

Element group	element	value	format	
Common Data Elements	Castor record ID		String	
	Pseudonym		String	
	Date of inclusion		date	
	Date of birth	partial date		Date (mm/yyyy)
	Age at inclusion			number
	Gender			radiobutton
			Female	string
			Male	string
			Undetermined	string
			Fetus (Unknown)	string
	Patient status			
			Alive	string
			Dead	string
			Lost in follow-up	string
			Opted-out	string
	Date of death			Date (mm/yyyy)
	First contact with specialised centre			Date (dd/mm/yyyy)
	Moment of onset	radiobutton		string
			Antenatal	string
			At birth	string
			Date	string
			Undetermined	string
Age at onset	age in years		number	
Age check	0 or 1		number	
Date of onset	calculation: date_of_birth + age_at_onset		partial date	
Is there a diagnosis?	yes/no			
Moment of diagnosis	radiobutton			
		Antenatal	string	
		At birth	string	
		Date	string	

		Undetermined	string
	Date of diagnosis	date	Date (dd/mm/yyyy)
	Diagnosis	ORPHA codes	
	Undiagnosed case	HPO codes	
First line family history			
	Is there information on family history?	yes/no	
	parents affected	yes/no/unknown	
	which parents		checkboxes
		mother/ father	
	do you have (other) children	yes/no	string
	Are some of the children affected (if yes)	yes/no/unknown	string
	number of male children affected	number	number
	number of female children affected	number	number
	Are some of the children unaffected (if yes)	yes/no/unknown	string
	number of male children unaffected	number	number
	number of female children unaffected	number	number
	Do you have siblings (if yes)	yes/no	string
	Are some of these siblings affected (if yes)	yes/no/unknown	string
	number of male siblings affected	number	number
	number of female siblings affected	number	number
	Are some of these siblings unaffected (if yes)	yes/no/unknown	string
	number of male siblings unaffected	number	number
	number of female siblings unaffected	number	number
Second line family history			
	grandparents affected	yes/no/unknown	
		grandmother	checkboxes
		grandfather	checkboxes
	Uncles affected	yes/no/unknown	string

	number of uncles affected	number	number
	Aunts affected (if yes)	yes/no/unknown	string
	number of aunts affected	number	number
Body Parts involved			
	arm(s) (if yes)	yes/no	string
	arms	left/ right/ both	string
	leg(s) (if yes)	yes/no	string
	legs	left/ right/ both	string
	other bodyparts	yes/no	string
	which bodyparts		string
		face/ ears/ conjunctiva/ trunk/ genitals	
	generalized bodyparts affected	yes/no	string
	which generalized bodyparts are affected		
		thorax i.e. pleural effusions	string
		abdomen i.e. ascites	string
		gut – bowel i.e. intestinal lymphangiectasia	string
		urinary tract i.e. chylouria	string
		genitals i.e. chylous reflux	string
intervention			
	has intervention taken place?	yes/no	
	Intervention type?		checkbox
		conservative	string
		operative	string
		interventional radiology	string
	Location of conservative treatment		radiobutton
		ambulatory	string
		in hospital	string
	operative		checkbox
		reconstructive	string
		reductive	string
		others	string
	interventional radiology	checkbox	string
genetics			
	has genetic testing been done (if yes)?	yes/no	
	array CGH/ chromosome testing (if yes)	yes/no	

	sample type		
		blood	string
		tissue	string
	was genetic abnormality detected?	yes/no	
	copy number variation	One of the 23 chromosomes	string
	type of CNV	deletion/duplication/triplication	string
	gene panel (if yes)	yes/no	
	from which sample		
		blood	string
		tissue	string
	was a single gene abnormality detected? (if yes)	yes/no	
	Add genetic diagnosis	repeating measure	
			HGVS/HGNC/OMIM
	Did the DNA analysis confirm the mutation?		
		Pathogenic variant detected - class 5	string
		Likely pathogenic variant detected - class 4	string
		Variant of uncertain significance detected - class 3	string
		Likely benign variant detected - class 2	string
		Benign variant detected - class 1	string
	Is the patient homo- or heterozygous for the mutation?		
		Homozygous -biallelic	string
		Heterozygous - monoallelic	string
		Compound heterozygous -biallelic	string
	Is the mutation found in a germline or mosaicism?		
		germline	string
		somatic mosaicism	string
	single gene analysis	yes/no	
	Sample type		
		blood	string
		tissue	string
	was a single gene abnormality detected?	yes/no	
	Add genetic diagnosis	repeating measure	
			HGVS/HGNC/OMIM
	Did the DNA analysis confirm the mutation?		
		Pathogenic variant detected - class 5	string

		Likely pathogenic variant detected - class 4	string
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	Is the mutation found in a germline or mosaicism?		
		germline	string
		somatic mosaicism	string
	whole genome/ exome	yes/no	
	Sample type		
		blood	string
		tissue	string
	was genetic abnormality detected?	yes/no	
	Add genetic diagnosis		
			HGVS/HGNC/OMIM
	Did the DNA analysis confirm the mutation?		
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