

Casimir
“User Interactions”
WP 8 , Participant 3

Klaus Schughart
Braunschweig 2.7.08

Agenda

WP8 meeting

- Develop phenotype-driven - use case no. 2

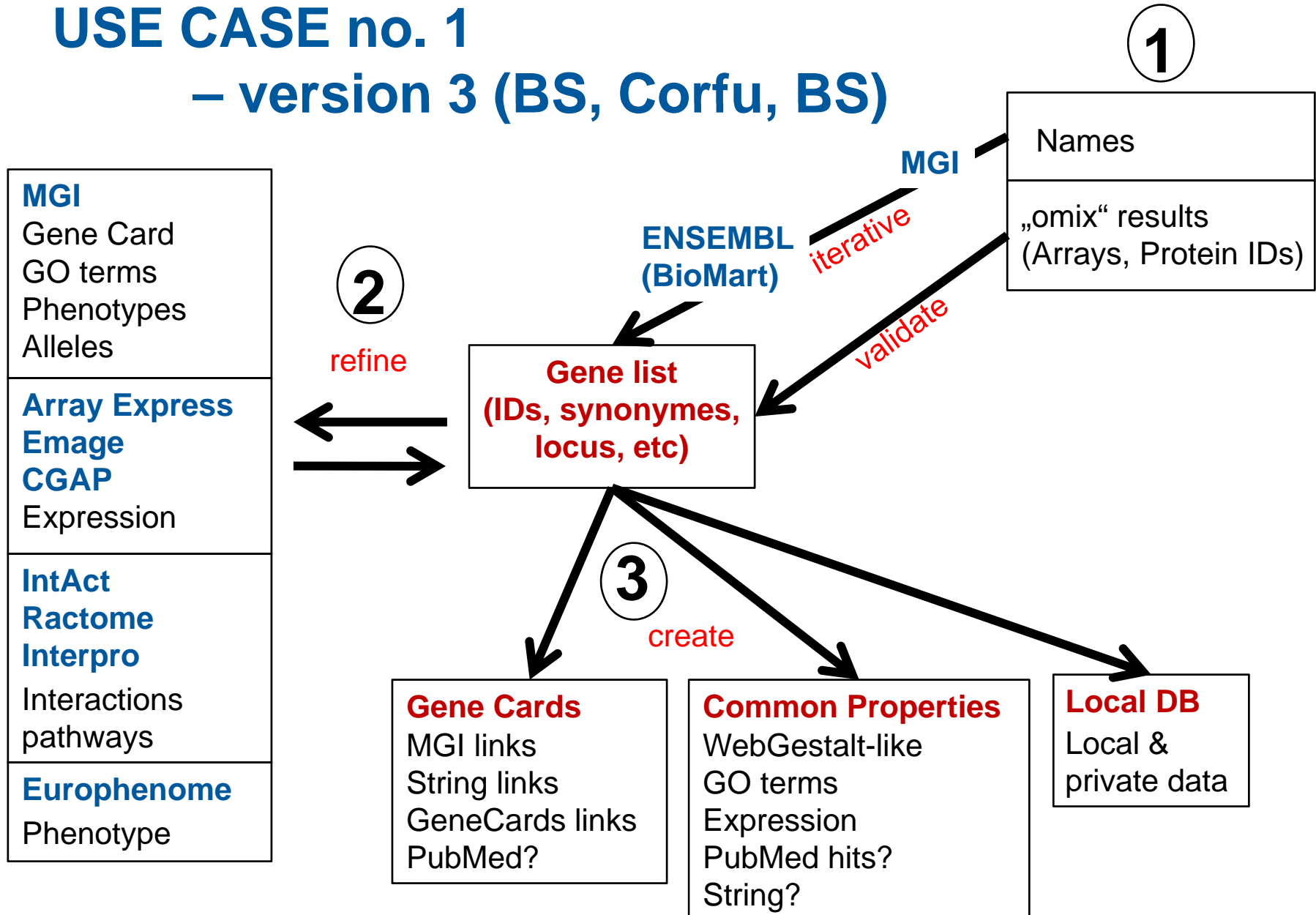
WP5 & WP8 joint meetings

- WP8 present current status of use case no. 1
K. Schughart & M. Grunberger
- WP5 present use case no. 2 (Taverna & Molgenis)
MD. Smedley
- Identify problems to solve for use case no. 1

- Present dbGG
M. Swertz
- Review use case no 2
- Next milestones and beyond ...

USE CASE no. 1

– version 3 (BS, Corfu, BS)



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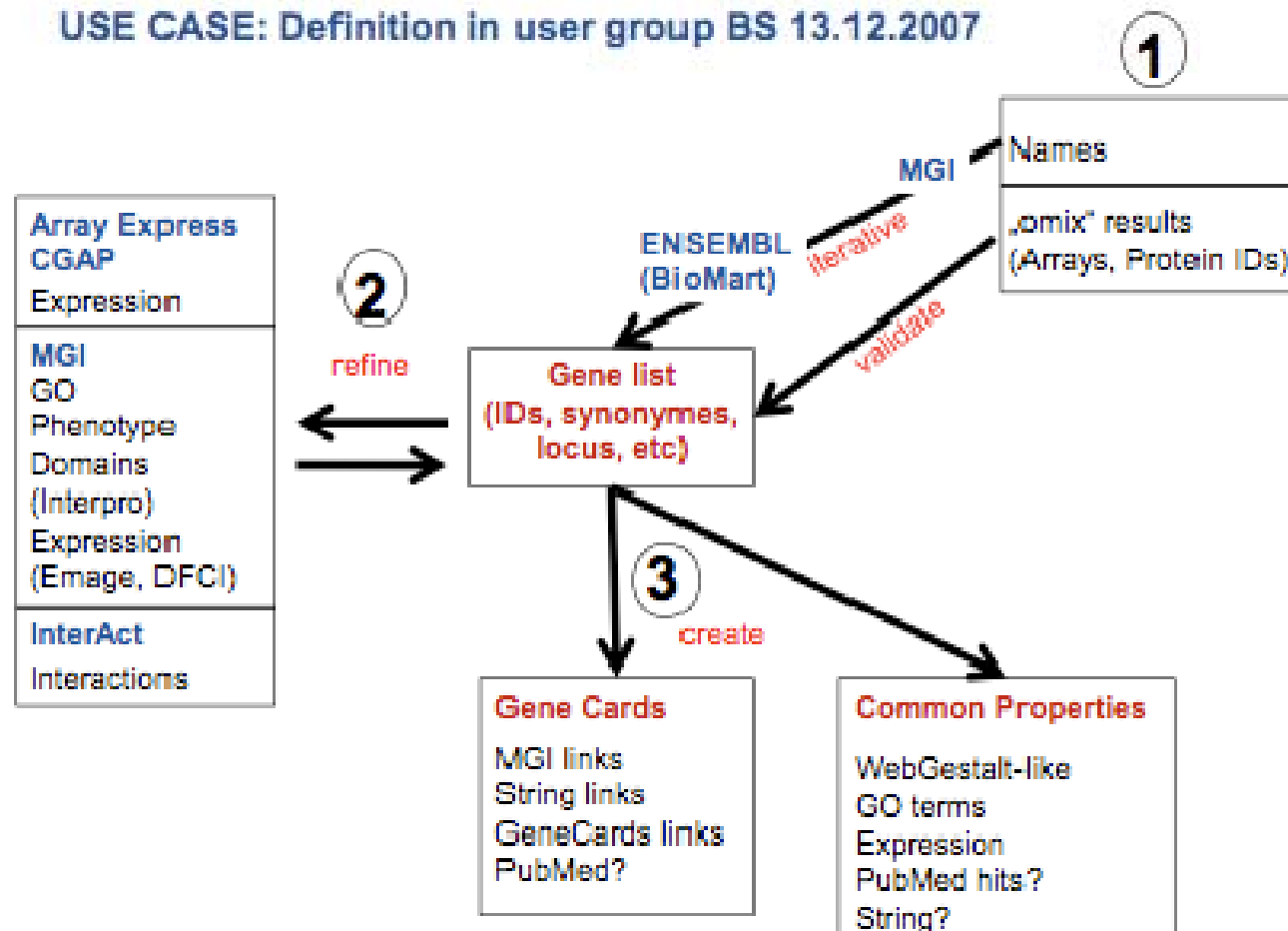
Use-case Development

Current Status
And
Planned Features

Presented by
Michael Grunberger

Use-case: Specification

USE CASE: Definition in user group BS 13.12.2007



Use-case: Alpha version

Initial input and output selection

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http://www.casimir.org.uk/usecase1/alpha_version/

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Gene Shopping Carts:

2008-04-15	🔍	+	🗑️
2008-04-15	🔍	+	🗑️
2008-04-21	🔍	+	🗑️
2008-05-22	🔍	+	🗑️
2008-06-23	🔍	+	🗑️
2008-06-30	🔍	+	🗑️
2008-06-30	🔍	+	🗑️

WP8 - Use Case 1: Web Service Gene Shopping Cart

Gene symbols

Enter list: (1 symbol per line)

pax3
igf2

Or upload a file:

Browse...

File Type:

tab delimited comma separated

symbols to search are in column number: 1

(all symbols must be in a single column)

Search the following data sources:

MGI Ensembl

Output

Gene Attributes:

Nomenclature Genome Location Ensembl ID Entrez Gene ID VEGA ID

Additional Information:

MGI ID MGI Allele ID

Marker RefSeq ID Gene Ontology (GO) Mammalian Phenotype (MP)

Search Reset

Done 0:6


Gene selection

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- 2008-06-23
- 2008-06-30

WP8 - Use Case 1: Web Service Gene Shopping Cart

Add to cart (Select all)	Input Symbol	Marker Name	Marker Type	Ensembl Gene ID	GO Term ID	Marker Synonym
<input type="checkbox"/>	pax3 (searched marker_symbol)	paired box gene 3	Gene	ENSMUSG 00000004872	GO:0007507, GO:0007517, GO:0006355, GO:0000122, GO:0009887, GO:0006350, GO:0045944, GO:0001755, GO:0048066, GO:0001843, GO:0043565, GO:0045449, GO:0021915, GO:0005634, GO:0007275, GO:0003677, GO:0005515, GO:0005667, GO:0003700, GO:0016563, GO:0003682, GO:0003705, GO:0016566, GO:0008283, GO:0016477, GO:0008284,	Pax-3,
<input type="checkbox"/>	igf2 (searched marker_symbol)	insulin-like growth	Gene	ENSMUSG 00000048583	GO:0005576, GO:0008083, GO:0008283, GO:0005179, GO:0018445, GO:0005615, GO:0005515, GO:0005159, GO:0009887,	Mpr, M6pr, Peg2, Igf-2, Igf-II,

Add selected genes to cart Reset

Download selected gene data as [Excel spreadsheet](#) / [CSV file](#).

New gene cart creation and cart details

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http://www.casimir.org.uk/usecase1/alpha_version/index.php?stage=4&cartid=78

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Search :

WP8 - Use Case 1: Web Service Gene Shopping Cart

Cart: 2008-06-30

Created on: 2008-06-30 08:06:47 Last modified on:

pax3

No.	Symbol	Marker Name	Marker Type	Ensembl Gene ID	GO Term ID
1	pax3	paired box gene 3	Gene	ENSMUSG0000004872	GO:0007507,GO:0007517,GO:0006355,GO:0000122,GO:0009887,GO:0006350,GO:0045944,GO:0001755,GO:004

igf2

No.	Symbol	Marker Name	Marker Type	Ensembl Gene ID	GO Term ID
2	igf2	insulin-like growth	Gene	ENSMUSG00000048583	GO:0005576,GO:0008083,GO:0008283,GO:0005179,GO:0018445,GO:0005615,GO:0005515,GO:0005159,GO:

Download these genes as [Excel spreadsheet](#) / [CSV file](#).

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Gene Shopping Carts:

- 2008-04-15
- 2008-04-15
- 2008-04-21
- 2008-05-22
- 2008-06-23
- 2008-06-30
- 2008-06-30
- 2008-06-30

Gene details and more web service options

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http://www.casimir.org.uk/usecase1/alpha_version/index.php?stage=5&cartcontentsid=103&cartid=7

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WP8 - Use Case 1: Web Service Gene Shopping Cart

pax3

No.	Symbol	Marker Name	Marker Type	Ensembl Gene ID	GO Term ID
1	pax3	paired box gene 3	Gene	ENSMUSG0000004872	GO:0007507,GO:0007517,GO:0006355,GO:0000122,GO:0009887,GO:0006350,GO:0045944,GO:0001755,GO:004

Search : [Load more data from Intact](#)

[Download these genes as Excel spreadsheet / CSV file.](#)

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Gene Shopping Carts:

- 2008-04-15
- 2008-04-15
- 2008-04-21
- 2008-05-22
- 2008-06-23

Done 0:6


Example web service: Intact

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- 2008-06-23
- 2008-06-30

WP8 - Use Case 1: Web Service Gene Shopping Cart

accession number molecule a	accession number molecule b	names molecule a	names molecule b	interaction details
P56693 & EBI-1167533	P23760 & EBI-1167564	SOX10	PAX3	EBI-1167594 & EBI-1167721
P23760 & EBI-1167564	P20265 & EBI-1167176	PAX3	POU3F2	EBI-1167600 & EBI-1167739
P24610 & EBI-1208116	P55036 & EBI-359318	Pax3	PSMD4	EBI-1380085 & EBI-1380067 & EBI-1379723
P54727 & EBI-954531	P24610 & EBI-1208116	RAD23B	Pax3	EBI-1380343 & EBI-1380351 & EBI-1380361 & EBI-1379990
P24610 & EBI-1208116	P28749 & EBI-971402	Pax3	RBL1	EBI-1208339 & EBI-1208574 & EBI-1208245
Q08999 & EBI-971439	P24610 & EBI-1208116	RBL2	Pax3	EBI-1208346 & EBI-1208144 & EBI-1208259
P24610 & EBI-1208116	P13405 & EBI-971782	Pax3	Rb1	EBI-1208206 & EBI-1208294 & EBI-1208454
P62988 & EBI-413034	P24610 & EBI-1208116	RPS27A	Pax3	EBI-1379672 & EBI-1380062

Add these fields to the cart

Done 0:6

Use-case: Beta version

Planned features

- **Make the cart more user-friendly:**
 - Modify/ delete functions for carts and genes
 - Change the initial order of input/ output fields
 - Make various fields links to details on web pages (e.g. MGI ID, Ensembl ID)
 - Make the initial search wider and allow wild-card searches
 - Load more human readable data and display additionally to ID fields (e.g. GO, MP)
- **Add more services:**
 - Intact, Eurexpress, GNF, SNP, Eimage
 - Other DB's from original use-case specification don't have web services yet

Use-case: Beta version

Current status

- Demo Biomart now running on Casimir servers
- Ontology server for GO+MP data now running on Casimir servers
- New backend implemented. Rewrite was needed to store data in different formats and additional information for each field.
- The new system is more flexible and any web service/ field and data can now be added in future.
- Let users add their own services ?

- ToDo:
 - Finish new user interface
 - Load data from additional web services

Use-case: Beta version

What next?

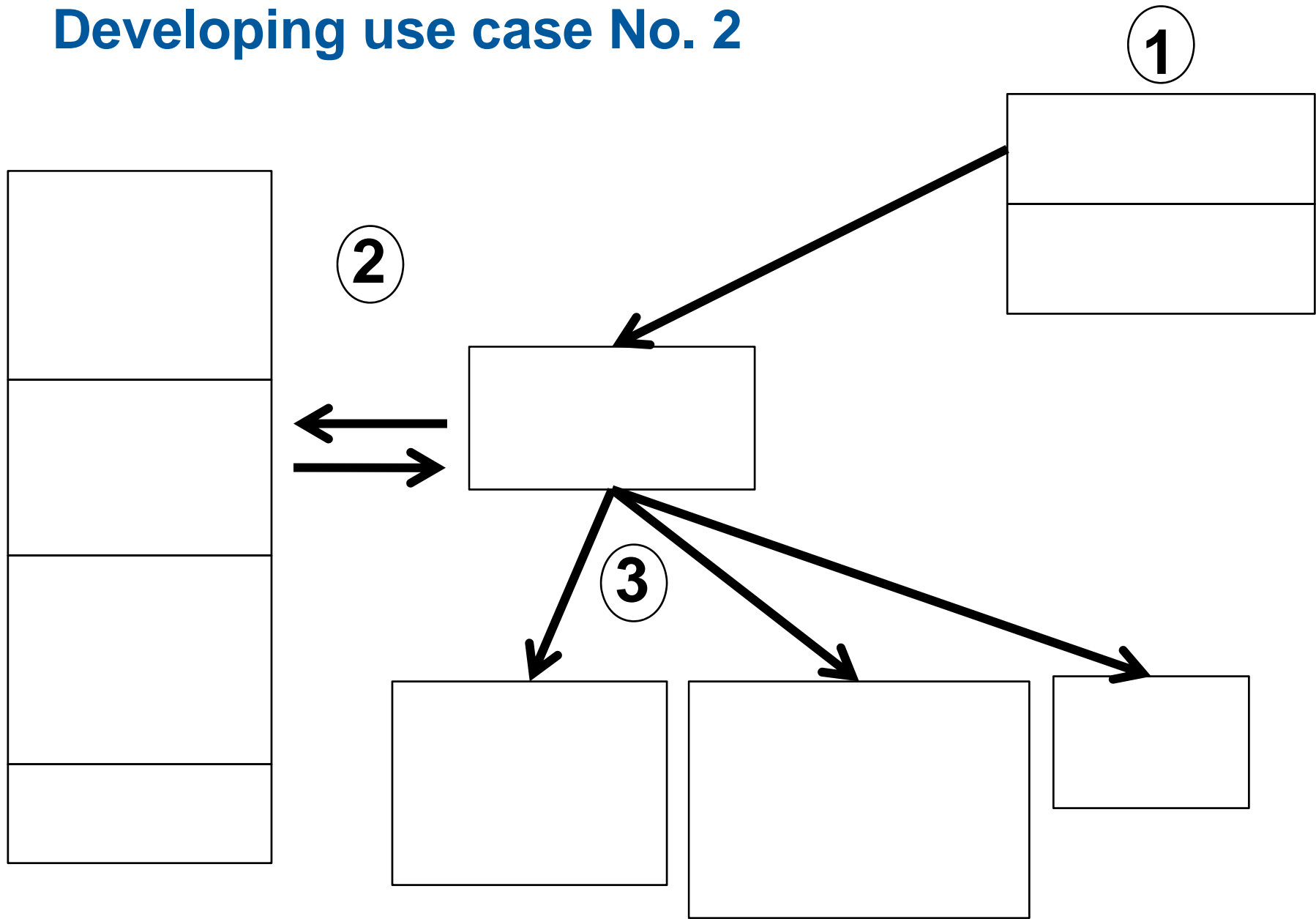
- September meeting: New version should be ready for internal use and discussions
- December meeting: Fully functional version should be ready for presentation and use

Use-case: Beta version

Questions

- Should more services be added that we have missed?
- Can we create more services via Biomart/ Molgenis?
- Which other features should we add now or in future?
- Any other suggestions?

Developing use case No. 2



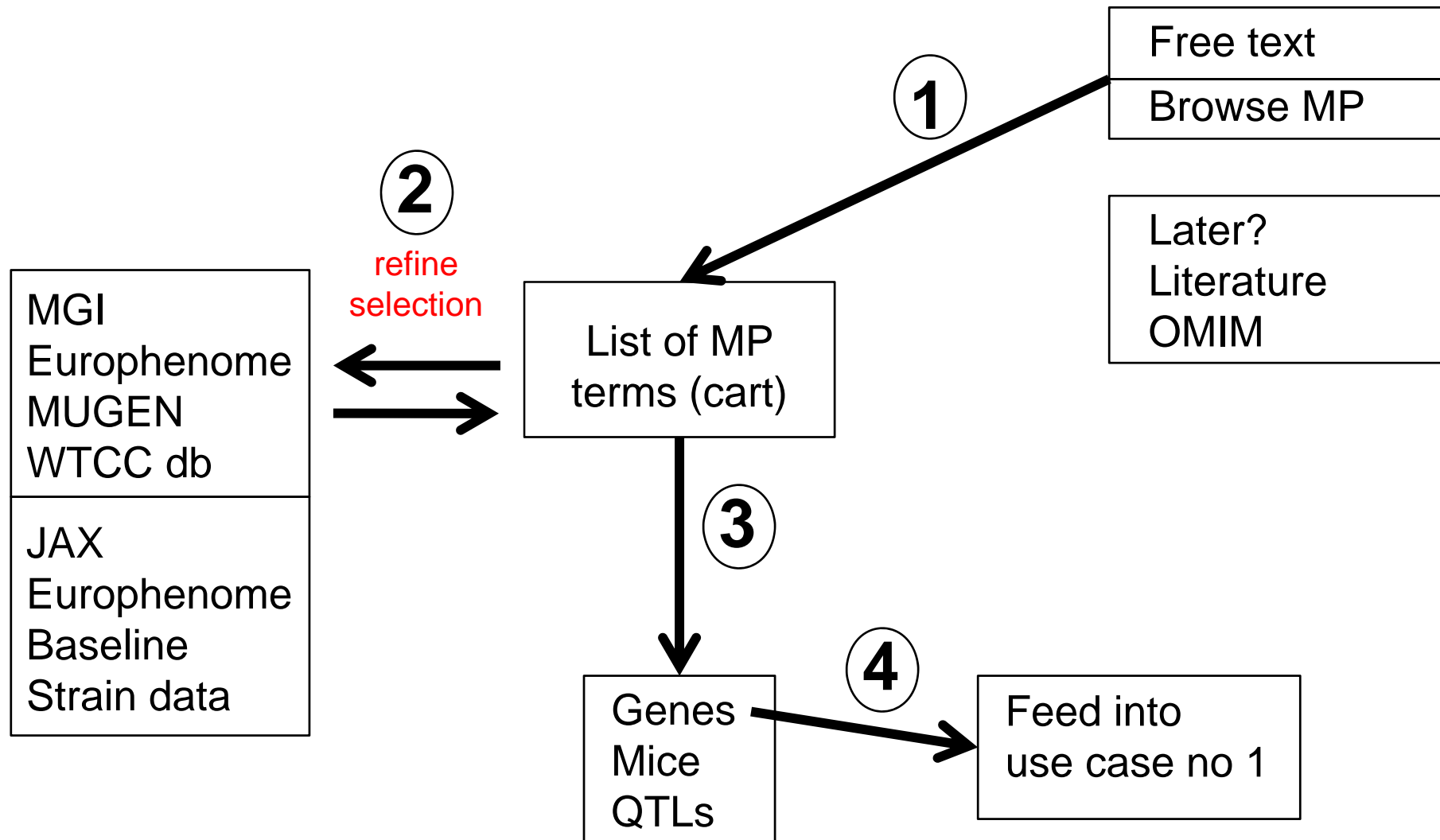
Phenotype-driven use case

- Objective
phrase a question/query, involve multiple databases, visualize results
- Goals:
define entry point and result
identify databases to be used
phrase queries and individual steps

Phenotype-driven use case

- Steps
 - >> define start point
 - >> define end point
- How are you performing phenotype-centric searches
- Which databases are you using?
- How would you like to have the results presented?

USE CASE no. 2 – phenotype driven



Use case no 2

- As entry point, free text and browsing / selecting MP database terms should be provided, free text will suggest closest MP terms (ways to use literature searches may be considered later – feasibility to be investigated)
- This step can be done multiple times to collect a list of MP terms in a shopping cart fashion
- The list of MP terms will then be used to search databases, these database need to be using M terms for phenotype descriptions
- Currently, databases using MP terms are:
 - MGI
 - MUGEN
 - Euromphenome (to be set up soon in MP-searchable fashion)
 - WTC database for QTL mapping of heterogenous stock (to be checked)
- Hits for the MP terms in baseline strain databases should be established (JAX, Euromphenome)
- The result of this search will be a list of genes, mice and QTLs
- The genes list can then be usable as input list for use case no 1

More ideas & comments

- **The key to phenotype searches across databases is a standard ontology**
- Literature search
check if EBI could index the literature with MP terms (Damian)
or: search only literature deposited in MGI, these are linked to genes which are linked to MP term, in this way generate MP list and then validate and select
- Integration is rather needed in the second step than at the beginning
- list of phenotypes, mutants strains, and then ...
- OMIM may be used by going via text searches, identify the human gene, identify the orthologous mouse gene (assuming that orthologous human genes result in a similar phenotype in mice & human)
- OMIM: one may also link human and mouse via genomic regions (an analysis in humans identifies a genomic region, this could be linked to mouse syntenic regions – and vice versa)
- RIKEN could be used if there were a link via MP terms
- Eumodic, Eucom & ICS (IGBMC) data will go into Europhenome, these data will be associated with MP terms
- Human phenotype in OMIM to human genes to mouse genes to pathways, may provide new genes that could be associated with human phenotype, could be checked in GWA studies

Next steps

- working up the milestones

- Implement use case no 1
until midterm meeting (17.9.08), ?integrate private data
- Casimir users to test
- Implement use case no 2
until December meeting (2.12.08)
- Add use case no 3
until December meeting, integrate into use case no 1
- Casimir users to test
- **Publish** use case in Spring 2009
- Get user feed back
- Analyze and report to other WPs

- Small “use cases” BiMart bookmarks with user friendly interphase

Time table

Task	Month
Develop use cases	
User group meetings	
Publish use case	
Collate feed-back from community	11-15
User group meetings, adjust user case according to feed-back & finalize	15-20
Report use case	22

Next steps

- future perspectives, add on's

- Suggest ways to integrate mouse and human databases and simple cases to realize
- Design a (simple) use case for clinicians a la “Mouggles” (input phenotype as free text and genomic location or gene; output a hyperlink text with link to mouse gene(s) or syntenic region with associated phenotype)
- Collect a set of future challenging user queries for bioinformaticians to see where the future needs are, see C. Lengger cases
- Queries that require a quantitative analysis
get all strains with a glucose level of 10% above mean, above a p-threshold (> 95% percentile after permutation)
- Query images & large datasets
(MRI, FACS, microarrays, microscopic images)
- iMouse as a one-stop-shop?
Web Portal at the most