

Appendix 1  
 SOPeIT-87 Curator requirements and competency testing  
 GBA gene- Gaucher disease association

Version

1

**EXTERNAL DATABASES**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
OMIM -based queries	D			
PubMed- based queries	D			
GeneReviews- based queries	D			
Orphanet- based queries	D			
HPO- based queries	D			

**IN HOUSE SOFTWARE**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
Gepado handling	D			
UniDB handling	D			
CuRepo handling	D			

**DIGITAL PRODUCTS**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
CentoMD	D			
CentoLSD	D			

**GBA- GAUCHER ASSOCIATIONS**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
Gaucher disease description	D			
Gaucher types and specifics	D			
GBA gene	D			
G2P correlation (GBA2Gaucher)	D			
Specific literature and current knowledge	D			

	Name		Signature	Date
<b>Trainee</b>				
<b>Trainer</b>				
<b>Head of department</b>				
SUMMARY: statement competency test passed- valid until- (MM.YYYY)				

Appendix 2  
 SOPeIT-87 Curator requirements and competency testing  
 GLA gene- Fabry disease association

Version

1

**EXTERNAL DATABASES**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
OMIM -based queries	D			
PubMed- based queries	D			
GeneReviews- based queries	D			
Orphanet- based queries	D			
HPO- based queries	D			

**IN HOUSE SOFTWARE**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
Gepado handling	D			
UniDB handling	D			
CuRepo handling	D			

**DIGITAL PRODUCTS**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
CentoMD	D			
CentoLSD	D			

**GBA- GAUCHER ASSOCIATIONS**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
Fabry disease description	D			
Fabry types and specifics	D			
GLA gene	D			
G2P correlation (GLA2Fabry)	D			
Specific literature and current knowledge	D			

	Name		Signature	Date
<b>Trainee</b>				
<b>Trainer</b>				
<b>Head of department</b>				
SUMMARY: statement competency test passed- valid until- (MM.YYYY)				

Appendix 3  
 SOPeIT-87 Curator requirements and competency testing  
 GBA variant curation

Version

1

**EXTERNAL DATABASES**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
OMIM -based queries	D			
PubMed- based queries	D			
GeneReviews- based queries	D			
Orphanet- based queries	D			
HPO- based queries	D			
HGVS nomenclature	D			

**IN HOUSE SOFTWARE**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
Gepado handling	D			
UniDB handling	D			
CuRepo handling	D			

**DIGITAL PRODUCTS**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
CentoMD	D			
CentoLSD	D			

**GBA variant evaluation**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
Variant ID	D			
Variant ID	D			
Variant ID	D			

Variant ID	D			
Variant ID	D			

**GBA -based warnings**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
Warning ID	D			
Warning ID	D			
Warning ID	D			
Warning ID	D			
Warning ID	D			

	Name	Signature	Date
<b>Trainee</b>			
<b>Trainer</b>			
<b>Head of department</b>			
SUMMARY: statement competency test passed- valid until- (MM.YYYY)			

Appendix 4  
 SOPeIT-87 Curator requirements and competency testing  
 GLA variant curation

Version

1

**EXTERNAL DATABASES**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
OMIM -based queries	D			
PubMed- based queries	D			
GeneReviews- based queries	D			
Orphanet- based queries	D			
HPO- based queries	D			
HGVS nomenclature	D			

**IN HOUSE SOFTWARE**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
Gepado handling	D			
UniDB handling	D			
CuRepo handling	D			

**DIGITAL PRODUCTS**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
CentomD	D			
CentomLSD	D			

**GLA variant evaluation**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
Variant ID	D			
Variant ID	D			
Variant ID	D			
Variant ID	D			

Variant ID	D			
------------	---	--	--	--

**GLA -based warnings**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
Warning ID	D			
Warning ID	D			
Warning ID	D			
Warning ID	D			
Warning ID	D			

	Name	Signature	Date
<b>Trainee</b>			
<b>Trainer</b>			
<b>Head of department</b>			

SUMMARY: statement competency test passed- valid until- (MM.YYYY)



Appendix 5  
 SOPeIT-87 Curator requirements and competency testing  
 GBA case curation

Version

1

**EXTERNAL DATABASES**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
OMIM -based queries	D			
PubMed- based queries	D			
GeneReviews- based queries	D			
Orphanet- based queries	D			
HPO- based queries	D			

**IN HOUSE SOFTWARE**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
Gepado handling	D			
UniDB handling	D			
CuRepo handling	D			

**DIGITAL PRODUCTS**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
CentoMD	D			
CentoLSD	D			

**Gaucher- based decision trees**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
Case ID	D			
Case ID	D			
Case ID	D			
Case ID	D			
Case ID	D			

<b>Gaucher -based warnings</b>

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
Warning ID	D			
Warning ID	D			
Warning ID	D			
Warning ID	D			
Warning ID	D			

	Name	Signature	Date
<b>Trainee</b>			
<b>Trainer</b>			
<b>Head of department</b>			

SUMMARY: statement competency test passed- valid until- (MM.YYYY)

Appendix 6  
 SOPeIT-87 Curator requirements and competency testing  
 GLA case curation

Version

1

**EXTERNAL DATABASES**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
OMIM -based queries	D			
PubMed- based queries	D			
GeneReviews- based queries	D			
Orphanet- based queries	D			
HPO- based queries	D			

**IN HOUSE SOFTWARE**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
Gepado handling	D			
UniDB handling	D			
CuRepo handling	D			

**DIGITAL PRODUCTS**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
CentoMD	D			
CentoLSD	D			

**Fabry- based decision trees**

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
Case ID	D			
Case ID	D			
Case ID	D			
Case ID	D			
Case ID	D			

Fabry -based warnings
-----------------------

Item	Expected results	Comment in case deviation from SOPs/ Criteria definitions	Trainee signature	Date/ Trainer signature
Warning ID	D			
Warning ID	D			
Warning ID	D			
Warning ID	D			
Warning ID	D			

	Name	Signature	Date
<b>Trainee</b>			
<b>Trainer</b>			
<b>Head of department</b>			

SUMMARY: statement competency test passed- valid until-

(MM.YYYY)