NeuroLab 2.0: Co-Designing a STEM+M Storyline – Santschi, et al.

Appendix B. Resources for Storyline Discovery and Incremental Model Building

Slide series showing embedded text prompts, web page screenshots, and expanded views of various page features and elements to help students navigate through authoritative databases to obtain information relevant to storyline discovery and incremental model building.

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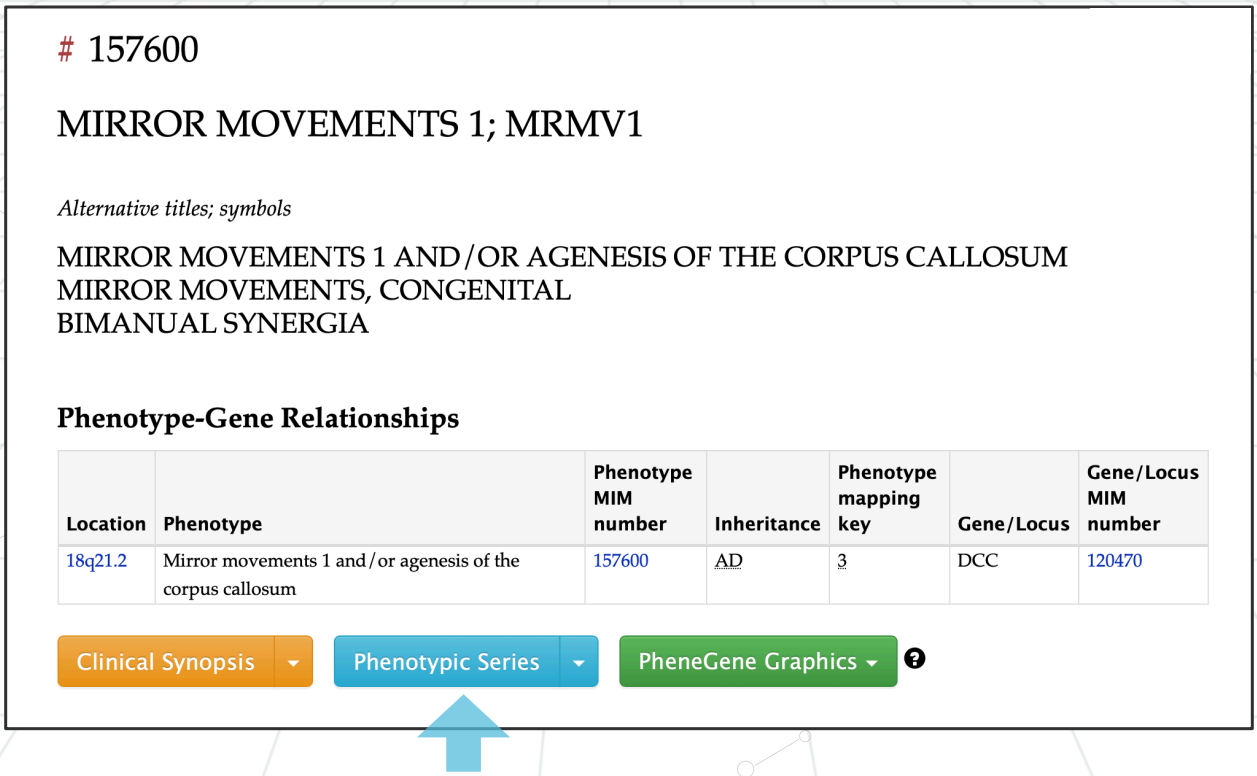
The MRMV1 data record contains a <Phenotypic Series> tab



157600

MIRROR MOVEMENTS 1; MRMV1

Alternative titles; symbols

MIRROR MOVEMENTS 1 AND/OR AGENESIS OF THE CORPUS CALLOSUM
MIRROR MOVEMENTS, CONGENITAL
BIMANUAL SYNERGIA

Phenotype-Gene Relationships

Location	Phenotype	Phenotype MIM number	Inheritance	Phenotype mapping key	Gene/Locus	Gene/Locus MIM number
18q21.2	Mirror movements 1 and/or agenesis of the corpus callosum	157600	AD	3	DCC	120470

Clinical Synopsis | Phenotypic Series | PheneGene Graphics

NeuroLab

A phenotypic series is a group of similar or identical phenotypes that arise from different gene mutations

157600

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Clinical Synopsis | Phenotypic Series | PheneGene Graphics

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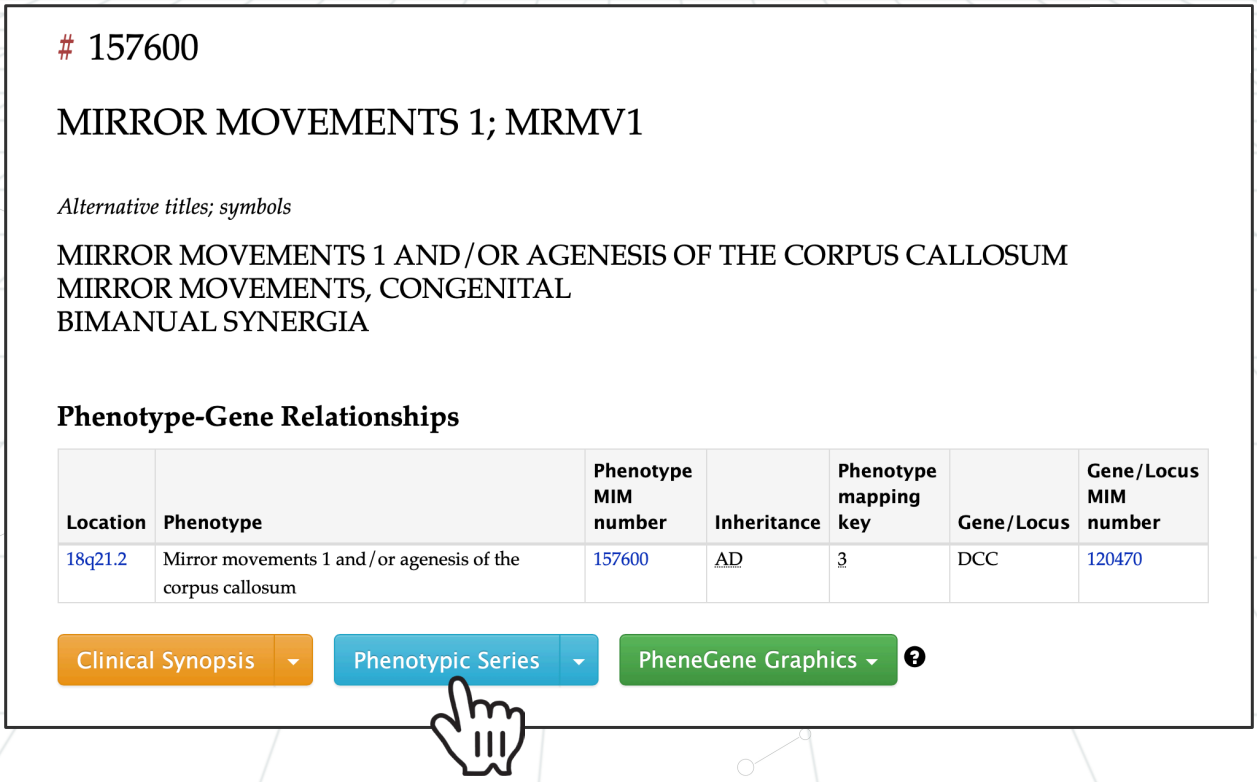
Click the tab to view a table that summarizes the phenotypic series



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Clinical Synopsis | Phenotypic Series | PheneGene Graphics

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How many similar phenotypes are displayed in the table? Which genes are associated with the phenotypes?

Phenotypic Series – PS157600

Mirror movements – PS157600 – 4 Entries

View corresponding clinical synopses as a table

Location	Phenotype	Inheritance	Phenotype mapping key	Phenotype MIM number	Gene/Locus	Gene/Locus MIM number
15q15.1	Mirror movements 2	AD	3	614508	RAD51	179617
17p13.1	Mirror movements 4	AD	3	618264	NTN1	601614
18q21.2	Mirror movements 1 and/or agenesis of the corpus callosum	AD	3	157600	DCC	120470
22q13.1	?Mirror movements 3	AR	3	616059	DNAL4	610565

View corresponding clinical synopses as a table

PheneGene Graphics

NeuroLab

The table displays 4 phenotypes corresponding to 4 separate genes

Phenotypic Series – PS157600

Mirror movements – PS157600 – 4 Entries

View corresponding clinical synopses as a table

Location	Phenotype	Inheritance	Phenotype mapping key	Phenotype MIM number	Gene/Locus	Gene/Locus MIM number
15q15.1	Mirror movements 2	AD	3	614508	RAD51	179617
17p13.1	Mirror movements 4	AD	3	618264	NTN1	601614
18q21.2	Mirror movements 1 and/or agenesis of the corpus callosum	AD	3	157600	DCC	120470
22q13.1	?Mirror movements 3	AR	3	616059	DNAL4	610565

View corresponding clinical synopses as a table

PheneGene Graphics

NeuroLab

Click the link at the bottom right to open a table that summarizes the clinical findings for each phenotype

Phenotypic Series – PS157600

Mirror movements – PS157600 – 4 Entries

View corresponding clinical synopses as a table

Location	Phenotype	Inheritance	Phenotype mapping key	Phenotype MIM number	Gene/Locus	Gene/Locus MIM number
15q15.1	Mirror movements 2	AD	3	614508	RAD51	179617
17p13.1	Mirror movements 4	AD	3	618264	NTN1	601614
18q21.2	Mirror movements 1 and/or agenesis of the corpus callosum	AD	3	157600	DCC	120470
22q13.1	?Mirror movements 3	AR	3	616059	DNAL4	610565

View corresponding clinical synopses as a table

PheneGene Graphics

NeuroLab

How would you describe the similarity of neurologic findings displayed in the shaded row of this table?

NUMBER	# 614508	# 618264	# 157600	# 616059
TITLE	MIRROR MOVEMENTS 2; MRMV2	MIRROR MOVEMENTS 4; MRMV4	MIRROR MOVEMENTS 1; MRMV1	MIRROR MOVEMENTS 3; MRMV3
INHERITANCE (in 4/4)	- Autosomal dominant	- Autosomal dominant	- Autosomal dominant	- Autosomal recessive
ABDOMEN (in 1/4) ▼				
		Gastrointestinal - Irritable bowel syndrome, constipation-dominant (in some patients)		
MUSCLE, SOFT TISSUES (in 2/4) ▼	- Pain or cramping during sustained manual activity		- Pain or cramping during sustained manual activity	
NEUROLOGIC (in 4/4) ▼	*Central Nervous System* - Mirror movements, involuntary, usually of the upper limb and hand - Difficulties in fine bimanual activities - Writing fatigability - Abnormal corticospinal tract decussation	*Central Nervous System* - Mirror movements, involuntary, usually of the upper limb and hand - Difficulties in fine bimanual activities - Abnormal corticospinal tract decussation	*Central Nervous System* - Intellectual disability, mild (in some patients) - Mirror movements, involuntary, usually of the upper limb and hand - Difficulties in fine bimanual activities - Writing fatigability - Abnormal corticospinal tract decussation - Agenesis of the corpus callosum (in some patients) - Absence of hippocampal commissure	*Central Nervous System* - Mirror movements, involuntary, affecting the hand and fingers - Difficulties in fine bimanual activities
MISCELLANEOUS (in 4/4) ▼	- Onset in infancy or early childhood - Disorder usually remains stable over time - Incomplete penetrance (50%)	- Onset in early childhood - Three unrelated families have been reported (last curated December 2018)	- Onset in infancy or early childhood - Disorder usually remains stable over time - Highly variable phenotype - Incomplete penetrance	- Onset in infancy or early childhood - One consanguineous Pakistani family has been reported (last curated October 2014)
MOLECULAR BASIS (in 4/4) ▼	- Caused by mutation in the homolog of S. cerevisiae RAD51 gene (RAD51, 179617.0003)	- Caused by mutation in the netrin 1 gene (NTN1, 601614.0001)	- Caused by mutation in the deleted in colorectal carcinoma gene (DCC, 120470.0001)	- Caused by mutation in the dynein, axonemal, light chain 4 gene (DNAL4, 610565.0001)

NeuroLab

Which phenotypes are associated with involuntary mirror movements?

NUMBER	# 614508	# 618264	# 157600	# 616059
TITLE	MIRROR MOVEMENTS 2; MRMV2	MIRROR MOVEMENTS 4; MRMV4	MIRROR MOVEMENTS 1; MRMV1	MIRROR MOVEMENTS 3; MRMV3
INHERITANCE (in 4/4)	- Autosomal dominant	- Autosomal dominant	- Autosomal dominant	- Autosomal recessive
ABDOMEN (in 1/4) ▼				
		Gastrointestinal - Irritable bowel syndrome, constipation-dominant (in some patients)		
MUSCLE, SOFT TISSUES (in 2/4) ▼	- Pain or cramping during sustained manual activity		- Pain or cramping during sustained manual activity	
NEUROLOGIC (in 4/4) ▼	*Central Nervous System* - Mirror movements, involuntary, usually of the upper limb and hand - Difficulties in fine bimanual activities - Writing fatigability - Abnormal corticospinal tract decussation	*Central Nervous System* - Mirror movements, involuntary, usually of the upper limb and hand - Difficulties in fine bimanual activities - Abnormal corticospinal tract decussation	*Central Nervous System* - Intellectual disability, mild (in some patients) - Mirror movements, involuntary, usually of the upper limb and hand - Difficulties in fine bimanual activities - Writing fatigability - Abnormal corticospinal tract decussation - Agenesis of the corpus callosum (in some patients) - Absence of hippocampal commissure	*Central Nervous System* - Mirror movements, involuntary, affecting the hand and fingers - Difficulties in fine bimanual activities
MISCELLANEOUS (in 4/4) ▼	- Onset in infancy or early childhood - Disorder usually remains stable over time - Incomplete penetrance (50%)	- Onset in early childhood - Three unrelated families have been reported (last curated December 2018)	- Onset in infancy or early childhood - Disorder usually remains stable over time - Highly variable phenotype - Incomplete penetrance	- Onset in infancy or early childhood - One consanguineous Pakistani family has been reported (last curated October 2014)
MOLECULAR BASIS (in 4/4) ▼	- Caused by mutation in the homolog of S. cerevisiae RAD51 gene (RAD51, 179617.0003)	- Caused by mutation in the netrin 1 gene (NTN1, 601614.0001)	- Caused by mutation in the deleted in colorectal carcinoma gene (DCC, 120470.0001)	- Caused by mutation in the dynein, axonemal, light chain 4 gene (DNAL4, 610565.0001)

NeuroLab

Which phenotypes are associated with difficulties in fine bimanual activities?

NUMBER	# 614508	# 618264	# 157600	# 616059
TITLE	MIRROR MOVEMENTS 2; MRMV2	MIRROR MOVEMENTS 4; MRMV4	MIRROR MOVEMENTS 1; MRMV1	MIRROR MOVEMENTS 3; MRMV3
INHERITANCE (in 4/4)	- Autosomal dominant	- Autosomal dominant	- Autosomal dominant	- Autosomal recessive
ABDOMEN (in 1/4) ▼				
		Gastrointestinal - Irritable bowel syndrome, constipation-dominant (in some patients)		
MUSCLE, SOFT TISSUES (in 2/4) ▼	- Pain or cramping during sustained manual activity		- Pain or cramping during sustained manual activity	
NEUROLOGIC (in 4/4) ▼	*Central Nervous System* - Mirror movements, involuntary, usually of the upper limb and hand - Difficulties in fine bimanual activities - Writing fatigability - Abnormal corticospinal tract decussation	*Central Nervous System* - Mirror movements, involuntary, usually of the upper limb and hand - Difficulties in fine bimanual activities - Abnormal corticospinal tract decussation	*Central Nervous System* - Intellectual disability, mild (in some patients) - Mirror movements, involuntary, usually of the upper limb and hand - Difficulties in fine bimanual activities - Writing fatigability - Abnormal corticospinal tract decussation - Agenesis of the corpus callosum (in some patients) - Absence of hippocampal commissure	*Central Nervous System* - Mirror movements, involuntary, affecting the hand and fingers - Difficulties in fine bimanual activities
MISCELLANEOUS (in 4/4) ▼	- Onset in infancy or early childhood - Disorder usually remains stable over time - Incomplete penetrance (50%)	- Onset in early childhood - Three unrelated families have been reported (last curated December 2018)	- Onset in infancy or early childhood - Disorder usually remains stable over time - Highly variable phenotype - Incomplete penetrance	- Onset in infancy or early childhood - One consanguineous Pakistani family has been reported (last curated October 2014)
MOLECULAR BASIS (in 4/4) ▼	- Caused by mutation in the homolog of S. cerevisiae RAD51 gene (RAD51, 179617.0003)	- Caused by mutation in the netrin 1 gene (NTN1, 601614.0001)	- Caused by mutation in the deleted in colorectal carcinoma gene (DCC, 120470.0001)	- Caused by mutation in the dynein, axonemal, light chain 4 gene (DNAL4, 610565.0001)

NeuroLab

Which phenotypes are associated with abnormal corticospinal tract crossing (decussation)?

NUMBER	# 614508	# 618264	# 157600	# 616059
TITLE	MIRROR MOVEMENTS 2; MRMV2	MIRROR MOVEMENTS 4; MRMV4	MIRROR MOVEMENTS 1; MRMV1	MIRROR MOVEMENTS 3; MRMV3
INHERITANCE (in 4/4)	- Autosomal dominant	- Autosomal dominant	- Autosomal dominant	- Autosomal recessive
ABDOMEN (in 1/4) ▼				
		Gastrointestinal - Irritable bowel syndrome, constipation-dominant (in some patients)		
MUSCLE, SOFT TISSUES (in 2/4) ▼	- Pain or cramping during sustained manual activity		- Pain or cramping during sustained manual activity	
NEUROLOGIC (in 4/4) ▼	*Central Nervous System* - Mirror movements, involuntary, usually of the upper limb and hand - Difficulties in fine bimanual activities - Writing fatigability - Abnormal corticospinal tract decussation	*Central Nervous System* - Mirror movements, involuntary, usually of the upper limb and hand - Difficulties in fine bimanual activities - Abnormal corticospinal tract decussation	*Central Nervous System* - Intellectual disability, mild (in some patients) - Mirror movements, involuntary, usually of the upper limb and hand - Difficulties in fine bimanual activities - Writing fatigability - Abnormal corticospinal tract decussation - Agenesis of the corpus callosum (in some patients) - Absence of hippocampal commissure	*Central Nervous System* - Mirror movements, involuntary, affecting the hand and fingers - Difficulties in fine bimanual activities
MISCELLANEOUS (in 4/4) ▼	- Onset in infancy or early childhood - Disorder usually remains stable over time - Incomplete penetrance (50%)	- Onset in early childhood - Three unrelated families have been reported (last curated December 2018)	- Onset in infancy or early childhood - Disorder usually remains stable over time - Highly variable phenotype - Incomplete penetrance	- Onset in infancy or early childhood - One consanguineous Pakistani family has been reported (last curated October 2014)
MOLECULAR BASIS (in 4/4) ▼	- Caused by mutation in the homolog of S. cerevisiae RAD51 gene (RAD51, 179617.0003)	- Caused by mutation in the netrin 1 gene (NTN1, 601614.0001)	- Caused by mutation in the deleted in colorectal carcinoma gene (DCC, 120470.0001)	- Caused by mutation in the dynein, axonemal, light chain 4 gene (DNAL4, 610565.0001)

NeuroLab

Which phenotypes are associated with abnormal corticospinal tract crossing?

NUMBER	# 614508	# 618264	# 157600	# 616059
TITLE	MIRROR MOVEMENTS 2; MRMV2	MIRROR MOVEMENTS 4; MRMV4	MIRROR MOVEMENTS 1; MRMV1	MIRROR MOVEMENTS 3; MRMV3
INHERITANCE (in 4/4)	- Autosomal dominant	- Autosomal dominant	- Autosomal dominant	- Autosomal recessive
ABDOMEN (in 1/4) ▼				
		Gastrointestinal - Irritable bowel syndrome, constipation-dominant (in some patients)		
MUSCLE, SOFT TISSUES (in 2/4) ▼	- Pain or cramping during sustained manual activity		- Pain or cramping during sustained manual activity	
NEUROLOGIC (in 4/4) ▼	*Central Nervous System* - Mirror movements, involuntary, usually of the upper limb and hand - Difficulties in fine bimanual activities - Writing fatigability - Abnormal corticospinal tract decussation	*Central Nervous System* - Mirror movements, involuntary, usually of the upper limb and hand - Difficulties in fine bimanual activities - Abnormal corticospinal tract decussation	*Central Nervous System* - Intellectual disability, mild (in some patients) - Mirror movements, involuntary, usually of the upper limb and hand - Difficulties in fine bimanual activities - Writing fatigability - Abnormal corticospinal tract decussation - Agenesis of the corpus callosum (in some patients) - Absence of hippocampal commissure	*Central Nervous System* - Mirror movements, involuntary, affecting the hand and fingers - Difficulties in fine bimanual activities
MISCELLANEOUS (in 4/4) ▼	- Onset in infancy or early childhood - Disorder usually remains stable over time - Incomplete penetrance (50%)	- Onset in early childhood - Three unrelated families have been reported (last curated December 2018)	- Onset in infancy or early childhood - Disorder usually remains stable over time - Highly variable phenotype - Incomplete penetrance	- Onset in infancy or early childhood - One consanguineous Pakistani family has been reported (last curated October 2014)
MOLECULAR BASIS (in 4/4) ▼	- Caused by mutation in the homolog of S. cerevisiae RAD51 gene (RAD51, 179617.0003)	- Caused by mutation in the netrin 1 gene (NTN1, 601614.0001)	- Caused by mutation in the deleted in colorectal carcinoma gene (DCC, 120470.0001)	- Caused by mutation in the dynein, axonemal, light chain 4 gene (DNAL4, 610565.0001)

NeuroLab

Explain how abnormal corticospinal tract decussation is relevant to the EMG, TMS, and fMRI data that you analyzed

NUMBER	# 614508	# 618264	# 157600	# 616059
TITLE	MIRROR MOVEMENTS 2; MRMV2	MIRROR MOVEMENTS 4; MRMV4	MIRROR MOVEMENTS 1; MRMV1	MIRROR MOVEMENTS 3; MRMV3
INHERITANCE (in 4/4)	- Autosomal dominant	- Autosomal dominant	- Autosomal dominant	- Autosomal recessive
ABDOMEN (in 1/4) ▼				
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MUSCLE, SOFT TISSUES (in 2/4) ▼	- Pain or cramping during sustained manual activity		- Pain or cramping during sustained manual activity	
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NeuroLab

Explain how abnormal corticospinal tract decussation is relevant to your models?

NUMBER	# 614508	# 618264	# 157600	# 616059
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ABDOMEN (in 1/4) ▼				
		Gastrointestinal - Irritable bowel syndrome, constipation-dominant (in some patients)		
MUSCLE, SOFT TISSUES (in 2/4) ▼	- Pain or cramping during sustained manual activity		- Pain or cramping during sustained manual activity	
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