

# **Genotype-To-Phenotype Databases - A Holistic Solution**

**Experiences of an European Union FP7 project**

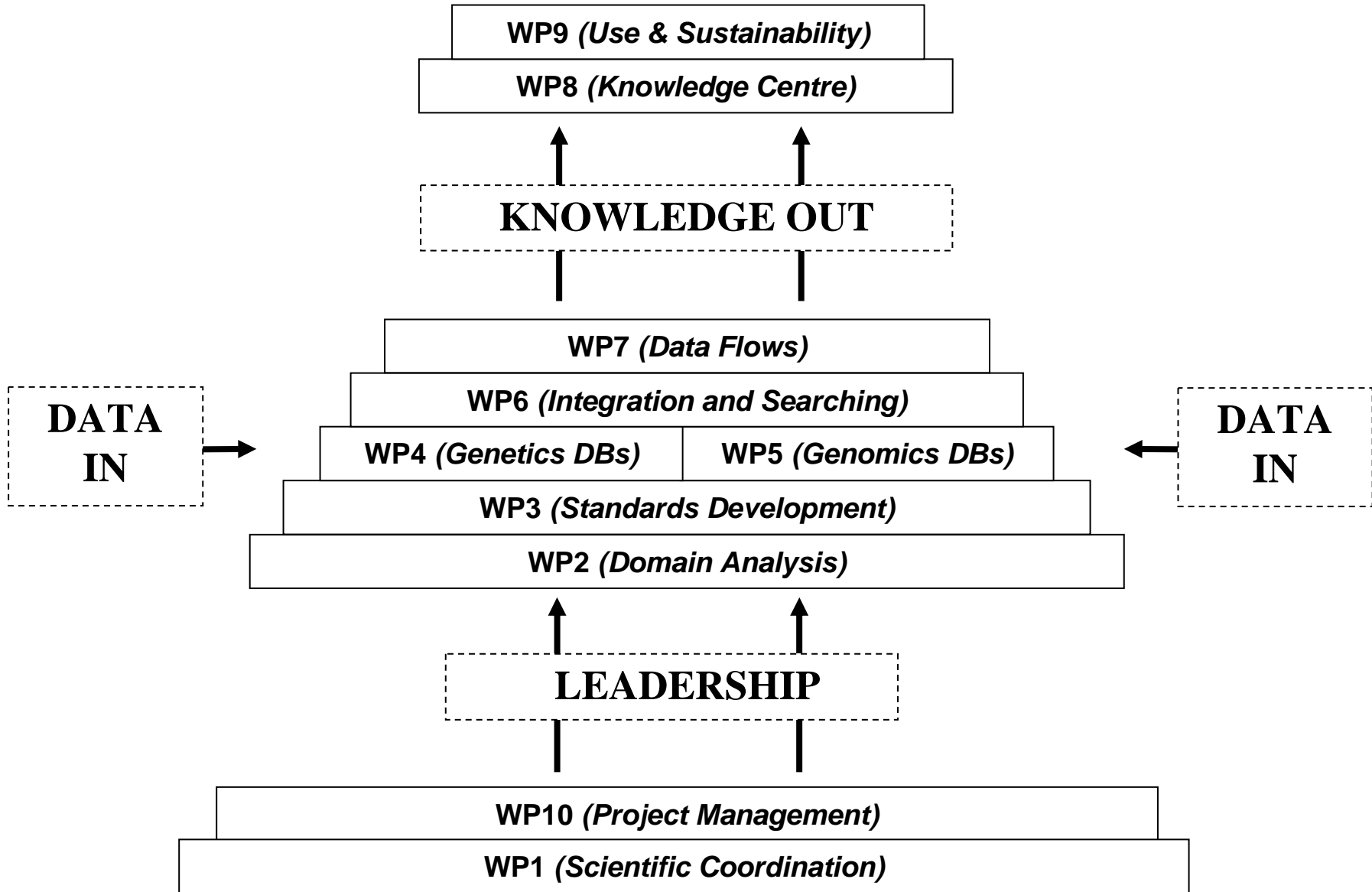
**Debasis Dash  
IGIB, CSIR, INDIA**

## GEN2PHEN

### Genotype-To-Phenotype Databases - A Holistic Solution

(EC FP7 Integrated Project, EUR 12M, 5 year duration)

University of Leicester	UK
European Molecular Biology Laboratory	Germany
Fundació IMIM	Spain
Leiden University Medical Center	Netherlands
Institut National de la Santé et de la Recherche Médicale	France
Karolinska Institute	Sweden
Foundation for Research and Technology – Hellas	Greece
Centre National de Génotypage	France
Erasmus University Medical Center	Netherlands
University of Helsinki	Finland
University of Aveiro – IEETA	Portugal
University of Western Cape	South Africa
Institute of Genomics and Integrative Biology	India
Swiss Institute of Bioinformatics	Switzerland
University of Manchester	UK
BioBase GmbH	Germany
deCODE genetics	Iceland
PhenoSystems	Belgium
Biocomputing Platforms	Finland




## GEN2PHEN: Core Objectives

- 1: Analyse current needs and practices (global perspective)
- 2: Develop key standards for the G2P field [nb. PML]
- 3: Create generic components, services and integration structures
- 4: Create search and presentation solutions, anchored on Ensembl
- 5: Promote and facilitate data population into G2P databases
- 6: Assist deployment of GEN2PHEN solutions to the community
- 7: Consider system durability and long-term financing
- 8: Execute system validation pilots, with biomedical relevance
- 9: Build a major 'G2P Knowledge Center', as a nexus for the field  
*(enabling user discussion and comments on specific database entries)*

# IGVdb: A NEW INTERACTIVE WEB SERVER FOR ANALYSIS OF INDIAN GENOME VARIATION DATA

Indian Genome Variation Database: chr3:8330000..427780000 - Windows Internet Explorer

http://localhost/cgi-bin/browse/igvdb/#search



## Indian Genome Variation Browser

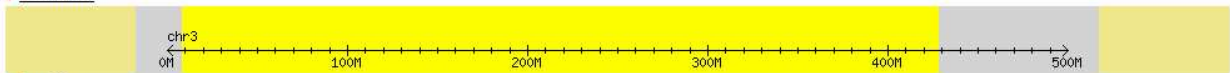
Showing 419.5 Mbp from chr3, positions 8,330,000 to 427,780,000

**Instructions**  
**Searching:** Search using a sequence name, gene name, locus, or other landmark. The wildcard character \* is allowed.  
**Navigation:** Click one of the rulers to center on a location, or click and drag to select a region. Use the Scroll/Zoom buttons to change magnification and position.

**Examples:** chr3.  
[\[Bookmark this\]](#) [\[Upload your own data\]](#) [\[Hide banner\]](#) [\[Share these tracks\]](#) [\[Link to Image\]](#) [\[High-res Image\]](#) [\[Help\]](#) [\[Reset\]](#)

**Search**  
 Landmark or Region: chr3:8330000.427780000    
 Data Source: Indian Genome Variation Database  
 Scroll/Zoom:     Show 419.5 Mbp

**Overview**



**Details**

**PHASE:**

rs1801282	rs333
rs709158	rs187084

IGVdb Portal

Welcome to IGVDB

Page 1 of 36

Chromosome	Gene	Gene Description
22	3-Sep	NIL
22	ABCA1F	Apical 1-4 galactose/branched-chain (dibutyryl)coenzyme synthase
7	ABCB1	ATP-binding cassette, sub-family B (MDR/TAP), member 1
16	ABCC1	ATP-binding cassette, sub-family C (CFTR/MRP), member 1
11	ABCC8	ATP-binding cassette, sub-family C (CFTR/MRP), member 8
17	ACACA	Acetyl-Coenzyme A carboxylase alpha
3	ACDC	NIL
17	ACR	angiotensin I converting enzyme (peptidyl-dipeptidase A) 1
22	ACOD	acetylase 2, mitochondrial
15	ACTC	actin, alpha, cardiac muscle
14	ACTN1	actinin, alpha 1
20	ADA	adenosine deaminase
5	ADAM19	ADAM metalloproteinase domain 19 (metrin beta)
2	ADAM33	ADAM metalloproteinase domain 33
4	ADPA	Adiponectin 1 (AdipoQ)

IGVdb Portal

SEARCH: rs129116

rs129116 SNP Report

dbSNP rsID	rs129116	SNP ID	SNP00589
SNP Type	Intron	Variation Type	SNP
Reference Allele	G	Chromosome	16
Alt Allele 1	A	Alt Allele 2	G

SNP rs129116 mapped to gene ABCC1

Gene ID	780	Locus Position	351
Gene Position	22607	Chromosome Position	15973541
Gene Symbol	ABCC1	Contig Position	7379119
5' Flank Seq	ACOTTGATGGGCTTTGAGTCAACCTGCTG	Start Position	15956925
3' Flank Seq	ACOTTGATGGACAGTTAAGATGGCAGG	End Position	16143774

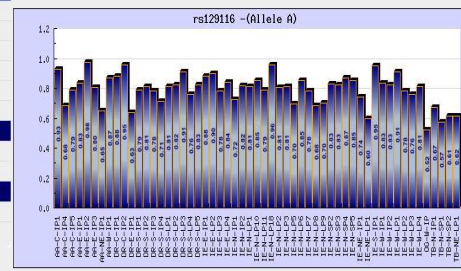
**SNP INFORMATION**

SNP Information

Population Code	No. of Samples	Frequency(G)	Frequency(A)	Frequency(M)	Frequency(G)
A-L-G-PI	41	0.07	0.05	0.15	0

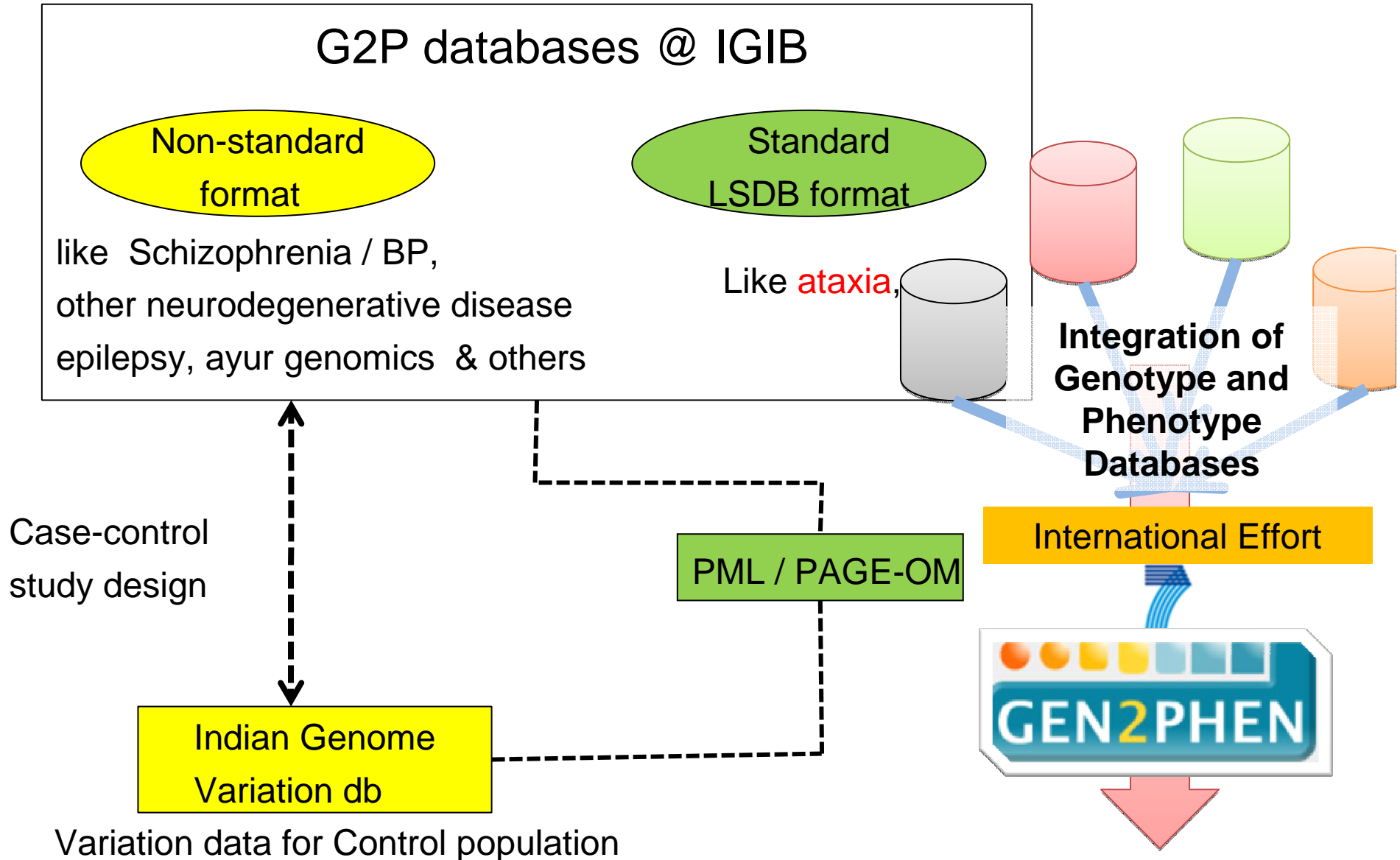
IGVdb Portal

rs129116 - (Allele A)

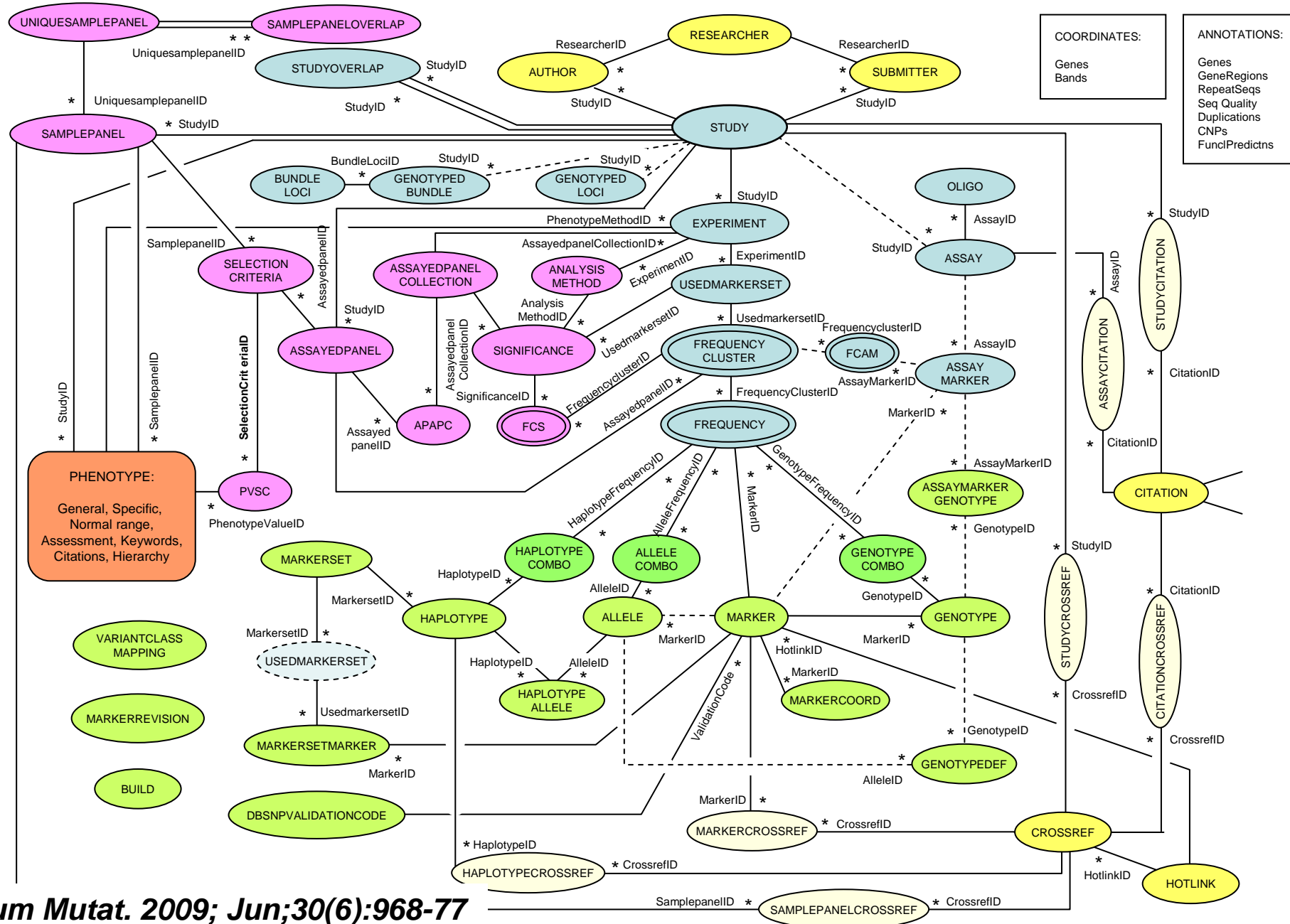


Population info

# Need for a standard format for interoperability of G2P Data



# The Phenotype and Genotype Experiment Object Model (PaGE-OM)

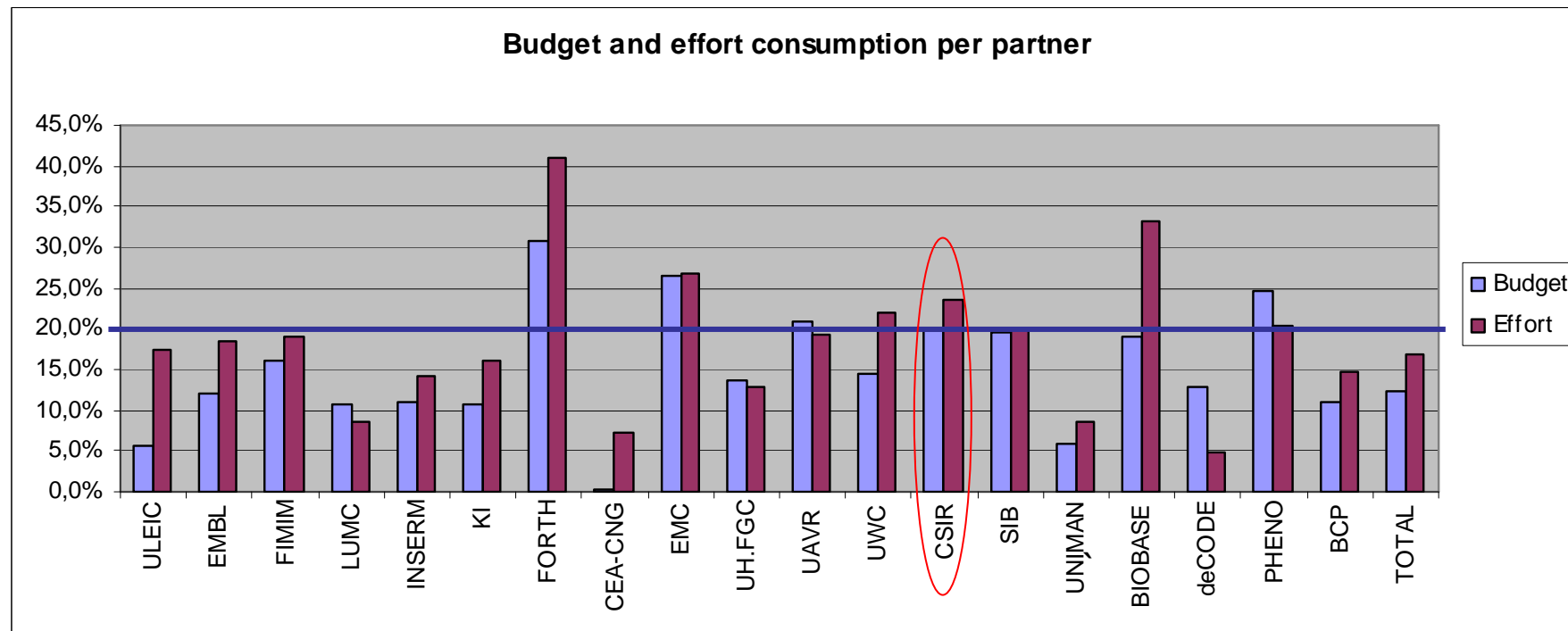


# 4<sup>th</sup> General Assembly Meeting



Reporting year 1

## Effort and budget consumption per partner, year 1



As compiled and presented in the 4th General Assembly Meeting, Hinxton, 8-9 June, 2009

**Long term collaboration needs freedom...**



**Collaborators need to share the same boat**

## Need extensive preparation to attend a Two day meeting

**Debasis Dash wrote:**

- > Dear Kerry,
- > I am yet to receive my VISA. Today in the VISA office they were **asking**
- > **for invitation letter from the host.** I furnished the meeting agenda
- > and my membership information in the Gene2Phen consortia from the
- > gen2Phen website. I do hope it should suffice as i have also provided
- > official letter of my travel. The reason i am telling you all these is
- > that i would know myself whether i am coming or not by this week end.
- > I have my ticket booked for sunday(7th midnight) and accommodation
- > (8th-10th) in Regent hotel.
- >
- > So many commitments (such as hotel, flight) without having the VISA in
- > hand is disturbing me. Anyway i have filled the dinner menu hoping for
- > the positive.
- >
- > Regards,
- > Debasis

# Getting entry to FORCE and not 'by Force'

Hi,

Still the same problem. It does not recognise my email ID. I Changed to external but no luck. even deleted browser cookies -- still the same.

Do they have my email id in their record??? Do i need to register for the first time?? Now it is mid night here. I will try again tomorrow. It seems once i get the password, i have to login within 90 mins.

Debasis

## Online submission of Form C ...not enough

Dear Debasis,

I am pleased to inform that we have received the Form C from CSIR.

Furthermore, please note that [we have not yet received the breakdown of efforts and costs, also relevant to the financial reporting](#). The templates are attached for reference.

Again, allow me to highlight that the effective deadline for the report is tomorrow, Friday the 26th of February.

Please let me know if there is anyone I should contact to follow up on this documentation.

Thank you very much once again for your kind assistance.



# Trust Researchers

A declaration to the attention of the  
European Council of Ministers and the Parliament.

[Home](#) [Declaration](#) [Background](#) [Sign up](#) [Who has signed?](#) [Who we are](#)

## Signatures

<b>Today</b>	<b>424</b>
<b>Total</b>	<b>6723</b>
Austria	933
Belgium	212
Bosnia-Herzegovina	3
Bulgaria	35
Croatia	18
Cyprus	34
Czech Republic	75
Denmark	59
Egypt	17
Estonia	25
Finland	143
France	692
Germany	365
Greece	187
Hungary	109
Ireland	20
Iceland	1
Israel	67
Italy	495
Latvia	141
Lithuania	38
Luxembourg	2

## DECLARATION

### Mutual Trust

Funding of research in Europe should be based on mutual trust and responsible partnering.

### Focused on research

Research has to be funded according to the nature of research, meaning concentrated on output.

### Consistency

The European Research Area (ERA) should benefit from a consistent vision shared by all actors for funding research throughout the different programmes that is avoiding all kinds of unnecessary technical and administrative details but instead promoting key funding principles based on an appropriate level of accountability.

### Reliability

Research and development is of utmost importance for Europe and its future oriented development. However, we need effective, reliable and stable funding principles to make it happen.

### Risk taking

Research and innovation are risk taking activities. An appropriate level of tolerable risks is vital for success and should be supported by European research programmes.

We ask the European Council of Ministers and the Parliament to urgently simplify the financial and administrative provisions related to the Framework Programme and other European funding instruments considering their important leverage effect for the competitiveness of the European Research Area.

# Simplification is essential

The new European research commissioner deserves political support from member states of the European Union to drastically reduce the dead weight of Brussels bureaucracy.

The head of a major research institute who categorically refused to allow any of his staff to apply for European Commission research funding. The science grandee who stood down from an advisory council in disgust at the paperwork. The highly regarded bright young scientist who was successfully awarded a grant but never took it up because others spotted his potential before the Euro-paperwork could be completed.

These are just a few examples of accumulated bad will and lost opportunities from decades of the Brussels experience. The principle of pan-European research collaboration, policy and infrastructures is laudable. The practice is dreadful.

In the wake of a 1999 corruption scandal involving the then research commissioner Edith Cresson, the European Court of Audi-

or modified or transferred between partners. The following year's money will not be released until this documentation is submitted and approved — a process that can take weeks.

The fear of criticism from the Court of Auditors is a miasma that envelops the commission, and in turn greatly undermines the motivation of the researchers it seeks to engage. Many scientific officers in Brussels are sympathetic and have managed to introduce some slight improvements, but their hands are tied. Moreover, others adopt a much more precautionary and burdensome interpretation.

The plain fact is that this rules- and process-based approach, appropriate enough for projects whose outcomes can be specified, is misguided in respect of research, whose outcomes are uncertain by definition. Risks that an investment might return less than hoped come

# Overhead for India is grossly underestimated

	H	I	J	K	L	M	N	O	P	Q	R	S
1	FIMIM	LUMC	INSERM		KI	FORTH	CEA	EMC	UH.FGC	UAYR	UWC	CSIR
2	Carlos Diaz	Johan den Dunnen	Christophe Beroud	Anne Cambon-Thomsen	Jan-Eric Litton	George Potamias	Ivo Gut & Simon Heath	George Patrinos	Juha Muilu	José Luis Oliveira	Heikki Lehvastaho	Samir Brahmachari
104												
	<b>Average personnel rate</b>		4000	4000	5650	3500	4000	4855	5297	5000	3000	3000
	<b>Overhead rate</b>		0.6	0.6	0.6	1.17	0.6	0.6	0.6	0.6	0.2	0.2
	<b>Other costs (travel, etc.)</b>		30000	30000	30000	30000	30000	50000	30000	30000	30000	30000
	<b>Contingency</b>											
	<b>total pm</b>		115.3	10.1	16.2	30.5	28.8	70.8	111.1	48.5	75.6	71.7
	<b>Personnel costs</b>		461201	40400	91531	106751	115201	343734	588497	242500	226802	215101
	<b>Subcontracting</b>											
	<b>Other direct costs</b>		29388	9266	26129	27812	27692	39775	27660	28585	29077	29028
	<b>Overheads max.</b>		294353	29799	70596	124898	85735	230105	369694	162651	51175	48825
	<b>Total costs</b>		784942	79465	188256	259461	228628	613614	985851	433736	307054	292954
	<b>EC contribution</b>		588706	59598	141192	194595	171471	460210	739388	325302	230290	219715
116												
117	74.4	3.7	0	0	0	0	0	3.7	0	0	0	0

Thank You