Elementchapter	elements	value
Common Data elements		
Pseudonym	Pseudonym	string
Personal information	Date of inclusion	date
	Date of birth	date
	Sex	string
Patient status	Patient's status	string
	Date of death	date
Care pathway	First contact with specialised centre	date
Disease history	Moment of onset	String / date
	Moment of diagnosis	String / date
Diagnosis	Diagnosis of the rare disease	Orphanet Code
	Genetic diagnosis	HGNC gene code
	Undiagnosed case	HPO code
Biological sample	If yes	yes/no
Link to a biobank	If yes	yes/no
	Biobank	string

Elementchapter	Elements	value
Family history		
1st line of family history		
Parents infected	If yes	yes/no
	mother	String
	father	String
Do you have children	If yes	yes/no
Are some of your children affected	if yes	yes/no
	male / female	number of each
Are some of your children unaffected	if yes	yes/no
	male / female	number of each
Do you have siblings	If yes	Yes/no
Are some of these siblings affected	if yes	yes/no
	male / female	number of each
Are some of these siblings unaffected	If yes	Yes/no
	male / female	number of each
2nd line of family history		
Grandparents affected?	If yes	yes/no
	Grandmother infected?	String
	Grandfather infected?	string
Aunts infected	If yes	
	Number affected	number
Uncles infected	If yes	
	Number affected	number

Elementchapter	Elements	value
Body Parts involved		
arm(s) (if yes)	If yes	Yes/no
	left/ right/ both	string
leg(s) (if yes)	If yes	Yes/no
	left/ right/ both	string
other	face/ ears/ conjunctiva/ trunk/ genitals	string
generalized	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?	(if yes)	Yes/no
	Conservative (if chosen)	string
	In hospital	string
	Ambulatory	string
	operative (if chosen))	string
	reconstructive	string
	reductive	string
	others	string
	interventional radiology	
genetic tested		
has genetic testing been done?	(if yes)	Yes/no
	type of testing	Dropdown 4 options
array CGH/ chromosome testing (if yes)		string
	from which sample?	
	blood	string
	tissue	string
	was genetic abnormality detected? (if yes)	Yes/no

	copy number variation / chromosome number	
	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 genetic abnormality detected? (if yes)	Yes/no
	HGVS/HGNC/OMIM	
single gene analysis (if yes)		
	from which sample?	
	blood	
	tissue	
	was genetic abnormality detected? (if yes)	
	HGVS/HGNC/OMIM	
whole genome/ exome (if yes)		
	from which sample?	
	blood	
	tissue	
	was genetic abnormality detected? (if yes)	
	HGVS/HGNC/OMIM	
genetic outcomes		
Did the DNA analysis confirm the mutation?		
	pathogenic variant detected	string
	variant of uncertain significance	String
	no pathogenic variant or VUS detected	String
	likely benign	String
	benign	string
Is the patient homo- or heterozygous for the mutation?		
	heterozygous - monoallelic	string
	homozygous - biallelic	string
	compound heterozygous - biallelic	string
Is the mutation found in a germline or mosaicism?		

	germline	string
	somatic mosaicism	string