

# Phenotype Annotation Tool and Ontologies at dictyBase

# Phenotype/PATO 2003

<b>\$</b>	Α	В	С	D	E	F	G	Н		J	K
1	locus	gen condition/mutan	environm. conditions	observable	attribute	qualifier	assay	values	unit	PMID	extra notes
2	sadA	KO/AX3	petridish HL5	substrate adhesion	ability	abolished/defective	attachment assay	<2	%WT	12499361	
3	sadA	н	petridish HL5	cytokinesis	ability	abolished/defective	DAPI staining	1-12	nuc/cell		52% have a
4	sadA	н	buffer, shaking	phagocytosis	ability	abolished/defective	uptake of latex beads	0	%WT		
5	sadA	11		vegetative growth	rate	increased	counting of cells	20	%		
6	sadA	н	petridish HL5	actin filament organization	cellular distribution	aberrant	Phalloidin staining				GO:7015
7	sadA	11	vegetative growth	cell motility	rate	increased	speed meas./cell tracking	6.3	um/min		WT 3.2
8	yelA	KO/AX4	solid substratum	mound formation	progression	abolished/defective	observation				
9	yelA	11	solid substratum	spore formation	progression	precocious	observation	6.00	hours	9254905	autonomou
10	yelA	11	bacterial lawn	spore	viability	reduced	plaque formation after det	2	%WT		
11	yelA	11	solid substratum	spore	morphology	aberrant	observation	round	rel WT		WT elongat
12	yelA	11	solid substratum	spore coat	morphology	aberrant	electron microscopy				
13	yelA	II .	solid substratum	spore specific expression	timing/progression	precocious	spiA Northern	4	hours		
	yelA	"	solid substratum	prestalk specific gene expr		reduced	ecm A Northern				
15	yelA	db yelA- tacB-or tacC	solid substratum	spore formation	ability	partially rescued	plaque formation after det	15	%	9543721	tagB/C-0%
	mlcE	KO/JH10		cytokinesis	ability	abolished/defective	DAPI staining	>10	nuc./cell		evtl. lethal
17	mlcE	11	cAMP gradient	chemotaxis	rate	reduced	Zigmond Chamber	5.8	um/min		(by 40-50%
18	mlcE	II .	solid substratum	aggregation (loose agg)	progression	delayed	observation	2-3	hours		
19	mlcE	"		stalk formation	size	reduced	observation	short +	thick (50%	?)	
20	mlcE	11	solid substratum	mound formation	progression	abolished/defective	observation	50.00	%WT		
21	mlcE	"	growth	actin-activated ATPase activ	level	reduced	enzyme activity assay with	<10	%WT		new GO?
22	mlcE	11	high salt		ability	increased	myosin solubilization in hi		%		WT >80%
23	mlcE	11	growth	MHC-MLCR binding	ability	reduced	IP whole cells	56	%WT		
24	m lcE	11	growth	MHC-MLCR binding	ability	reduced	IP purified myosin	10	%WT		
25	mlcE	"	development/solid substr	prespore specific gene exp	localization	aberrant	reporter gene assay, pspA				
26	ampA	KO/AX3	starvation in buffer suspe	cell-cell adhesion	ability	increased	agglutination assay	450	%Wt	11884033	
27	ampA	11	growth in suspension	tip formation	progression	delayed	observation	4.5	hours		
28	ampA	11	growth in suspension	fruiting body formation	efficiency	reduced	observation	some	cells		
29	ampA	n .	growth on bacteria 2d	mound formation	progression	delayed	observation	7-8	hours		
30	ampA	II .	growth on bacteria 2d	mound formation	progression	abolished/defective	observation				
31	ampA	11	growth on bacteria 3d	mound formation	progression	abolished/defective	observation				
32	ampA	11	growth on bacteria 3d	stream formation	progression	abolished/defective	observation				
33	ampA	II .	growth in suspension	substrate adhesion	ability	increased	protein retention assay aff	200	%WT		
34	ampA	II .	growth in suspension	substrate adhesion	ability	increased	protein retention assay aff	140	%WT		
35	ampA	II .	growth on bacteria 3d		ability	increased	protein retention assay aff		%WT		
36	ampA	11	growth on bacteria 3d	substrate adhesion	ability	increased	protein retention assay aff	260	%WT		
37	ampA	OE/AX3 6x	starvation in buffer suspe	cell-cell adhesion	ability	reduced	size meas, of cell clumps	50	%Wt		
38	ampA	OE/AX3 6x	growth in suspension	mound formation	progression	delayed	observation				
39	ampA	OE/AX3 6x	growth on bacteria 3d	fruiting body formation	progression	abolished/defective	observation				
40	ampA	KO/AX3			preference/efficiency	increased	counting of pspD cells at 1	200	%WT	11973270	

## **Phenotype Ontology**

- 420 Phenotype Terms
- 1780 Phenotype Annotations
- 470 Genes with Phenotype Annotation(s)

```
		☐── Classes

    Dicty Phenotypes

□←⑥ aberrant cellular process

    ⊕ ♠ aberrant cell adhesion

           □←  aberrant cell differentiation

    □ ← ① aberrant prespore cell differentiation

                    ← 
    abolished prespore cell differentiation
                    ← ① decreased prespore cell differentiation
                    ← 
 increased prespore cell differentiation

    ⊕ ⊕ aberrant spore germination

→ ↑ aberrant stalk cell differentiation

                ← ① increased sorting to prestalk region

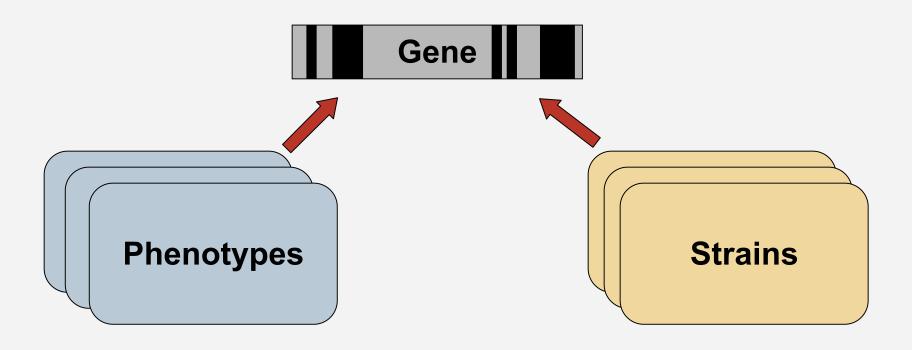
    ⊕ ← ⑥ aberrant signal transduction

    ⊕ ♠ aberrant response to stimulus

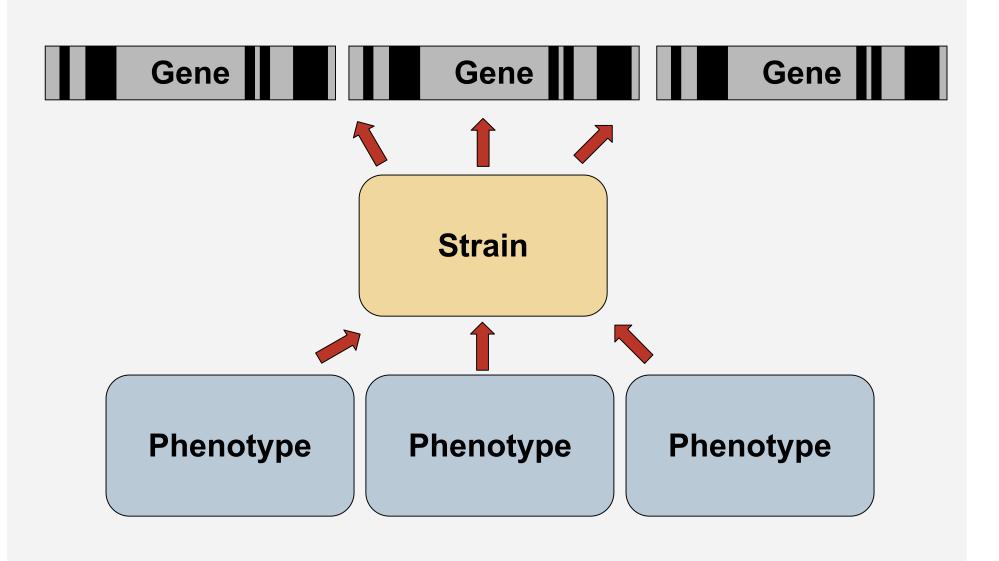
⊕ ← f) inviable

        ←  wild type
```

## Previous Phenotype Representation in dictyBase



# New Phenotype Representation in dictyBase



# Strain and Phenotype Display on the Gene Page

Cellular Component contractile ring (IDA), cytoplasm (IDA)

Expression

UCSD Expression Profile | BCM Expression Profile |



#### Strains and Phenotypes

mhkB-	increased myosin II assembly					
mhkA-/mhkB-	decreased growth rate, increased myosin II assembly					
mhkA-/mhkB-/mhkC-	decreased growth rate, increased myosin II assembly					
[oot15]:mbkB:ELAC	aborrent autokinosia					

#### Links

mhkB Researchers | GeneDB | STKE | Entrez Nucleotide | Entrez Protein | UniProt |

#### **Community Annotations**

mhkB Community Annotations Page

## Phenotype and Strain Details Page

### Phenotype and Strain Details for [act15]:mhkB:FLAG

Phenotype	Phenotype			Reference
aberrant cytokinesis		mhkB activity disrupts myo thus overexpression mimic cytokinesis defect Assay: nuclei count Environment: in suspens	cks the mhcA null	Rico & Egelhoff (2003) 'Myosin heavy chain kinase B participates in the regulation of myosin assembly into the cytoskeleton.' <i>J Cell Biochem</i> 88:521-32
Strain Details				
Strain Descriptor	[act15]:mhkB:FLAG		Strain ID	DBS0236665
Synonyms	MHCK-B OE		Systematic Name	DBS0236665
Description	FLAG epitope fused at the a overexpressed in wild type	amino-terminus of mhkB Genotype		axeA2, axeB2, axeC2, mhkB-FLAG [pTX-MKB2], neoR
Mutant Type	overexpression		Mutagenesis Method	Extrachromosomal
Strain Type	Axenic, Drug resistant		Depositor	
Parental Strain	AX2		Species	Dictyostelium discoideum
Plasmid	pTX-MKB2		Vector	
References	12532328			
Associated Genes	<u>mhkB</u>			

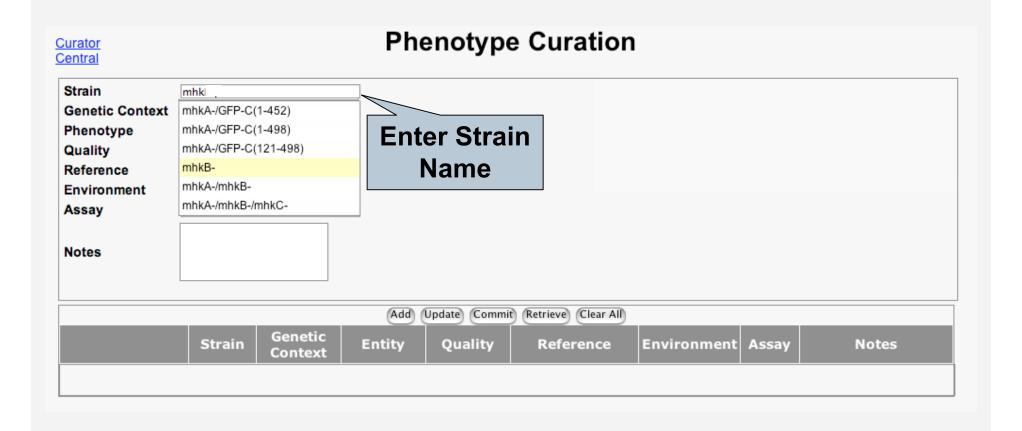
# Development of a New Phenotype Annotation Tool

- Developed in collaboration with NCBO
- Similar in functionality to Phenote
- Web-based tool that can be accessed from any computer

# Features of the New Phenotype Annotation Tool

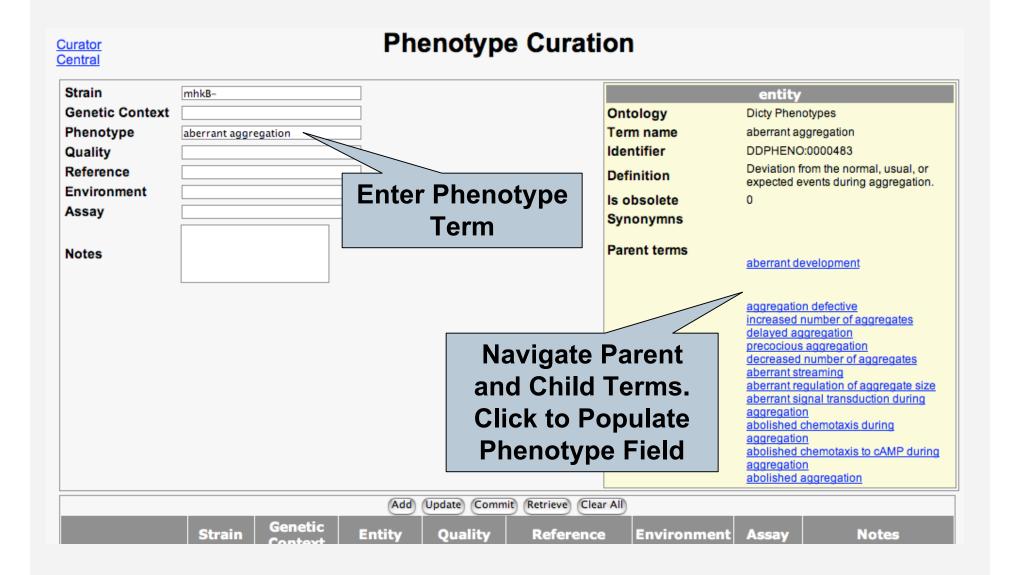
- Runs on top of the Chado database schema
- Uses terms from ontologies in .obo format for phenotype annotation
- Auto-completes ontology terms similar to Google Suggest
- Built using Web 2.0 technologies such as AJAX

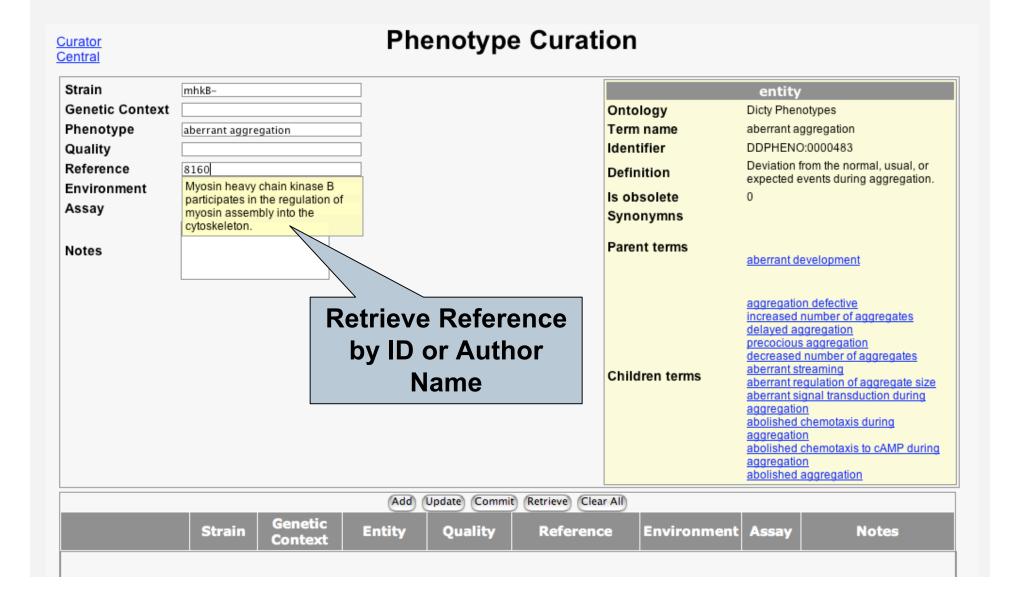
<u>Curator</u> <u>Central</u>			Ph	enotype	<b>Curation</b>	า		
Strain Genetic Context Phenotype Quality Reference Environment Assay Notes								
	0	Genetic		Update Commi				Notes
	Strain	Context	Entity	Quality	Reference	Environment	Assay	Notes



<u>Curator</u> <u>Central</u>			Ph	enotype	Curat	ion		
Strain Genetic Context Phenotype Quality Reference Environment Assay Notes	mhkB-					DBS ID Strain ID Strain name Associated Genes	Strain  DBS0235328  371  mhkB- mhkB	
			Add	Update Commit	Retrieve Clea	ar All)		
	Strain	Genetic Context	Entity	Quality	Refer	Environment	t Assay	Notes
						Retrieve E Phenot Annotat	ype	

train	mhkB-						Strain	
Senetic Context Phenotype Quality Reference Environment Assay					Str Str	SS ID rain ID rain name sociated Genes	DBS02353 371 mhkB- mhkB	328
otes								
			Add	Update Comm	nit Retrieve Clear All	)		
	Strain	Genetic Context	(Add) Entity	Update Comm	nit Retrieve Clear All	Environment	Assay	Notes
<b>X</b> €DII 造	Strain mhkB-					Environment  n undefined	Assay biochemical assay	Notes into Triton-resistant cytoskeletal fractions
¥ ŒĐIT ₽		Context	Entity increased myosin II	Quality	Reference  Rico & Egelhoff (2003) 'Myosin heavy chain kinase B participates in the regulation of myosin assembly into the cytoskeleton.' J Cei	Environment  n undefined	biochemical	into Triton-resistant





<u>Curator</u> <u>Central</u>			Ph	enotyp	e Curatio	n		
Strain	mhkB-						Referen	ce
<b>Genetic Context</b>					R	eference no	8160	
Phenotype	aberrant aggr	egation			Pi	ubmed ID	12532328	
Quality								elhoff (2003) 'Myosin heavy
Reference	Myosin heavy	chain kinase B pa	artici		C	itation		se B participates in the of myosin assembly into the
Environment								on.' J Cell Biochem 88:521-32
Assay							dictyBase paper	PubMed Access Full Text
Notes								
			Add	Update Comm	nit Retrieve Clear A	<u> </u>		
	Strain	Genetic Context	Entity	Quality	Reference	Environment	Assay	Notes
X ENT P	mhkB-	undefined	increased myosin II assembly	undefined	Rico & Egelhoff (2003 'Myosin heavy chain kinase B participates the regulation of myosin assembly into the cytoskeleton.' J Co Biochem 88:521-32	in undefined	biochemical assay	into Triton-resistant cytoskeletal fractions

Curator Central			Ph	enotyp	e Curati	on					
Strain	mhkB-							assay			
Genetic Context						Ontolo	ду	Dictyostelium Assay			
Phenotype	aberrant aggr	regation				Term na	ame	light micros	scopy		
Quality						ldentifi	er	DDASSAY	:0000016		
Reference	Myosin heavy	chain kinase B pa	artici			Definiti	on				
Environment	on bacterial p	terial plates					lete	0			
Assay	light microsco	ору				Synonymns					
Notes					Parent terms  Dictyostelium Assay  Children terms						
	Strain	Genetic Context	(Add) Entity	Update Comm	Reference		vironment	Assay	Notes		
X EDIT E	mhkB-	undefined	increase myosin I assembl		nnotatio List tne cytoskeleton. J Blochem 88:521-32	Cell	efined	biochemical assay	into Triton-resistant cytoskeletal fractions		

<u>Curator</u> Central			Ph	enotyp	e Curati	on		
Strain	mhkB-						assay	,
Genetic Context						Ontology		ium Assay
Phenotype	aberrant agg	regation				Term name	light micro	scopy
Quality						Identifier	DDASSAY	r:0000016
Reference	Myosin heavy	chain kinase B p	artici			Definition		
Environment	on bacterial p	lates				Is obsolete	0	
Assay	light microsc	ору				Synonymns		
Notes						Parent terms	ium Assay	
Notes						Children terms		
			Add	Update Com	mit Retrieve Clea	r All)		
	Strain	Genetic Context	Entity	Quality	ference	Environme	ent Assay	Notes
X (EDIT) Pa	mhkB-		aberrant aggregation		kinase B pa the regulation myos the c	Commi	t	
X DIT	mhkB- undefined incr			undefined	Rico 'Myo' kinas the re myosin assembly i the cytoskeleton.' J Biochem 88:521-3	J Cell		into Triton-resistant cytoskeletal fractions

Cu	rato
Ce	ntral

Strain	mhkB-	assay	
Genetic Context		Ontology Dictyostelium Assay	
Phenotype	aberrant aggregation	Term name light microscopy	
Quality		Identifier DDASSAY:0000016	
Reference	Myosin heavy chain kinase B partici	Definition	
Environment	on bacterial plates	Is obsolete 0	
Assay	light microscopy	Synonymns	
Notos		Parent terms  Dictyostelium Assay	
Notes		Children terms	

			Add	Update Comn	nit Retrieve Clear All			
	Strain	Genetic Context	Entity	Quality	Reference	Environment	Assay	Notes
× EDIT E	mhkB-		aberrant aggregation		Myosin heavy chain kinase B participates in the regulation of myosin assembly into the cytoskeleton.	on bacterial plates	light microscopy	
¥ ŒĐIT ₽	mhkB-	undefined	increased myosin II assembly	undefined	Rico & Egelhoff (2003) 'Myosin heavy chain kinase B participates in the regulation of myosin assembly into the cytoskeleton.' J Cell Biochem 88:521-32	undefined	biochemical assay	into Triton-resistant cytoskeletal fractions
MESSAGE								
Successfully co	mmitted t	he data						

# Resulting Phenotype Display on the Gene Page

Cellular Component contractile ring (IDA), cytoplasm (IDA)

Expression

UCSD Expression Profile | BCM Expression Profile |



#### Strains and Phenotypes

mhkB- increased myosin II assembly aberrant aggregation

mhkA-/mhkB- decreased growth rate, increased myosin II assembly

mhkA-/mhkB-/mhkC- decreased growth rate, increased myosin II assembly

[act15]:mhkB:FLAG aberrant cytokinesis

#### Links

mhkB Researchers | GeneDB | STKE | Entrez Nucleotide | Entrez Protein | UniProt |

#### **Community Annotations**

mhkB Community Annotations Page

Cu	rato
Ce	ntral

Strain	mhkB-		assay
Genetic Context		Ontology	Dictyostelium Assay
Phenotype	aberrant aggregation	Term name	light microscopy
Quality		<u>Identifier</u>	DDASSAY:0000016
Reference	Myosin heavy chain kinase B partici	Definition	
Environment	on bacterial plates	Is obsolete	0
Assay	light microscopy	Synonymns	
Notes		Parent terms Children terms	<u>Dictyostelium Assay</u>

Add Update Commit Retrieve Clear All								
	Strain	Genetic Context	Entity	Quality	Reference	Environment	Assay	Notes
X EDIT	mhkB- aberrant aggregation			Myosin heavy chain kinase B participates in the regulation of myosin assembly into	on bacterial plates	light microscopy		
	Edit Existing				the cytoskeleton.  Rico & Egelhoff (2003)			
Phenotype  MnkB- Appotation		'Myosin heavy chain kinase B participates in the regulation of myosin assembly into the cytoskeleton.' J Cell	undefined	biochemical assay	into Triton-resistant cytoskeletal fractions			
					Biochem 88:521-32			
MESSAGE								
Successfully committed the data								

Cu	rator
Ce	ntral

mhkB-	assay
ontext	Ontology Dictyostelium Assay
aberrant aggregation	Term name light microscopy
	Identifier DDASSAY:0000016
Myosin heavy chain kinase B partici	Definition
on bacterial plates	Is obsolete 0
light microscopy	Synonymns
	Parent terms  Dictyostelium Assay
	Children terms

(Add) (Update) (Commit) (Retrieve) (Clear All)								
	Strain	Genetic Context	Entity	Quality	Reference	Environment	Assay	Notes
X EDIT E	mhkB-		aberrant aggregation		Myosin heavy chain kinase B participates in the regulation of myosin assembly into the cytoskeleton.	on nacterial histes	light microscopy	
X EDIT E	mhkB-	undefined	increased myosin II assembly	undefined	Rico & Egelhoff (2003) 'Myosin heavy chain kinase B participates in the regulation of myosin assembly into the cytoskeleton.' J Cell Biochem 88:521-32	undefined	biochemical assay	into Triton-resistant cytoskeletal fractions

Cu	ra	tor
Ce	nti	ral

Strain	mhkB-		environment
<b>Genetic Context</b>		Ontology	environment
Phenotype	aberrant aggregation	Term name	on solid surfaces
Quality		Identifier	DDENVIR:0000008
Reference	Myosin heavy chain kinase B partici	Definition	
<b>Environment</b>	on solid surfaces	Is obsolete	0
Assay	light microscopy	Synonymns	
Notes		Parent terms	environmental condition
		Children terms	

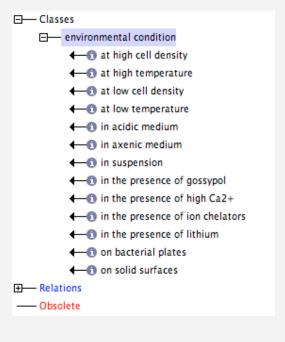
Add (Update Commit Retrieve Clear All)								
	Strain	Genetic Context	Entity	Quality	Reference	Environment	Assay	Notes
X EDIT 🖺	mhkB-		aberrant aggregation		Myosin heavy chain kinase B participates in the regulation of myosin assembly into the cytoskeleton.	on solid surfaces	light licroscopy	
🗶 (EDIT) 🖺	mhkB-	undefined	increased myosin II assembly	undefined	Rico & Egelhoff (2003) 'Myosin heavy chain kinase B participates in the regulation of myosin assembly into the cytoskeleton.' J Cell Biochem 88:521-32	undefined	biochemical assay	into Triton-resistant cytoskeletal fractions

## **Additional Ontologies**

### Assay

### F- Classes Dictyostelium Assay ← agglutination assay ← attachment to glass ← ① attachment to plastic ← biochemical assay ← ① cell count ← ① developmental time course ← ① electron microscopy ← ① germination assay ← immunofluorescence ← ① light microscopy ← measurement of surface area ← ① motility assay ← Northern blot ← nuclei count ← ① observation of a marker gene ← f) phalloidin staining ← plaque count ← size measurement ← ① uptake of latex beads ← ① uptake of yeast particles ← ① viability assay → Relations — Obsolete

### Environment



### **Future Directions**

- Dissection of Dicty phenotype ontology terms to fit PATO Entity-Quality model
- Collaboration with other groups on PATO and phenotype-related ontologies (e.g., genetic context, environmental conditions)
- Provide phenotype annotation tool to GMOD as open-source software

# dictyBase Acknowledgements

## **Principal Investigators**

- Rex Chisholm
- Warren Kibbe

## **Software Developers**

- Eric Just
- Sohel Merchant

### **Curators**

- Petra Fey
- Pascale Gaudet
- Karen Pilcher

### **Funding**

- NIH
- GO Consortium