosine	
			Deoxyadenosine	
			Guanosine	
			Deoxyguanosine	
			DBS 22q deletion	
			22q deletion	
			CF Mutation 1	
			CF Mutation 2	
			PolyT tract (T/T)	
			Polymorphism(s)	
			CF MUT Other 1	
			CF MUT Other 2	
			C2	
			C3:1	
			C3	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO STFU Follow-Up Profile Diagnostic Analyte Name	NSOSTFU012	The name(s) of the analyte(s) measured in the diagnostic evaluation of the infant / child	C4	
			C5:1	
			C5	
			C4OH	
			C6	
			C5OH	
			Benzoyl	
			C6OH	
			Phenylacetyl	
			C8:1	
			C8	
			C3DC	
			C10:3	
			C10:2	
			C10:1	
			C10	
			C4DC	
			C5DC	
			C12:1	
			C12	
			C6:DC	
			C12-OH	
			C14:2	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO STFU Follow-Up Profile Diagnostic Analyte Name	NSOSTFU012	The name(s) of the analyte(s) measured in the diagnostic evaluation of the infant / child	C14:1	
			C14	
			C8:DC	
			C14:1OH	
			C14OH	
			C16:1	
			C16	
			C10:DC	
			C16:1OH	
			C16OH	
			C18:2	
			C18:1	
			C18	
			C18:2-OH	
			C18:1-OH	
			C18-OH	
			C16:DC	
			C18:1:DC	
			Gene	
			Thyroid Scan (location)	Thyroid Scan (location)
			Thyroid Scan (uptake)	
NSO STFU Follow-Up Profile Diagnostic Analyte Value	NSOSTFU013	The numeric or alpha numeric value (result) of the analyte measured in the diagnostic evaluation of the infant / child		

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO STFU Follow-Up Profile Name	NSOSTFU017		Additional Diagnostic Investigations	
			Hemoglobin percentages	
			Immunophenotyping	
			Acylglycine profile	
			Urinary pterin analysis	
			Adrenocorticotropic hormone stimulation test	
			Amino acid profile	
			CBC (infant)	
			CBC (maternal)	
			SCID investigations	
			22q11 deletion results	
			Repeat NSO purine profile on dried blood spot	
			Purine profile	
			CFTR molecular results	
			Acylcarnitine profile (infant)	
			Acylcarnitine profile (mother)	
			Molecular results (in standard nomenclature)	
			Organic acid profile (newborn/patient)	
			Beutler test results	
			Hemoglobin pattern	
			Hemoglobin pattern determined by	
			Hemoglobin H bodies	
			Organic acid profile (maternal)	
			Biopterin loading test	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO STFU Follow-Up Profile Name	NSOSTFU017		Acylcarnitine probe results	
			Enzymology probe results	
			Palmitoyl-CoA-oxidation studies	
			Venous blood gases	
NSO STFU Follow-Up Profile Profile Type	NSOSTFU0020	Not available to end user. Used to define types of profile for UI rendering		
NSO STFU Follow-Up Profile Result	NSOSTFU014	The result of the profile performed in the diagnostic evaluation of the infant / child.	Abnormal	
			Absent	
			Negative	
			Normal	
			Positive	
			Present	
			Hgb Electrophoresis	
			Hgb Isoelectric Focusing	
			Hgb HPLC	
NSO STFU Follow-Up Profile Result Test	NSOSTFU014TXT			
NSO Treatment Initiated - Other (CCPID)	NBS0141CCPID	Other treatment initiated on the infant after the diagnosis (CCPID)		
NSO Treatment Initiated - Other (RTC)	NBS0141	Other treatment initiated on the infant after the diagnosis.		
NSO Treatment Initiated (CCPID)	NBS0140CCPID	Treatment initiated on the infant after the diagnosis (CCPID)	BMT	
			PEG-ADA	
			Gene Therapy	
			Other, specify:	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSO Treatment Initiated (RTC)	NBS0140	Treatment initiated on the infant after the diagnosis (RTC)	BMT	
			PEG-ADA	
			Gene Therapy	
			Other, specify:	
NSODERF Gene Variant	NDERFGENEVAR	NSODERF Gene Variant	5T	
			7T	
			9T	
			c.1001A>G (p.Lys334Arg; p.K334R)	
			c.1009_1011del (p.Leu337del)	
			c.100G>A (p.Gly34Ser; p.G34S)	
			c.1010A>C (p.Tyr337Ser; p.Y337S)	
			c.1016C>T (p.Ser339Leu; p.S339L)	
			c.1027G>C (p.Gly343Arg; p.G343R)	
			c.1040G>C (p.Arg347Pro; p.R347P)	
			c.1045C>T (p.Arg349Ter; p.R349X)	
			c.1046C>T (p.Ala349Val; p.A349V)	
			c.1049 C>A (p.Ser350Tyr; p.S350Y)	
			c.105_109del (p.Arg37Alafs*20)	
			c.1052delC (p.Thr351Lysfs*12; p.T351Kfs*12)	
			c.1066-11G>A	
			c.1068T>A	
			c.1091-2_1092del4ins18	
			c.1115C>A (p.Ala372Asp; p.A372D)	
			c.1135G>A (p.Ala379Thr; p.A379T)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSODERF Gene Variant	NDERFGENEVAR	NSODERF Gene Variant	c.1148T>A (p.Phe383Tyr; p.F383Y)	
			c.1168G>A (p.Gly390Arg; p.G390R)	
			c.1169A>G (p.Glu390Gly; p.E390G)	
			c.1171C>T (p.Pro391Ser; p.P391S)	
			c.1173C>A (p.Phe391Leu; p.F391L)	
			c.1182+1G>A	
			c.1193C>A (p.Ala398Asp; p.A398D)	
			c.-11G>T	
			c.1208C>T (p.Ala403Val; p.A403V)	
			c.1213A>G (p.Met405Val; p.M405V)	
			c.1222C>T (p.Arg408Trp; p.R408W)	
			c.1241A>G (p.Tyr414Cys; p.Y414C)	
			c.1247T>C (p.Ile416Thr; p.I416T)	
			c.1253G>A (p.Ser418Asn; p.S418N)	
			c.1312T>A (p.Tyr438Asn; p.Y438N)	
			c.1315+1G>A	
			c.1322G>A (p.Gly441Asp; p.G441D)	
			c.-132-329A>G	
			c.1324G>T (p.Val442Phe; p.V442F)	
			c.133G>A (p.Gly45Arg; p.G45R)	
			c.1348T>G (p.Trp450Gly; p.W450G)	
			c.1360G>A (p.Asp454Asn; p.D454N)	
			c.1361A>G (p.Tyr454Cys; p.Y454C)	
			c.1363delC (p.Leu455Phefs)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSODERF Gene Variant	NDERFGENEVAR	NSODERF Gene Variant	c.1367G>A (p.Arg456His; p.R456H)	
			c.1375dupC (p.Arg459Profs)	
			c.1376G>A (p.Arg459Gln; p.R459Q)	
			c.-137C>G	
			c.1388G>A (p.Gly463Glu; p.G463E)	
			c.138C>T	
			c.-138C>T	
			c.1408A>G (p.Met470Val; M470V)	
			c.142G>C (p.Asp48His; Hb Hasharon)	
			c.143T>C (p.Leu48Ser; p.L48S)	
			c.1451_1452delGGinsC (p.Arg484Profs)	
			c.1451A>G (p.His484Arg; p.H484R)	
			c.1475A>C (p.Asp492Ala; p.D492A)	
			c.1475C>T (p.Thr492Ile; p.T492I)	
			c.1489C>T (p.Pro497Ser; p.P497S)	
			c.1500_1502del (p.Leu502del)	
			c.151C>T (p.His51Tyr; p.H51Y)	
			c.1528G>C (p.Glu510Gln; p.E510Q)	
			c.157C>T (p.Arg53Cys; p.R53C)	
			c.157G>A (p.Asp53Asn; Hb Osu Christiansborg)	
			c.1584G>A (p.Glu528Glu; p.E528E)	
			c.1603C>G (p.Leu535Val; p.L535V)	
			c.1612C>T (p.Arg538Cys; p.R538C)	
			c.1616A>G (p.Gln539Arg; p.Q539R)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSODERF Gene Variant	NDERFGENEVAR	NSODERF Gene Variant	c.1627delG	
			c.163_167delTTTTTinsAA	
			c.165del (c.165delT)	
			c.1678+1G>A	
			c.17_18del	
			c.17_del	
			c.1700G>A (p.Arg567Gln; p.R567Q)	
			c.-170G>A (Sardinian HPFH)	
			c.1717-1G>A	
			c.1717-1G>T	
			c.1727G>C (p.Gly576Ala; p.G576A)	
			c.1741G>T (p.Val581Phe; p.V581F)	
			c.1793A>G (p.His598Arg; p.H598R)	
			c.1843C>T (p.Arg615Ter)	
			c.1844G>A (p.Arg615Gln; p.R615Q)	
			c.1868G>T (p.Gly623Val; p.G623V)	
			c.194T>C (p.Ile65Thr; p.I65T)	
			c.199-10T>G	
			c.19G>A (p.Asp7Asn; Hb Dunn)	
			c.1A>G (p.Met1Val; p.M1V)	
			c.2002C>T (p.Arg668Cys; p.R668C)	
			c.200C>T (p.Thr67Met; p.T67M)	
			c.2015_2052delAAinsG (p.Lys684SerfsX38; 2183AA>G)	
			SNP rs77680021	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSODERF Gene Variant	NDERFGENEVAR	NSODERF Gene Variant	SNP rs8064573	
			c.847_850delCTGG	
			SNP rs3817641 (benign)	
			SNP rs41274865 (benign)	
			SNP rs41274867 (benign)	
			SNP rs60400822 (benign)	
			SNP rs6145976 (benign)	
			c.971G>T (p.Gly324Val; p.G324V)	
			c.977T>C (p.Val326Ala; p.V326A)	
			c.990dupT (p.Glu331Terfs)	
			c.991T>C (p.Cys331Arg; p.C331R)	
			c.996+17G>A	
			SNP rs11316888 (intron variant)	
			SNP rs12022011 (intron variant)	
			SNP rs140871321	
			SNP rs150422765 (intron variant)	
			SNP rs17671352 (benign)	
			SNP rs2277202 (benign)	
			c.896A>T (p.Lys299Met; p.K299M)	
			c.896C>T (p.Ala299Val; p.A299V)	
			c.896T>G (p.Phe299Cys; p.F299C)	
			c.898G>T (p.Ala300Ser; p.A300S)	
			c.916A>G (p.Ile306Val; p.I306V)	
			c.92+1G>T	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSODERF Gene Variant	NDERFGENEVAR	NSODERF Gene Variant	c.923dupT (p.Leu308Phefs)	
			c.925G>A (p.Gly309Arg; p.G309R)	
			c.92C>T (p.Pro31Leu; p.P31L)	
			c.92G>C (p.Arg31Thr; Hb Kairouan)	
			c.93-21G>C	
			c.937C>T (p.Arg313Trp; p.R313W)	
			c.939A>G (p.Ile313Met; p.I313M)	
			c.945+1G>C	
			c.946-4C>G	
			c.948delT (1078delT)	
			c.950T>C (p.Val317Ala; p.V317A)	
			c.955C>T (p.Gln319Ter)	
			c.956C>T (p.Ser319Phe; p.S319F)	
			c.958T>G (p.Leu320Val; p.L320V)	
			c.970C>T (p.Arg324Ter)	
			c.629A>G (p.Tyr210Cys; p.Y210C)	
			c.631C>T (p.Arg211Cys; p.R211C)	
			c.632G>A (p.Arg211His; p.R211H)	
			c.638T>C (p.Leu213Pro; p.L213P)	
			c.641A>C (p.Asn214Thr; p.N214T)	
			c.641A>G (p.Asn214Ser; p.N214S)	
			c.641T>A (p.Ile214Lys; p.I214K)	
			c.645C>T (p.Leu215Leu; p.L215L)	
			c.64C>T (p.Pro22Ser; p.P22S)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSODERF Gene Variant	NDERFGENEVAR	NSODERF Gene Variant	c.652C>T (p.Leu218Leu; p.L218L)	
			c.656G>A (p.Arg219His; p.R219H)	
			c.658_660delAAG (p.Lys220del)	
			c.680A>T (p.Asp227Val; p.D227V)	
			c.682G>T (p.Asp228Tyr; D228Y)	
			c.683A>G (p.Asn228Ser; p.N228S)	
			c.688-2A>G	
			c.701C>T (p.Thr234Ile; p.T234I)	
			c.701G>A (p.Arg234Gln; p.R234Q)	
			c.709G>A (p.Ala237Thr; p.A237T)	
			c.710T>A (p.Ile237Asn; p.I237N)	
			c.713T>A (p.Val238Glu; p.V238E)	
			c.719T>A (p.Met240Lys; p.M240K)	
			c.722G>A (p.Arg241His; p.R241H)	
			c.728G>A (p.Arg243Gln; p.R243Q)	
			c.734T>C (p.Val245Ala; p.V245A)	
			c.744-33GATT[7]+[7]	
			c.753-23_753-22del	
			c.753-2A>C	
			c.757G>A (p.Asp253Asn; p.D253N)	
			c.775C>T (p.Arg259Trp; p.R259W)	
			c.779C>T (p.Thr260Met; p.T260M)	
			c.782G>A (p.Arg261Gln; R261Q)	
			c.787G>A (p.Val263Met; p.V263M)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSODERF Gene Variant	NDERFGENEVAR	NSODERF Gene Variant	c.855G>T (p.Lys285Asn; p.K285N)	
			c.8591A>C (p.His2864Pro; p.H2864P)	
			c.860G>A (p.Gly287Asp; p.G287D)	
			c.865G>A (p.Gly289Arg; p.G289R)	
			c.877G>A (p.Ala293Thr; p.A293T)	
			c.881G>A (p.Gly294Glu; p.G294E)	
			c.887_888delCT (p.Pro296Argfs)	
			c.887G>A (p.Arg296Gln; p.R296Q)	
			c.895C>T (p.Arg299Cys; p.R299C)	
			c.896_898delAGA (p.Lys299del)	
			c.209C>T (p.Ser70Leu; p.S70L)	
			c.2128A>T (p.Lys710Ter)	
			c.216+2T>C	
			c.223C>T (p.Arg75Ter)	
			c.224G>A (p.Arg75Gln; R75Q)	
			c.2260G>A (p.Val754Met; V754M)	
			c.227T>C (p.Ile76Thr; Hb F Waynesboro)	
			c.227T>C (p.Leu76Pro; Hb Atlanta)	
			c.233T>C (p.Ile78Thr; p.I78T)	
			c.236G>A (p.Arg79His; p.R79H)	
			c.2490+1G>A (2622+1G>A)	
			c.253G>T (p.Gly85Cys; p.G85C)	
			c.2562T>G (p.Thr854Thr; 2694T>G)	
			c.2604A>G	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSODERF Gene Variant	NDERFGENEVAR	NSODERF Gene Variant	c.262_264delGAG (p.Glu88del; p.E88del)	
			c.276G>T (p.Glu92Asp; p.E92D)	
			c.281 A>G (p.Asp94Gly; p.D94G)	
			c.283G>A (p.Asp95Asn; Hb F Columbus-GA)	
			c.287G>T (p.Gly96Val; p.G96V)	
			c.290A>T (p. Asp97Val; p.D97V)	
			c.293-13C>G	
			c.3067_3072delTAGTG (p.Ile1023Serfs)	
			c.315+1G>A	
			c.316-2A>G	
			c.3196C>T (p.Arg1066Cys; p.R1066C)	
			c.3197G>A (p.Arg1066His; p.R1066H)	
			c.3222T>A (p.Phe1074Leu; p.F1074L)	
			c.331C>T (p.Arg111Ter)	
			c.332_339delGAGACTAC (p.Gly111Valfs)	
			c.3486_3487delAG	
			c.349C>T (p.Arg117Cys; p.R117C)	
			c.34G>A (p.Ala12Thr; p.A12T)	
			c.362C>T (p.Thr121Ile; p.T121I)	
			c.366T>G (p.Asn122Lys; p.D122K)	
			c.372T>A (p.Asp142Glu; p.D142E)	
			c.377G>A (p.Gly126Asp; p.G126D)	
			c.3794G>T (p.Gly1265Val; p.G1265V)	
			c.380C>T (p.Pro127Leu; p.P127L)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSODERF Gene Variant	NDERFGENEVAR	NSODERF Gene Variant	c.382_384delTAC (p.Tyr128del)	
			c.383_385del (p.Val128del)	
			c.384-20A>G	
			c.3846G>A (p.Trp1282Ter; W1282X)	
			c.387+91dupA	
			c.3879delA	
			c.3889dupT(p.Ser1297Phefs*5)	
			c.3905T>C (p.Met1302Thr; p.M1302T)	
			c.394C>T (p.Arg132Stop; p.R132X)	
			c.397G>T (p.Val133Leu; p.V133L)	
			c.-41A>C	
			c.4272C>T (p.Tyr1424Tyr; p.Y1424Y)	
			c.428A>G (p.Asp143Gly; p.D143G)	
			c.42C>A	
			c.436C>T (p.Arg146Trp; pR146W)	
			c.437delT (p.Leu146Trpfs)	
			c.4389G>A (p.Gln1463Gln; p.Q1463Q)	
			c.43G>T (p.Gly15Trp; p.G15W)	
			c.44 T>C (p.Val5Ala; p.V5A)	
			c.442delA (p.Ile148Leufs)	
			c.443A>G (p.Gln148Arg; p.Q148R)	
			c.443C>T (p.Thr148Ile; p.T148I)	
			c.443G>A (p.Arg148Lys; p.R148K)	
			c.443T>C (p.Ile148Asn; p.I148N)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSODERF Gene Variant	NDERFGENEVAR	NSODERF Gene Variant	c.456G>A (p.Trp152Ter)	
			c.470G>A (p.Arg157His; p.R157H)	
			c.473G>A (p.Arg158Gln; p.R158Q)	
			c.507delG (p.Glu169AspfsX11)	
			c.50G>A (p.Gly17Asp; Hb J-Baltimore)	
			c.511G>A (p.Ala171Thr; p.A171T)	
			c.518T>A (p.Ile173Asn; p.I173N)	
			c.51C>G (p.Phe17Leu; p.F17L)	
			c.51dupT (p.Ala5CysfsX5)	
			c.535G>T (p.Gly179Trp; p.G179W)	
			c.547C>T (p.Arg183Trp; p.R183W)	
			c.556C>T (p.Arg186Trp; p.R186W)	
			c.556C>T (p.His186Tyr; p.H186Y)	
			c.557G>A (p.Cys186Tyr; p.C186Y)	
			c.56_57delGCinsAA (p.Arg19Gln; p.R19Q)	
			c.568C>T (p.His190Tyr; p.H190Y)	
			c.572T>C (p.Met191Thr; p.M191T)	
			c.575T>G (p.Phe192Cys; p.F192C)	
			c.577C>T (p.Arg193Trp; p.R193W)	
			c.579+1G>T (711+1G>T)	
			c.604T>C (p.Phe202Leu; p.F202L)	
			c.609G>A (p.Trp203Ter)	
			c.622C>T (p.Arg208Ter)	
			619 bp deletion (g.71609_72227del619)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSODERF Gene Variant	NDERFGENEVAR	NSODERF Gene Variant	c.1000C>T (p.Arg334Trp; R334W)	
			c.1040G>A (p.Arg347His; R347H)	
			c.1062+5G>A (IVS12+5 G>A)	
			c.-119_-116delGTCA (5'UTR-119delGTCA)	
			c.1193C>T (p.P398L)	
			c.126_129delCTTT (p.Phe42fs)	
			c.1316G>A; (p.Gly439Asp; p.G439D)	
			c.1327_1330dup (p.Ile444Argfs)	
			c.1330G>C (p.D444H)	
			c.1364C>A (p.Ala455Glu; A455E)	
			c.1367T>C (p.Val456Ala)	
			c.1368A>C (p.Q456H)	
			c.1413T>C (p.C471C)	
			c.1436C>T (p.P479L; p.Pro479Leu)	
			c.1463G>A (p.R488H)	
			c.1466C>A (p.Ser489X; S489X)	
			c.1508_1512delCCATG	
			c.1519_1521delATC (p.Ile507del; I507del; DI507)	
			c.1521_1523delCTT (p.Phe508del; F508del; dF508)	
			c.1523T>G (p.Phe508Cys; F508C)	
			c.1558G>T (p.Val520Phe; V520F)	
			c.1595C>T (p.T532M)	
			c.1616C>A	
			c.1624G>T (p.Gly542X; G542X)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSODERF Gene Variant	NDERFGENEVAR	NSODERF Gene Variant	c.1630G>A (p.Gly544Ser)	
			c.1646G>A (p.Ser549Asn; S549N)	
			c.1652G>A (p.Gly551Asp; G551D)	
			c.1657C>T (p.Arg553X; R553X)	
			c.1675G>A (p.Ala559Thr; A559T)	
			c.1679G>C (p.Arg560Thr; R560T)	
			c.1705T>G (p.Tyr569Asp; Y569D)	
			c.1766+1G>A (1898+1G>A)	
			c.178G>T (p.Glu60X; E60X)	
			c.190A>G (p.Met64Lys)	
			c.192G>T (p.Gln64His; Q64H)	
			c.199 T>C (p.Tyr67His; p.Y67H))	
			c.19G>A (HbC; p.Glu7Lys)	
			c.200C>T (p.Pro67Leu; P67L)	
			c.20A>T (HbS; p.Glu7Val)	
			c.216+1G>A	
			c.2162T>A (p.Val721Glu)	
			c.220G>A (p.Asp74Asn; Hb G-Accra; Korle-Bu variant of Hb)	
			c.223G>C (p.Asp74Gly; HbQ-Thailand)	
			c.2246G>T (p.R749L; p.Arg749Leu)	
			c.233C>A (p.Pro78His; HbToulon)	
			c.237C>A (p.Asn79Lys; HbStanleyville-II)	
			c.254G>A (p.Gly85Glu; G85E)	
			c.262_263delTT (p.Leu88IlefsX22; 394delTT)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSODERF Gene Variant	NDERFGENEVAR	NSODERF Gene Variant	c.263T>A (p.Leu88X)	
			c.2657+2_2657+3insA (2789+2insA)	
			c.2657+5G>A (2789+5G>A)	
			c.271dupA (p.Arg91Lysfs)	
			c.2734T>A (p.Ser912Thr)	
			c.278-41del	
			c.27dupG (c.27_28insG; p.Ser10Valfs*14)	
			c.2988+1G>A (3120+1G>A)	
			c.305G>A (p.Arg117His; R117H)	
			c.307A>C (p.Ser103Arg; HbManitoba I)	
			c.3080T>C (p.Ile1027Thr; I1027T)	
			c.3276C>A (p.Tyr1092X; Y1092X)	
			c.3276C>G (p.Tyr1092X; Y1092X)	
			c.3454G>C (p.Asp1152His; D1152H)	
			c.3484C>T (p.Arg1162X; R1162X)	
			c.364G>C (p.Glu121Gln; HbD)	
			c.364G>C (p.Glu121Gly; HbD-Punjab; Hb D-Los Angelese)	
			c.3659delC (p.Thr1220LysfsX8)	
			c.367G>A (p.Gly123Arg)	
			c.3717+12191C>T (3849+10kbC>T)	
			c.3731G>A (p.Gly1244Glu; G1244E)	
			c.3773_3774insT (3905insT)	
			c.3808G>A (p.Asp1270Asn; D1270N)	
			c.3909C>G (p.Asn1303Lys; N1303K)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSODERF Gene Variant	NDERFGENEVAR	NSODERF Gene Variant	c.3937C>T (p.Gln1313X; Q1313X)	
			c.395C>G (p.P132R)	
			c.424G>T (p.A142S)	
			c.427T>C (p.Ter143Glnext32; HbCS; HbConstant Spring)	
			c.46G>C (p.Gly15Arg; HbOttawa)	
			c.470G>A (p.R157H)	
			c.489+1G>T (621+1G>T)	
			c.511G>A (p.A171T)	
			c.513C>G (p.Asp171Glu; p.D171E)	
			c.51delC (p.Lys18Argfs)	
			c.52A>T (p.Lys18Ter)	
			c.554-1G>T (IVS6-1 G>T)	
			c.563A>G (p.Gln188Arg; p.Q188R)	
			c.583G>A (p.G195R)	
			c.607-6T>G (IVS7-6 T>G)	
			c.616C>T (p.R206C)	
			c.617T>G (p.Leu206Trp; L206W)	
			c.623-2_623-1del	
			c.744-33GATT[6]+[6] (TTAG repeats)	
			c.744-33GATT[6]+[7] (TTAG repeats)	
			c.776C>T (p.Ala259Val)	
			c.782C>T (p.Pro261Leu; P261L)	
			c.799 G>A (p.Gly267Arg; p.G267R)	
			c.-79A>G (-29A>G)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSODERF Gene Variant	NDERFGENEVAR	NSODERF Gene Variant	c.79G>A (p.Glu27Lys; HbE)	
			c.842G>C	
			c.848T>C (p.V283A)	
			c.869+11C>T (1001+11C>T)	
			c.889_891del (p.Glu297del; p.E297del))	
			c.91+5G>T (IVS1+5G>T; IVS2+5G>T)	
			c.91G>A (p.Glu31Gln; HbG-Honolulu)	
			c.92+5G>C (IVS1-5 G>C)	
			c.940A>G (p.Asn314Asp; N314D)	
			C.941C>T (p.Ala314Val)	
			c.98_104delGCGGCTGinsTCC (p.Cys33Phefs; c.98_104del7ins3)	
			c.985A>G (p.Lys329Glu; p.K329E)	
			Complete deletion	
			deletion exon 2	
			deletion exons 4-11	
			--FIL (g.11684_43534del31851)	
			--MED (g.24664_41064del16401)	
			--SEA (g.26264_45564del19301)	
			--South African (g.209051_232787delins157)	
			--THAI (g.10664_44164del33501)	
			-α3.7 (3.7-kb rightward deletion)	
			-α4.2 (4.2-kb leftward deletion)	
			Other, please specify:	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
NSODERF Gene Variant Other	NDERFGNVROTHER	NSODERF Gene Variant Other		
NSODERF Gene Variant Phase Confirmed flag	NSODERFPHCONF	NSODERF Gene Variant Phase Confirmed flag		
NSODERF Phase Confirm Method	NSOFUPHSCONF	NSODERF Phase Confirm Method	Family studies	
			Other, specify:	
Number of siblings of affected after diagnosis of newborn screen positive child	NBS0042	Number of affected full siblings to the screen positive child who diagnosed after the current screen positive	Unknown	
			0	
			1	
			2	
			3	
			4	
			5	
			6	
			7	
			8	
			9	
			10	
			>10	
Number of siblings of affected prior to diagnosis of newborn screen positive child	NBS0041	Number of affected full siblings to the screen positive child who diagnosed prior to the current screen positive	Unknown	
			0	
			1	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Number of siblings of affected prior to diagnosis of newborn screen positive child	NBS0041	Number of affected full siblings to the screen positive child who diagnosed prior to the current screen positive	2	
			3	
			4	
			5	
			6	
			7	
			8	
			9	
			10	
			>10	
Other affected family members of affected newborn screen positive child	NBS0032	Other family members of the infant / child who are affected with the disease in question. Specify relationship.	Mother	
			Father	
			Aunt, Maternal	
			Aunt, Paternal	
			Uncle, Maternal	
			Uncle, Paternal	
			Cousin, Maternal	
			Cousin, Paternal	
			Niece, Maternal	
			Niece, Paternal	
			Nephew, Maternal	
			Nephew, Paternal	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Other affected family members of affected newborn screen positive child	NBS0032	Other family members of the infant / child who are affected with the disease in question. Specify relationship.	Grandfather, Maternal	
			Grandfather, Paternal	
			Grandmother, Maternal	
			Grandmother, Paternal	
			Maternal half-sibling	
			Paternal half-sibling	
			Other, specify:	
Parent Diagnosed Prior to Positive NSO Screen Flag	NBS0015	Indicates whether the mother was diagnosed prior to the positive newborn screen obtained for the infant.		
Parents/Health Care Provider informed of positive newborn screen result	D0078	Have the parents or the infant's health care provider been informed of the positive newborn screen result		
Parents/Health Care Provider informed of positive newborn screen result Date	D0078DATE			
Prenatal Diagnostic Procedure Disposition	PSDP001	An indication of whether a prenatal diagnostic procedure was offered, declined, accepted or unknown.		
Reason comments for no definitive diagnosis established	NDERFNODEFC			
Reason for no definitive diagnosis established	NDERFNODEF	Please make a selection if the infant was lost to follow-up, or was deceased prior to establishment of diagnosis, or if no definitive diagnosis could be established for some reason. ** IF INFANT IS UNAFFECTED or a CARRIER (PRESUMED OR CONFIRMED), this is a "Definitive Diagnosis". Please click below and select "unaffected" or "carrier".		
Reason infant not retreived	NDERF0123	Indicates the reason(s) that an infant has not been retrieved at the time Retrieval Confirmation Information is submitted to BORN	Triaged	
			Awaiting call back from parents	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Reason infant not retreived	NDERF0123	Indicates the reason(s) that an infant has not been retrieved at the time Retrieval Confirmation Information is submitted to BORN	Incorrect contact information	
			No contact information for primary health care provider	
			Family has moved	
			Phone number out of service	
			Awaiting call back from primary health care provider	
			Unable to contact health care provider	
			Infant is deceased	
			Transferred to another treatment centre	
			Incorrect health care provider listed	
			No voice mail to leave message	
			Registered mail undeliverable	
			CAS involved to help locate	
			Family / parents out of country	
			Appointment pending	
			Other	
			No health care provider indicated	
			Parents contacted directly	
			No contact information for family	
Reason infant not retreived - Other	NDERF0123O	The reason that an infant has not been retreived at the time Retrieval Confirmation Information is submitted to BORN not represented in the pick list		
Reason Infant Was Not Referred NSO Followup	NDERFNRR	Reason Infant Was Not Referred for NSO Followup	Infant under 24 hours at sample collection and a normal repeat received	
			Infant under 24hrs at sample collection, no repeat received as infant is deceased	
			SCID prem, infant screen negative on repeat	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Reason Infant Was Not Referred NSO Followup	NDERFNRR	Reason Infant Was Not Referred for NSO Followup	SCID prem, infant deceased	
			SCID prem, infant referred on repeat	
			Other	
Receive Entered Results CCPID Flag	NSOSTFU2030	Receive Entered Results CCPID Flag		
Receive Entered Results Flag	NSOSTFU2040	Receive Entered Results Flag		
Regional Treatment Centre Responsible Physician	NBS0129	The name of the physician at the Regional Treatment Centre who was / is responsible for the care of the infant / child	ALABDOULSALAM, DR. TAREQ (Children's Hospital of Winnipeg)	
			ATHALE, DR. UMA (Hamilton Health Sciences Centre)	
			BABIC, DR. BOJANA (Hamilton Health Sciences Centre)	
			Brager, Dr.Rae HHSC paediatric immunologist	
			BOLAND, DR. MARGARET PATRICIA (Children's Hospital of Eastern Ontario)	
			BORICI-MAZI, DR.	
			CHAKRABORTY, DR. PRANESH (Children's Hospital of Eastern Ontario)	
			CLARSON, DR. CHERIL LINDA (London Health Sciences Centre - Endocrinology)	
			DEAN, HEATHER	
			DENT, DR. PETER	
			ELLIS, DR ANNE	
			GALLEGO, DR. P (London Health Sciences Centre - Endocrinology)	
			GERAGHTY, DR. MICHAEL (Children's Hospital of Eastern Ontario)	
			GOOBIE, DR. SHARAN LYNN (London Health Sciences Centre - Genetics)	
			GRASEMANN, HARTMUT (Hospital for Sick Children)	
			HADJIYANNAKIS, DR. ANASTASIA (Children's Hospital of Eastern Ontario)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Regional Treatment Centre Responsible Physician	NBS0129	The name of the physician at the Regional Treatment Centre who was / is responsible for the care of the infant / child	HAMILTON, DR. JILL KRYSTI (Hospital for Sick Children)	
			INBAR-FEIGENBERG, DR. MICHAL (Hospital for Sick Children)	
			JARDINE, DR. Lawrence (London Health Sciences Centre - Genetics)	
			JUSKEY, LIA (Hospital for Sick Children)	
			Karp, Dr. Natalya (London Health Sciences Center	
			KIRBY, DR. MELANIE-ANN (Hospital for Sick Children)	
			KLAASSEN, DR. ROBERT JOHN (Children's Hospital of Eastern Ontario)	
			KOVESI, DR. THOMAS ANDREW (Children's Hospital of Eastern Ontario)	
			KOZENKO, DR. MARIYA (Hamilton Health Sciences Centre)	
			KRONICK, DR. JONATHAN (Hospital for Sick Children)	
			LAPIERRE, DR. JEAN GUY (CHU St Justine)	
			LAWRENCE, DR. SARAH ELIZABETH (Children's Hospital of Eastern Ontario)	
			LI, DR. CHUMEI (Hamilton Health Sciences Centre)	
			MACKENZIE, DR. JENNIFER JANE (Kingston General Hospital)	
			MACKENZIE, DR. JENNIFER (Hamilton Health Sciences Centre)	
			MAHMUD, DR. FARID HUSSAIN (Hospital for Sick Children)	
			MAHMUTOGLU, DR. SAADET (Hospital for Sick Children)	
			MAZZA, DR. J.	
			McASSEY, DR. KAREN LYNN (Hamilton Health Sciences Centre)	
			MHANNI, DR AZIZ (WRHA Genetics & Metabolism Program)	
			MOOTE, DR. B.	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Regional Treatment Centre Responsible Physician	NBS0129	The name of the physician at the Regional Treatment Centre who was / is responsible for the care of the infant / child	NOWACZYK, DR. MALGORZATA JOANNA (Hamilton Health Sciences Centre)	
			ODAME, DR. ISAAC (Hospital for Sick Children)	
			PALMERT, DR. MARK RANEY (Hospital for Sick Children)	
			PASTERKAMP, DR. HANS (CHILDREN'S HOSPITAL OF WINNIPEG)	
			PEDDER, DR. LINDA (Hamilton Health Sciences Centre)	
			PERLMAN, DR. KUSIEL (Hospital for Sick Children)	
			PHAM-HUY, DR. ANNE (CHEO Infectious Diseases)	
			POTTER, DR. MURRAY ALEXANDER (Hamilton Health Sciences Centre)	
			PRASAD, DR. CHITRA (London Health Sciences Centre - Genetics)	
			PRICE, DR. APRIL KATHERINE (London Health Sciences Centre - Genetics)	
			RAIMAN, DR. JULIAN ANDREW JONATHON (Hospital for Sick Children)	
			RATJEN, DR. FELIX ALEXANDER (Hospital for Sick Children)	
			SALEH, DR. MAHA (London Health Sciences Centre)	
			ROIFMAN, DR. CHAIM	
			ROCKMAN-GREENBERG, DR. CHERYL (WRHA Genetics & Metabolism Program)	
			SALEH, DR.DAVID (Kingston General Hospital)	
			SAMAAN, DR. CONSTANTINE (Hamilton Health Sciences Centre)	
			SCHULZE, DR. ANDREAS (Hospital for Sick Children)	
			SELLERS, ELIZABETH	
			SILVA, DR. MARIANA (Kingston General Hospital)	
			SIRIWARDENA, DR. KOMUDI PULSARA (Hospital for Sick Children)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Regional Treatment Centre Responsible Physician	NBS0129	The name of the physician at the Regional Treatment Centre who was / is responsible for the care of the infant / child	SIU, DR. V.	
			SOCHETT, ETIENNE (Hospital for Sick Children)	
			SOLOMON, MELINDA (Hospital for Sick Children)	
			SOTTOSANTI, DR. M (London Health Sciences Centre - Endocrinology)	
			STEIN, DR. ROBERT IAN (London Health Sciences Centre - Endocrinology)	
			URBACH, DR. STACEY LISA (Hospital for Sick Children)	
			VAN WYLICK, DR. RICHARD CECIL (Kingston General Hospital)	
			VANDERMEULEN, DR. JOHN AUGUST (Hamilton Health Sciences Centre)	
			WASSERMAN, DR. JONATHAN (Hospital for Sick Children)	
			WHERRETT, DR. DIANE KATHERINE (Hospital for Sick Children)	
			WICKLOW, BRANDY	
			BASSILIOUS, DR. ERENY (McMaster Children's Hospital)	
			SIMPSON, EWURABENA (Children’s Hospital of Eastern Ontario)	
			REISMAN, JOE (Children’s Hospital of Eastern Ontario)	
			BERUBE, DR. DENIS (CHU St Justine)	
			FROSK, DR. PATRICK (WRHA Genetics & Metabolism Program)	
			Iqbal, Shaikh Mohammed (Children’s Hospital of Winnipeg)	
			WALIA, DR.JAGDEEP (Kingston General Hospital)	
			Kumar, Dr. Gautam (Sudbury CF Clinic)	
			Al-Somali, Dr. Faisal (WRHA Respirology)	
			Marcotte, Dr. Jacques (CHU St Justine)	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Regional Treatment Centre Responsible Physician	NBS0129	The name of the physician at the Regional Treatment Centre who was / is responsible for the care of the infant / child	BRUNEL-GUITTON, DR.CATHARINE (CHE Sainte Justine, Medical Genetics)	
			Guerin, Dr.Andrea (Kingston General Hosptial)	
			COHN, Dr. RONALD (The Hospital for Sick Children)	
			Sondheimer, Dr. Neal (The Hospital for Sick Children)	
			Rusnak, Dr. Allison (Kingston General Hospital)	
			HARRINGTON, DR. JENNIFER JEAN (Hospital for Sick Children)	
			SHULMAN, DR. RAYZEL MALKA (Hospital for Sick Children)	
Screen Positive Comments	NDERF02325			
Share information with NSO	NDERFSHARE	Parents/guardians have requested that the diagnostic evaluation information not be shared with NSO.		
Specify jurisdiction	NDERFOBTJURS	The location where the infant's newborn screening sample was obtained if it was obtained in a jurisdiction other than Ontario		
Test Not Done	NDERFTND	Indicates that the diagnostic investigation was not performed		
Total Number of affected siblings of affected newborn screen positive child	NBS0031	Number of full siblings to the infant / child who are affected with the disease in question.	Unknown	
			0	
			1	
			2	
			3	
			4	
			5	
			6	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Total Number of affected siblings of affected newborn screen positive child	NBS0031	Number of full siblings to the infant / child who are affected with the disease in question.	7	
			8	
			9	
			10	
			>10	
Total Number of deceased sibs of affected newborn screen positive child	NBS0108	Number of full siblings to the infant / child that are deceased.	Unknown	
			0	
			1	
			2	
			3	
			4	
			5	
			6	
			7	
			8	
			9	
			10	
			>10	
Total Number of siblings of affected newborn screen positive child	NBS0030	Total number of full siblings to the infant / child.	Unknown	
			0	
			1	

Encounter: NSO DERF

Data Element Name	BORN ID	Data Element Definition	Pick List Value	Pick List Definition
Total Number of siblings of affected newborn screen positive child	NBS0030	Total number of full siblings to the infant / child.	2	
			3	
			4	
			5	
			6	
			7	
			8	
			9	
			10	
			>10	
Unwell at retrieval	NBS0111-01	The symptom(s) and / or reason(s) the screen positive infant was unwell at the time of retrieval		
Was newborn screening obtained in another jurisdiction?	NDERFOBTJURFG	An indication of whether a dried blood spot or other sample type was obtained for newborn screening in a location other than Ontario as reported to the health care provider completing the SDRF.		
Was NSO obtained?	NDERFOBT	An indication of whether a dried blood spot was obtained for newborn screening in Ontario as reported to the health care provider completing the SDRF		
Was the diagnosis made in the infant?	NDERFDDINF	An indication of whether the definitive diagnosis was made in the infant.		
Was the diagnosis made in the mother?	NDERFDDMOTH	An indication of whether the definitive diagnosis was made in the mother.		