

MANDATORY ITEMS	MOLECULAR DATA	DMD registry Mutation name in <i>DMD</i> gene following HGVS rules (based on cDNA Ref Seq) Deletion: all exons tested (Yes / No / Unknown) Duplication: all exons tested (Yes / No / Unknown) Deletion/Duplication: boundaries known (Yes / No / Unknown) Point mutation: all exons sequenced (Yes / No / Unknown) Targeted mutation testing in the patient but testing of all exons in a relative male patient (Yes / No / Unknown)	SMA registry Mutation name in <i>SMN1</i> gene following HGVS rules (based on cDNA Ref Seq)	Definition
				NB: It is highly recommended to test all exons for deletions due to the occurrence of non-contiguous rearrangements.
				NB: It is highly recommended to test all exons for duplications due to the occurrence of non-contiguous rearrangements.
				This item should be filled in ONLY when the "No" answer has been selected for one of the three boxes above (deletion, duplication, point mutation).
	DIAGNOSIS	Diagnosis (Duchenne Muscular Dystrophy (DMD) / Becker Muscular Dystrophy (BMD) / Intermediate Muscular Dystrophy (IMD) / Female carrier / Unknown)	Diagnosis (SMA / Other / Unknown)	SMA: Spinal Muscular Atrophy
	MOTOR FUNCTION	Currently able to walk (Yes / No)	Currently able to walk (Yes / No)	Functional walking with or without help (orthoses or assistive device or human assistance), inside or outdoors
			Currently able to sit without support (Yes / No)	Able to maintain the sitting position on a chair or a wheelchair without support of upper limbs or leaning against the back of the chair
			Best motor function achieved (Walking [specify age of acquisition] / Sitting independently [specify age of acquisition] / Never able to walk or sit independently)	Walking: functional walking with or without help (orthoses or assistive device or human assistance), inside or outdoors / Sitting independently: able to maintain the sitting position on a chair or a wheelchair without support of upper limbs or leaning against the back of the chair
		Wheelchair use (if over 3 years of age) (Yes (permanent) / Yes (intermittent) / Never / Unknown; if Yes, specify age of beginning)	Wheelchair use (if over 3 years of age) (Yes (permanent) / Yes (intermittent) / Never / Unknown; if Yes, specify age of beginning)	Yes (permanent): patient is not able to walk and needs a wheelchair to move / Yes (intermittent): patient is still able to walk
	STEROIDS	Current steroid therapy (Yes, currently / No, but previously / Never / Unknown)		Any type of steroids
	SCOLIOSIS SURGERY	Scoliosis Surgery (Yes / No / Unknown)	Scoliosis Surgery (Yes / No / Unknown)	Scoliosis: lateral curvature of spine in the coronal plane with a Cobb angle measuring more than 10°; surgery: any type of surgical procedure
	FEEDING FUNCTION		Gastric/nasal tube (Yes / No / Unknown)	Nutritional supplementation via nasogastric or nasojejunal tube or gastrostomy
	HEART	Current cardiac medication (Yes / No / Unknown; if Yes, please list medication)		Specify each drug with its International Nonproprietary Name (INN). In a self-report registry, a list of substances should be given, together with the (country-dependent) name of the drug
	CLINICAL TRIALS	Currently included in a clinical trial (Yes, currently / No, but previously / Never / Unknown; if Yes, currently please specify the name of the drug)	Currently included in a clinical trial (Yes, currently / No, but previously / Never / Unknown; if Yes, currently please specify the name of the drug)	Specify the name of the drug in free text, plus the name of the trial, trial-sponsor (company) and trial phase/time/date. In a self-report registry, a list of (country-specific) trials should be given
TRANSFER ITEMS		Age Last follow-up Date (day/month/year) or Age ([In years])	Age Last follow-up Date (day/month/year) or Age ([In years])	month and year of birth Each registry has to choose one format : date (day/month/year) or age (in years). The two formats should not be mixed in the same column. In a self-report registry, the 'last follow-up' is the last update
		Regional localising information	Regional localising information	Province / State or part of ZIP code / Postcode

		DMD registry	SMA registry	Definition
HIGHLY ENCOURAGED ITEMS	MOTOR FUNCTION	Currently able to sit without support (Yes / No)		Able to maintain the sitting position on a chair or a wheelchair without support of upper limbs or leaning against the back of the chair
	HEART	Heart failure / cardiomyopathy (Yes / No / Unknown)		Clinical signs of heart failure
		Last LVEF (by ultrasound of the heart, in %, specify date [or age] of examination)		Left Ventricular Ejection Fraction determined by ultrasound examination of the heart, expressed in %: [(End diastolic volume - End systolic volume) / End diastolic volume] X 100
	RESPIRATORY FUNCTION	Non invasive ventilation (Yes (all day) / Yes (part-time) / No / Unknown)	Non invasive ventilation (Yes (all day) / Yes (part-time) / No / Unknown)	Mechanical ventilation with nasal or buccal mask / part-time: usually at night
		Invasive ventilation (Yes (all day) / Yes (part-time) / No / Unknown)	Invasive ventilation (Yes (all day) / Yes (part-time) / No / Unknown)	Mechanical ventilation with tracheostomy / part-time: usually at night
		Last FVC (% , specify date or [age] of examination)	Last FVC (% , specify date or [age] of examination)	Pulmonary function test with spirometry: Forced Vital Capacity expressed as percentage predicted for height (not in ml)
	MUSCLE BIOPSY	Previous muscle biopsy (Yes / No / Unknown)		
OTHER REGISTRIES	Signed up for other registries (Yes / No / Unknown; if Yes, please specify which registry)	Signed up for other registries (Yes / No / Unknown; if Yes, please specify which registry)	Registry name is in free text	
FAMILY HISTORY	Other affected family members (Yes / No / Unknown; if Yes: please specify)	Other affected family members (Yes / No / Unknown; if Yes: please specify)	If Yes: specify in free text	
SMA classification		SMA classification (SMA1 / SMA2 / SMA3)	SMA 1: onset between 0-6months, nevers sits, natural age of death <2 years / SMA 2: onset between 7-18 months, never stands, natural age of death >2 years / SMA 3: onset >18 months, stands and walks (this highest function may be lost during evolution)	
MOLECULAR DATA		Number of SMN2 copies		