



The Human Variome Project and the Brazilian participation

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What is the Human Variome Project?

(www.humanvariomeproject.org)

- Variome – Variation in a genome
- Human Variome Project – Collection & Distribution of Human Variation & its phenotype
- Initiated 20-23 June 2006 in Melbourne, Australia co-sponsored by WHO
- A community activity to collect for databasing

What is happening in Variation?

Genomics

- SNP, HapMap, and GWAS consortia providing variants and associations for NCBI/EBI

Genetics

- Diagnostic labs/Clinicians gathering data for patient care – Locus Specific Databases (LSDB)
- Little incentive for public sharing of latter data
- Diagnostic lab/Clinicians spend hours/days/weeks searching the web
- No agreed integrated procedure

Why is complete data needed?

- Genetic clinicians and diagnostic lab heads consult mutation databases daily (and for hours per patient)
- Data needed to refine pathogenicity predictions
- Cohorts needed for mutation specific therapeutics trials
- Individuals with fully sequenced DNA need lists of mutations to inform themselves of risk
- Pre-marital counselling

HVP in Brazil

Spain, 2008: Participation on HVP Planning Meeting

Amsterdam, 2008: Brazilian Database on Orofacial Clefts

Brazil, 2010: International Conference on Nutrigenomics and 10th. International Conference on Mechanisms of Mutagenesis and Anticarcinogenesis

Brazil, 2011: Congress of the Brazilian Society of Medical Genetics (July)

Paris (Unesco), 2010- : Biannual Meeting



Brazilian Database on Orofacial Clefts

Data collection aiming:

- information for epidemiologic approaches
- monitoring and surveillance
- investigation of causes and prevention
- evaluation of outcomes

International Database on Craniofacial Anomalies (IDCFA)

Who, 2001



Brazilian Database on Orofacial Clefts

Design:

- A nation-wide, hospital-based, prospective, voluntary and collaborative **clinical database on orofacial clefts**
- Continuous and flexible process in which new aims and tools can be aggregated according to the experience gained

Operational definitions: ICD-10

Manual and a case record form specially developed

Target population

- Orofacial cleft children assisted through the specialized hospitals (recorded as separate subsets)



Brazilian Database on Orofacial Clefts

MAIN CHARACTERISTICS:

- Standard clinical evaluation
- Data are collected during part of routine genetic health care practice
- Familial data
- Clinical follow-up
- Classification according to severity



HVP and Databases

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medical genetics

PART
A

Am J Med Genet A. 2012 Nov;158A(11):2763-6

Disease-Specific Databases: Why We Need Them and Some Recommendations From the Human Variome Project Meeting, May 28, 2011

Heather J. Howard,¹ Arthur Beaudet,² Vera Gil-da-Silva Lopes,³ Mike Lyne,⁴ Graeme Suthers,⁵ Peter Van den Akker,⁶ Katarzyna Wertheim-Tysarowska,⁷ Patrick Willems,⁸ and Finlay Macrae^{1*}

A SPECIFIC DISEASE IN A DEVELOPING COUNTRY

Vera Gil-da-Silva-Lopes presented the Brazilian efforts to establish a LSDB on orofacial clefts, describing the difficulties of working within a region where genetic education and services are poorly resourced and widely dispersed. Her project, a non-government, nation-wide, hospital-based, prospective, voluntary initiative is a collaborative model for LSDBs in developing countries. She reported useful genetic epidemiological data which forms the basis for monitoring changes over time, and possible environmental exposures, as well as capturing relevant aspects of healthcare delivery.

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Disease-Specific Databases: Why We Need Them

and
Variation

Heather
Peter Va

Hindawi Publishing Corporation
Plastic Surgery International
Volume 2013, Article ID 641570, 10 pages
<http://dx.doi.org/10.1155/2013/641570>



Hindawi

Research Article

Implementing the Brazilian Database on Orofacial Clefts

**Isabella Lopes Monlleó,^{1,2} Marshall Ítalo Barros Fontes,¹
Erlane Marques Ribeiro,³ Josiane de Souza,⁴ Gabriela Ferraz Leal,⁵ Têmis Maria Félix,⁶
Agnes Cristina Fett-Conte,⁷ Bruna Henrique Bueno,⁸ Luis Alberto Magna,⁸
Peter Anthony Mossey,⁹ and Vera Gil-da Silva-Lopes⁸**

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Heather J. Hov
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Research Article

Implementing the Brazilian Database on Orofacial Clefts



*CranFlow[®]:
Craniofacial anomalies, flow and management*

National Institute of Industrial Property # BR2015510005502: a web-based application for record-taking, follow-up and management of patients.

HVP and Brazilian Databases

Brazilian Database on Craniofacial Anomalies:

Standard terminology:

Human Phenotype Ontology (HPO) (<http://www.human-phenotype-ontology.org>).

Coding:

ICD-10 [<http://www.who.int/classifications/icd/en/>] (with extensions)

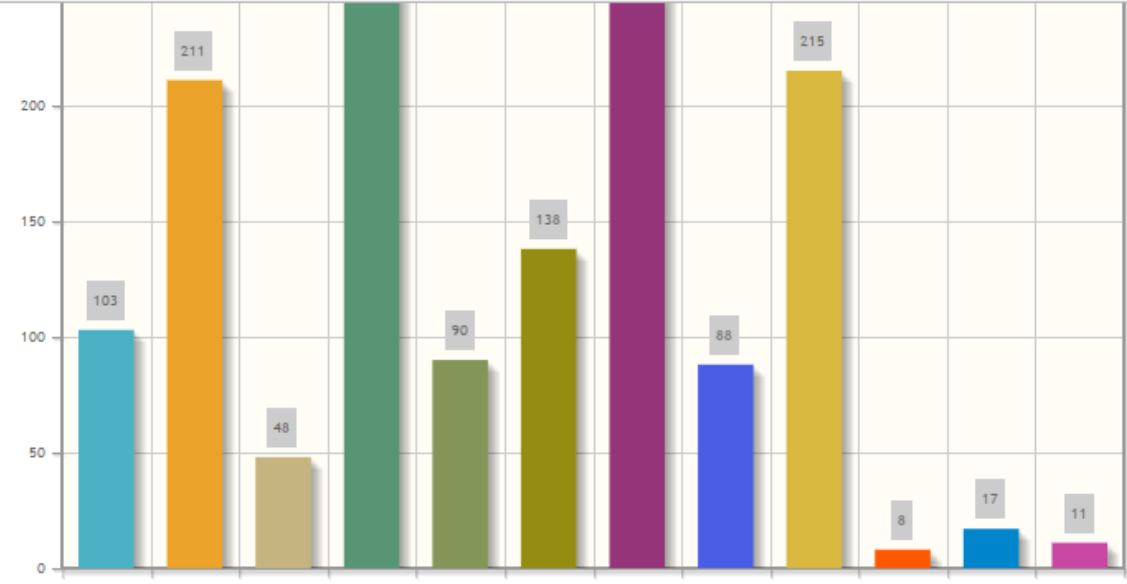
OMIM [<http://www.omim.org/>] number

Nomenclature:

International System for Human Cytogenetic Nomenclature (ISCN)

Human Genome Variation Society (HGVS) recommendations

Genomic data: Cytogenetics, Cytogenomics and Sequencing



Centrinho Prof. Luiz Gomes	138
CAIF - Centro de Atendimento Integral ao Fissurado Labio Palatal	305
CADEFI - Centro de Atenção aos Defeitos da Face	88
HUPAA - Hospital Universitário Prof. Alberto Antunes - UFAL	215
PPC - UERJ	8
APAE - São Paulo	17
CAISM - Unicamp	11
Total	1630

Resumo:

Total de Casos Encontrados: 1630

Data: 08/11/2016 16:21:38

Filtros Utilizados:

Paciente da e-BDCF: Todos

Anomalia: Todas as anomalias

Sexo Legal: Todos

HVP and Brazilian Databases

2015: BIPMed: First public [genomic database](#) in Latin America (dbSNP)

- Global Alliance for Genomics and Health: Beacon



HVP and Brazilian Databases

- Largest contry in Latin America
- 195 million inhabitants
- Very diverse genetic background



HVP Brazilian Node

Advisory Board:

Íscia Lopes-Cendes (UNICAMP)

Peter Pearson (USP)

Roberto Giugliani (UFRGS)

Vera Lúcia Gil-da-Silva-Lopes (UNICAMP)

Wilson Silva Júnior (FMRP – USP)

HVP Brazilian Node

MAIN CHALLENGES:

- To create a culture to exchange experiences and share data
- To discuss ethical parameters
- To determine short and long-term goals
- To have funding to support long-term human resources and technology

HVP Brazilian Node

- To discuss the ethical and legal parameters acceptable by Brazilian regulatory bodies:
 - patient's consent,
 - ethical approval - collection and release of genetic data
 - level of data encryption
 - data storage, data sharing and access

HVP Brazilian Node

- To establish a policy to request funding:
 - Disseminate information to research foundations, government bodies and other potential funding agencies
 - Have funds from budget of regular research programs
- Funding for:
 - Education
 - National investigation network
 - Regular meetings with different specialties and members of support groups



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