Genética Médica Populacional

CENISO –Censo Nacional de Isolados – Brasil

CELAISO – Censo Latino Americano de Isolados

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American Journal of Medical Genetics 138A:197–198 (2005)

Genetic Drift

On Being a Medical Geneticist

Eduardo E. Castilla*

ECLAMC, Genetica, Fiocruz, Rio de Janeiro RJ, Brazil

Unlike other medical specialties, medical genetics deals with families, rather than with individual persons, as patients.

venous return (TAPVR) (# 106700 OMIM), and although some familial cases with this form, as well as with the perhaps unrelated partial anomalous pulmonary venous return, or

stating: "In 2 brothers and a male paternal first cousin, Paz and Castilla [1971] observed total anomalous pulmonary venous return."

Thus,
as a **person** is the object of Medicine, and
a **family** is the object of Medical Genetics,
a **population** must be the object of Population Medical Genetics.

banding. Therefore, the normal karyotypes of the brother and sister who had fathered and mothered the three affected infants, and, as such, were presumable carriers, did not exclude the existence of a small, undetectable chromosome translocation segregating in this family. A second possible genetic mechanism was then hypothesized, namely, two loci

Thus, the updated knowledge indicates a not unexpected genetic heterogeneity for TAPVR.

These reports prompted me to recontact our patient family 30 years later, to offer them the eventual benefit of new technologies. At that time I was already living in another country, but the colleague in charge of the Department of

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- Angra dos Reis 1
- Assis Brasil 2
- Aracati 3
- Belém 4
- Campinas 5
- Cândido Godói 6
- Fortaleza 7
- Guaporé 8
- Tabuleiro do Norte 9
- Juazeiro do Norte 10
- Monte Negro 11
- Monte Santo 12
- Porto Alegre 13
- Porto Velho 14
- Pouso Alegre 15
- Rio de Janeiro 16
- Salvador 17
- São Gabriel 18
- São Paulo 19
- Triunfo 20

12

Monte Santo – BA

This group develops a comprehensive population medical genetics program in a remote area of Bahia State, which center is the town of Monte Santo (600 km far from Salvador). On this area several genetic diseases (Mucopolysaccharidosis VI, phenylketonuria and others) seem to be more frequent than in other places, possibly due to the combination of founder effect and endogamy. Prevalent mutations where identified and a customized newborn screening scheme associated to early management program is in progress, among other actions.

11

Monte Negro – RO

This group is devoted to multidisciplinary studies on infectious diseases that are endemic in the Amazon region (Malaria, Leishmaniasis, Chagas disease, Hepatitis, filariasis. Together with classical epidemiological surveys, research on genetic, mechanisms associated with these diseases are being pursued, employing modern methods of molecular genetics and powerful statistical tools of genetic epidemiology.

6

Candido Godoy – RS

Cândido Godói (CG) is a small municipality in South Brazil with approximately 6,000 inhabitants, and it is known as the "Twins' Town" due to its high rate of twin births. Recently this village has got attention from the international press after a journalist claimed that this high frequency of twinning could be connected to experiments performed by the german nazi doctor Joseph Mengele. After a call from the mayor of CG, INAGEMP started there a project to investigate the possible explanations for this twin phenomenon in this small village.

Health facts: positive

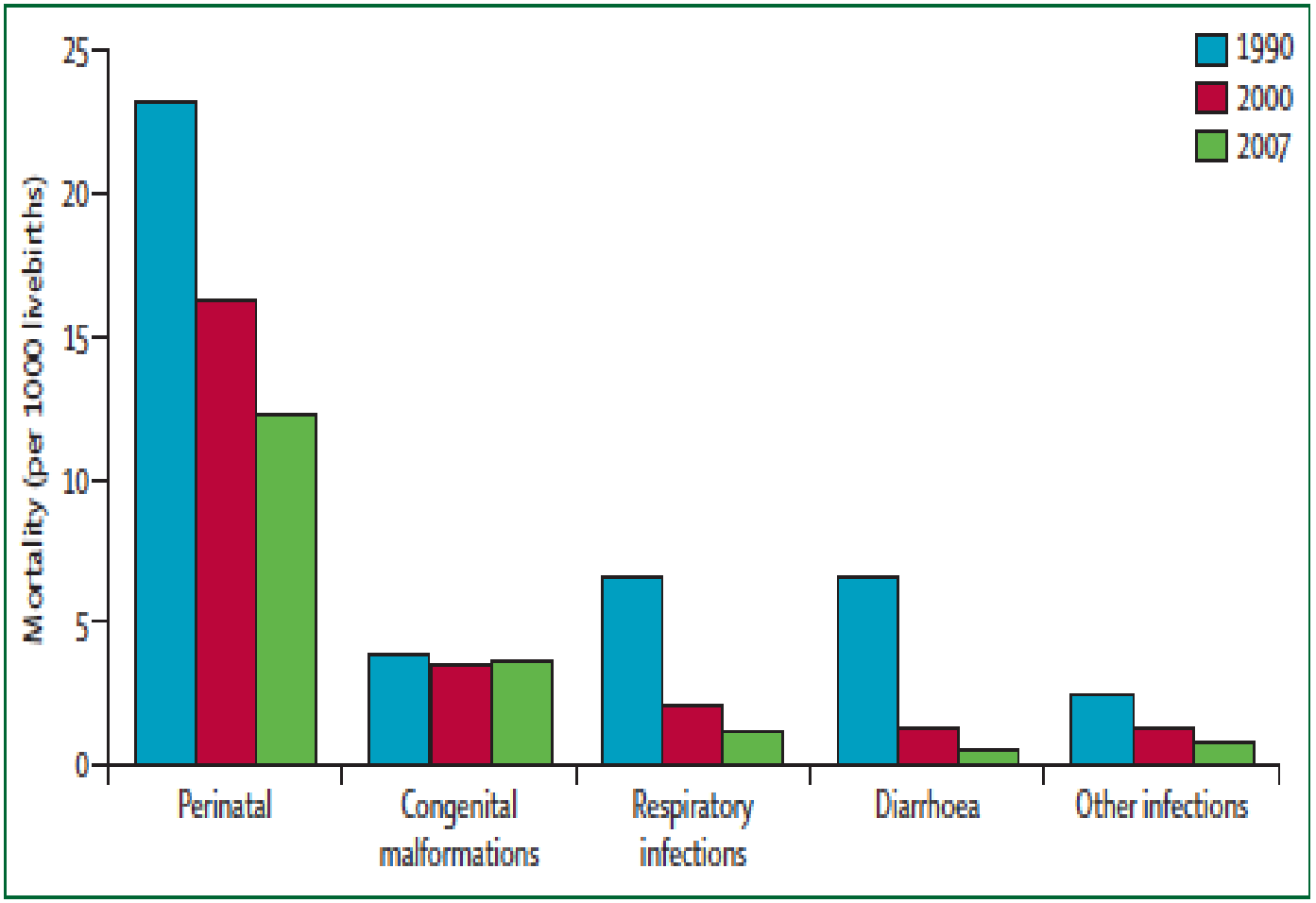

Figure 3: Infant mortality by cause and year

Victora et al., Lancet 2011; 377: 1863–76

SAÚDE BRASIL 2018

Uma análise da situação de saúde e das doenças e agravos crônicos: desafios e perspectivas

MINISTÉRIO DA SAÚDE
Secretaria de Vigilância em Saúde
Departamento de Vigilância de Doenças e
Agravos não Transmissíveis e Promoção da Saúde

Tabela 6 – Percentual de mortalidade por Causa GBD e componente – Brasil, 2000 e 2016

Ordem	Causa GBD	Percentual							
		0 a 6 dias		7 a 27 dias		28 a 364 dias		Menor de 1 ano	
		2000	2016	2000	2016	2000	2016	2000	2016
1	Complicações neonatais no parto pré-termo	48,3%	31,2%	26,2%	19,4%	4,4%	8,6%	35,2%	25,0%
2	Anomalias congênitas	10,7%	18,9%	17,9%	24,8%	22,1%	39,9%	14,3%	23,7%
3	Encefalopatia neonatal devido à asfixia e ao trauma no nascimento	19,5%	19,8%	8,4%	10,6%	2,5%	4,5%	14,0%	15,3%
4	Outros transtornos do período neonatal	10,0%	17,6%	9,8%	16,1%	2,1%	4,6%	8,2%	15,0%
5	Sepse neonatal e outras infecções neonatais	9,3%	9,4%	30,7%	23,6%	4,1%	6,2%	11,4%	11,6%
6	Corpo estranho	0,1%	0,1%	0,5%	0,8%	4,9%	7,2%	1,2%	1,5%
7	Doenças diarreicas	0,1%	0,0%	1,8%	0,4%	24,1%	5,4%	5,7%	1,0%
8	Doenças sexualmente transmissíveis, exceto HIV	0,2%	0,7%	0,2%	0,6%	0,1%	0,5%	0,2%	0,7%
9	Doença hemolítica e outra icterícia neonatal	0,7%	0,8%	1,2%	0,7%	0,1%	0,2%	0,7%	0,7%
10	Infeções de vias aéreas inferiores	0,0%	0,0%	0,1%	0,1%	2,6%	3,2%	0,6%	0,6%

continua

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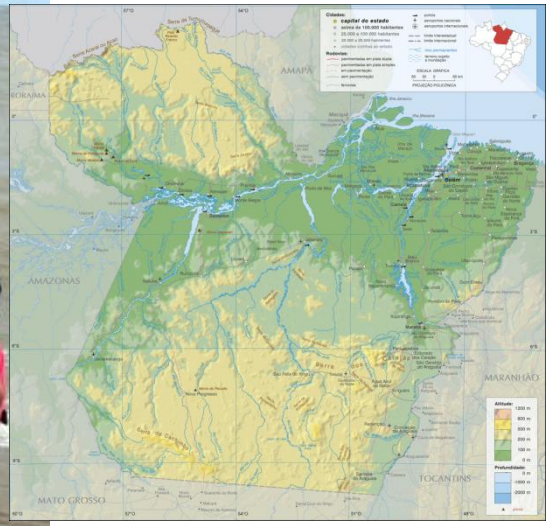
Instituto Nacional de Genética Médica Populacional

- **Population Genetics and Medical Genetics**
  - Demographic factors: migration, founder efect, isolation, inbreeding, consanguinity
  - Major predisposition gene

Geographic isolates



Cultural or Religious Isolates

- Jewish Populations
- Amish
- Romanis (ciganos)
- Menonitas (Brasil)

Signatures of founder effects, admixture, and selection in the Ashkenazi Jewish population

Steven M. Bray[a], Jennifer G. Mulle[a], Anne F. Dodd[a], Ann E. Pulver[b], Stephen Wooding[c], and Stephen T. Warr

16222–16227 | PNAS | September 14, 2010 | vol. 107 | no. 37

www.pnas.org/cgi/doi/10.1073/pnas.1004381107

The Ashkenazi Jewish (AJ) population has long been viewed as a genetic isolate, yet it is still unclear how population bottlenecks, admixture, or positive selection contribute to its genetic structure. Here we analyzed a large AJ cohort and found higher linkage disequilibrium (LD) and identity-by-descent relative to Europeans, as expected for an isolate. However, paradoxically we also found higher genetic diversity, a sign of an older or more admixed population but not of a long-term isolate. Recent reports have reaffirmed that the AJ population has a common Middle Eastern origin with other Jewish Diaspora populations, but also suggest that the AJ population, compared with other Jews, has had the most European admixture. Our analysis indeed revealed higher European admixture than predicted from previous Y-chromosome analyses. Moreover, we also show that admixture directly correlates with high LD, suggesting that admixture has increased both genetic diversity and LD in the AJ population. Additionally, we applied extended haplotype tests to determine whether positive selection can account for the level of AJ-prevalent diseases. We identified genomic regions under selection that account for lactose and alcohol tolerance, and although we found evidence for positive selection at some AJ-prevalent disease loci, the higher incidence of the majority of these diseases is likely the result of genetic drift following a bottleneck. Thus, the AJ population shows evidence of past founding events; however, admixture and selection have also strongly influenced its current genetic makeup.

GENÉTICA DE POBLACIONES

La historia de los gitanos europeos

El estudio de su genoma indica que migraron a Europa desde la India hace unos 1500 años

LOS GITANOS migraron del subcontinente indio a Europa hace unos 1500 años (*rojo*). Tras una rápida travesía por Oriente Próximo, en la que se produjo un escaso intercambio genético con las poblaciones autóctonas que encontraban en el camino, se asentaron en los Balcanes (*verde*). Empezó entonces, hace unos 900 años, la fragmentación que dio lugar a la diversidad de grupos actual.

—Isabel Mendizabal y David Comas
Instituto de Biología Evolutiva
CSIC-Universidad Pompeu Fabra

Americas – colonial period

Isolados Genéticos e Consanguinidade no Brasil.

Ann Hum Genet. 1977 Jul;41(1):99-102.

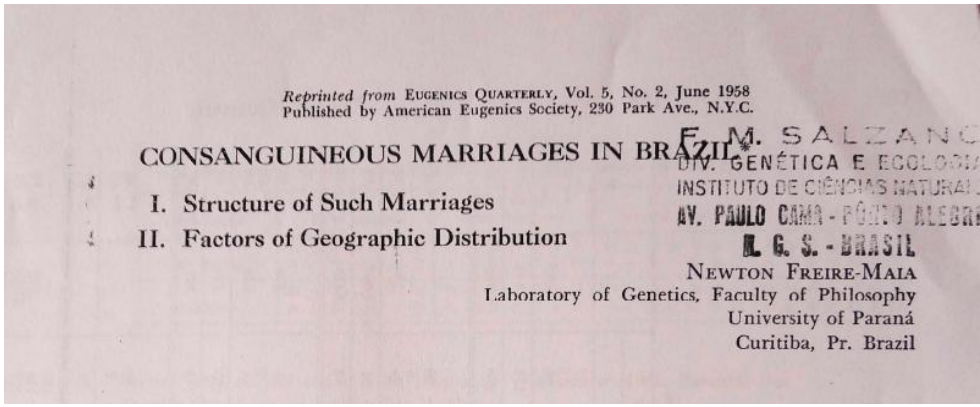
Inbreeding effect on precocious mortality in Japanese communities of Brazil.

Freire-Maia N, Takehara N.

Abstract

Data on precocious mortality (from abortions up to the age of 20) are presented for three Japanese communities in Brazil. The inbreeding load (B approximately 1-5) is estimated on the basis of these data. B/A lies between 7 and 30. Three general working hypotheses are presented.

PMID: 921222 [PubMed - indexed for MEDLINE]



Reprinted from EUGENICS QUARTERLY, Vol. 5, No. 2, June 1958
Published by American Eugenics Society, 230 Park Ave., N.Y.C.

CONSANGUINEOUS MARRIAGES IN BRAZIL

I. Structure of Such Marriages
II. Factors of Geographic Distribution

NEWTON FREIRE-MAIA
Laboratory of Genetics, Faculty of Philosophy
University of Paraná
Curitiba, Pr. Brazil

Am J Med Genet. 1990 Jan;35(1):115-7.

Genetic effects in Brazilian populations due to consanguineous marriages.

Freire-Maia N.

⊕ **Author information**

Abstract

Estimates of "relative risks" and "attributable risks" are presented for two Brazilian regions with the lowest (F = 0.00030) and the highest (F = 0.00395) inbreeding levels of the country, and for the whole country (F = 0.00088). The abolition of all consanguineous marriages (from second cousins up to and including uncle-niece/aunt-nephew marriages) in Brazil would eliminate only about 0.22, 3.05, and 0.65% of the "total damage," respectively. "Total damage" is defined as including abortions, miscarriages, stillbirths, infant-juvenile mortality (up to the age of 20 years), and anomalies in the survivors. The reduction of prenatal damage would be 0.11, 1.46, and 0.31%, and that of postnatal damage would be 0.49, 6.65, and 1.36%, respectively.

PMID: 2301460 [PubMed - indexed for MEDLINE]

Ann Hum Genet. 1964 Jun;27:329-39.

THE GENETICAL LOAD IN THE BAURU JAPANESE ISOLATE IN BRAZIL.

FREIRE-MAIA N, GUARACIABA MA, QUELCE-SALGADO A.

PMID: 14175198 [PubMed - OLDMEDLINE]

Human Heredity

Original Paper

Hum Hered 2001;51:27–34

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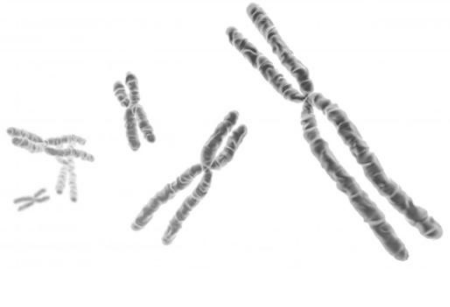
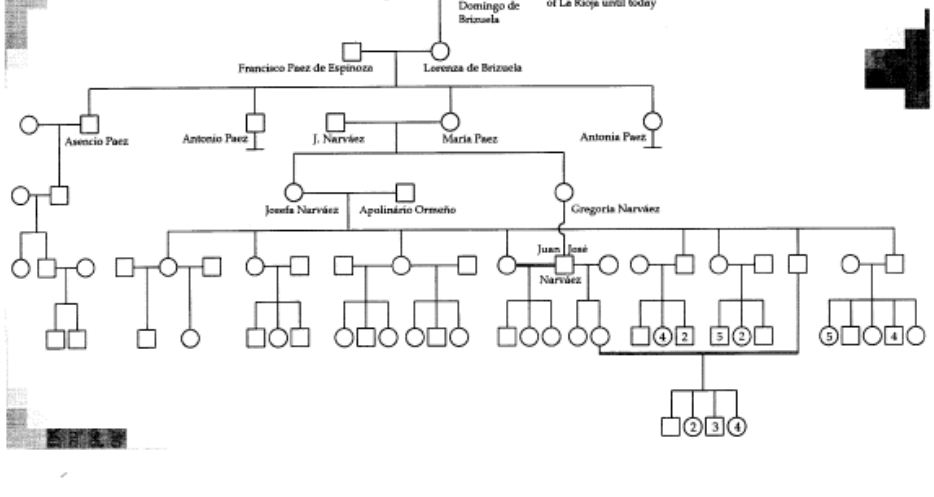
Revision received: August 31, 1999
Accepted: September 3, 1999



Consanguinity in South America: Demographic Aspects

Rosa Liascovich[a] Mónica Rittler[b] Eduardo E. Castilla[c]

[a]ECLAMC (Latin-American Collaborative Study of Congenital Malformations, WHO Collaborating Center for the Prevention of Birth Defects) at Centro Nacional de Genética Médica, ANLIS, and [b]ECLAMC at Hospital Maternoinfantil Ramon Sardá, Buenos Aires, Argentina, and [c]ECLAMC at Instituto Oswaldo Cruz, Rio de Janeiro, Brazil (EEC), and ECLAMC at Centro de Educación Médica e Investigaciones Clínicas, Buenos Aires, Argentina



Human Heredity

Original Paper

Hum Hered 2001;51:150–159

Received: Au
Revision rece
Accepted: De

Correlation between Molecular and Conventional Genealogies in Aicuña: A Rural Population from Northwestern Argentina

Graciela Bailliet[a] Eduardo E. Castilla[b] Julian P. Adams[c] Iêda M. Orioli[d] Verónica L. Martínez-Marignac[a] Silvina M. Richard[a] Néstor O. Bianchi[a]

[a]Instituto Multidisciplinario de Biología Celular, La Plata, Argentina; [b]Centro de Educación Médica e Investigaciones Clínicas, Buenos Aires, Argentina, and Instituto Oswaldo Cruz, Rio de Janeiro, Brasil; [c]Department of Biology, University of Michigan, Ann Arbor, Mich., USA; [d]Departamento de Genética, Universidade Federal do Rio de Janeiro, Rio de Janeiro, Brasil

Key Words
Mitochondrial genealogy · Y chromosome genealogy · Aicuña genealogy

Abstract
Aicuña is a village in the northwest of Argentina, located about 300 km south of La Rioja city, in the province of La Rioja. The population of Aicuña derives from a founder couple established in the uninhabited Aicuña valley in the early years of the 17th century. Due to land ownership litigation, the descendants maintained a well-documented genealogy that extends for 12 generations, comprising more than 8,000 individuals. From the historical pedigree of Aicuña, we selected 14 males with direct

which were shared by 6 and 3 donc
Three males selected as Ormeño patrili
showed a different Y haplotype (Y3), pro
by erroneous paternity registration du
The remaining case (haplotype Y4), a
belong to the Ormeño lineage, was prol
an erroneously registered paternity. Tw
showed an association of mtDNA mark
ing to the Amerindian haplotype A2. Th
matrilineage could be traced back for m
erations. The haplotype B of one rema
not correspond with the historical pedig
due to an error in the genealogy registra
showed an 85% agreement between c
molecular genealogies, with mtDNA

American Journal of Medical Genetics 9:31–41 (1981)

American Journal of Medical Genetics 2:321–330 (1978)

Acheiropodia

LMBR1

Am. J. Hum. Genet. 68:38–45, 2001

Am J Hum Genet 27:665–675, 1975

Genetics of Acheiropodia (the Handless and Footless Families of Brazil). VII. Population Dynamics

ADEMAR FREIRE-MAIA,[1] WEN-HSIUNG LI,[2] AND TAKEO MARUYAMA[3]

Under the title "Hereditary Absence of Hands and Feet," *Eugenical News* [1] published in 1929 a note and picture about a "remarkable family" from Brazil. This was the first of a series of notes published on "the handless and footless family of Brazil." For a long time, this family has been cited in a number of papers and textbooks, sometimes incorrectly.

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CENISO-BRA

Genetics and Molecular Biology, 37, 1 (suppl), 186-193 (2014)

www.sbg.org.br

Research Article

From rumors to genetic isolates

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[2]*Departamento de Genética, Instituto Nacional de Genética Médica Populacional, Universidade Federal do Rio Grande do Sul, Porto Alegre, RS, Brazil.*

Journal of Community Genetics
https://doi.org/10.1007/s12687-018-0369-1

ORIGINAL ARTICLE

Clusters of genetic diseases in Brazil

Gabriela Costa Cardoso [1] · Marcelo Zagonel de Oliveira [2] · Vanessa Rodrigues Paixão-Côrtes [3] · Eduardo Enrique Castilla [4,5] · Lavínia Schuler-Faccini [1,5]

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Genetics and Molecular Biology, 37, 1 (suppl), 171-185 (2014)

www.sbg.org.br

Genealogical data in population medical genetics: Field guidelines

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[2]*Latin American Collaborative Study of Congenital Malformations, Center for Medical Education and Clinical Research, Buenos Aires, Argentina.*
[3]*Departamento de Genética, Instituto de Biologia, Universidade Federal do Rio de Janeiro, Rio de Janeiro, RJ, Brazil.*

Abstract

This is a guide for fieldwork in Population Medical Genetics research projects. Data collection, handling, and analysis from large pedigrees require the use of specific tools and methods not widely familiar to human geneticists, unfortunately leading to ineffective graphic pedigrees. Initially, the objective of the pedigree must be decided, and the available information sources need to be identified and validated. Data collection and recording by the tabulated method is advocated, and the involved techniques are presented. Genealogical and personal information are the two main components of pedigree data. While the latter is unique to each investigation project, the former is solely represented by gametic links between persons. The triad of a given pedigree member and its two parents constitutes the building unit of a genealogy. Likewise, three ID numbers representing those three elements of the triad is the record field required for any pedigree analysis. Pedigree construction, as well as pedigree and population data analysis, varies according to the pre-established objectives, the existing information, and the available resources.

Keywords: medical genetics, population medical genetics, geographic clusters, isolates, rare diseases.

- *'Rumor'* is the systematic and scientific evaluation of any account of unusual occurrence of a given congenital anomaly or risk factor exposure.

- A lay person's anecdotal description of many cases of a given malformation in a given village is a rumor; a newspaper reporting dioxin contamination of the soil in a given area is a rumor.

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PERGUNTA:

VOCÊ SABE DE ALGUMA POPULAÇÃO COM PROBLEMAS GENÉTICOS ?

CENISO Censo Nacional de Isolados
INAGEMP Instituto Nacional de Genética Médica Populacional

inct
institutos nacionais
de ciência e tecnologia

https://www.surveymonkey.com/r/Rumor-LatinoAmericano

Usted sabe de alguna población con problemas genéticos?

El INAGEMP, Instituto Nacional de Ciencia y Tecnología, el CNPq, es un grupo de investigación dedicado a la Genética Médica de Poblaciones (www.inagemp.bio.br). Desde 2009 desarrolla un censo nacional de poblaciones brasileñas con alta frecuencia de enfermedades o características mono u oligogénicas o expuestas a factores de riesgo genético (ej.:endogamia), ambiental (ej.: talidomida), o factores desconocidos, para el ocurrencia de cualquier trastorno del desarrollo físico o mental.

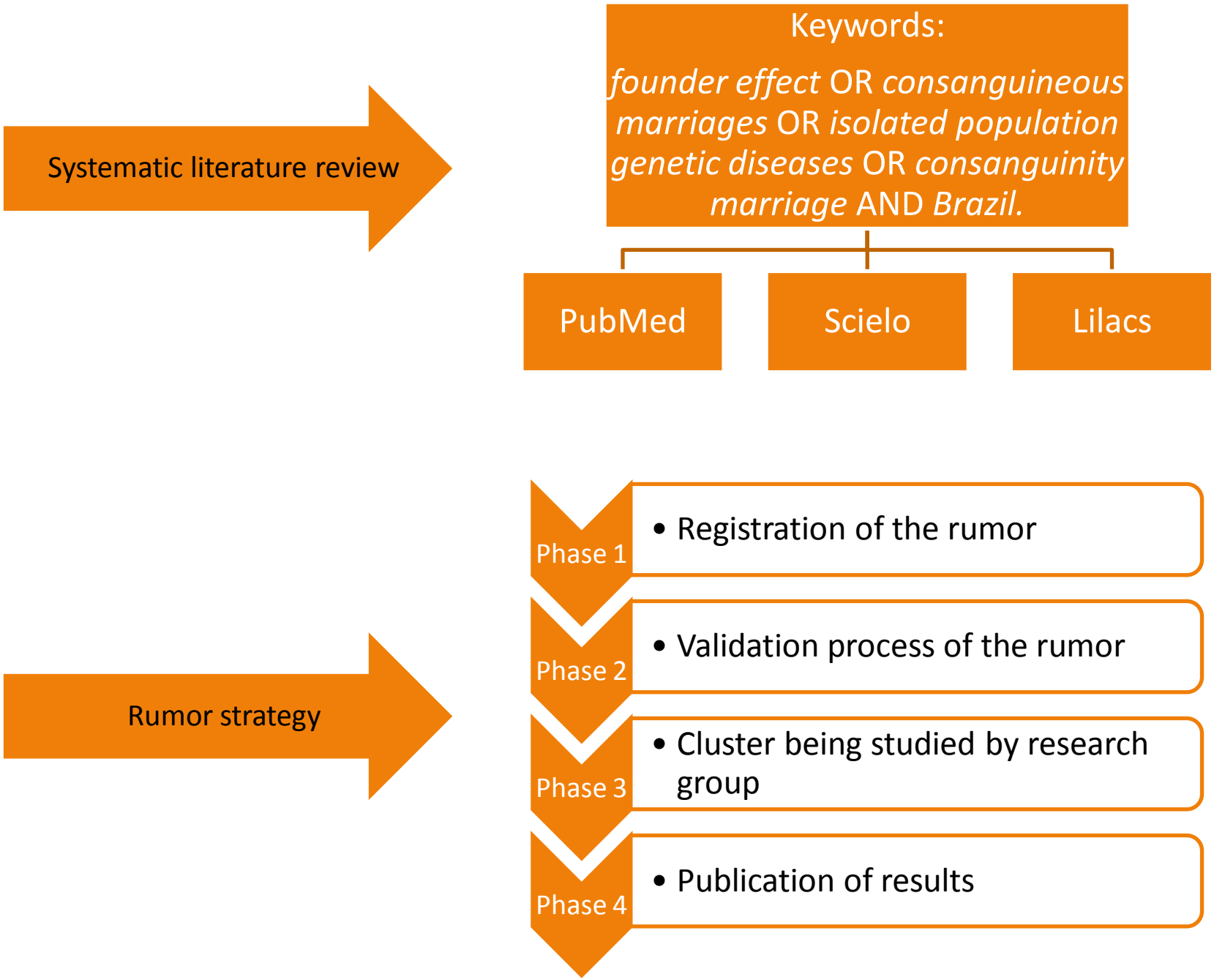
Este censo está especializado en poblaciones aisladas y grupos de enfermedades llamadas "genéticas" y constituye el primer paso para un centro especializado en investigación y normativas de atención médica de este tipos de patologías, ofreciendo servicios de soporte técnico y metodológico tanto a genetistas como a las comunidades afectadas.

Ahora se busca realizar un censo similar para las poblaciones aisladas de América Latina. En esta etapa inicial del censo estamos interesados en cualquier "rumor" o información exacta sobre poblaciones que usted considere pertinente para ser incluidas.

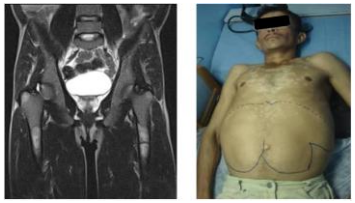
En caso de que se conozca más de una población afectada, favor diligenciar el formulario para cada una de ellas.

Agradecemos su interés y participación

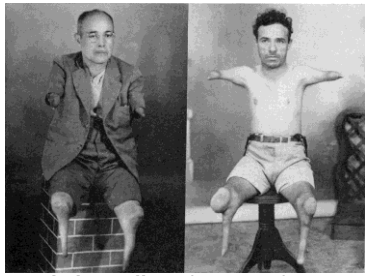
Equipo Rumor-Latinoamericano


Systematic literature review
Keywords:
founder effect OR *consanguineous marriages* OR *isolated population genetic diseases* OR *consanguinity marriage* AND *Brazil.*
PubMed
Scielo
Lilacs
Rumor strategy
Phase 1
• Registration of the rumor
Phase 2
• Validation process of the rumor
Phase 3
• Cluster being studied by research group
Phase 4
• Publication of results

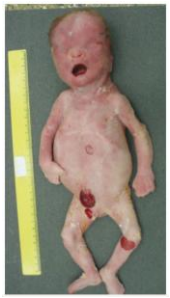
Results



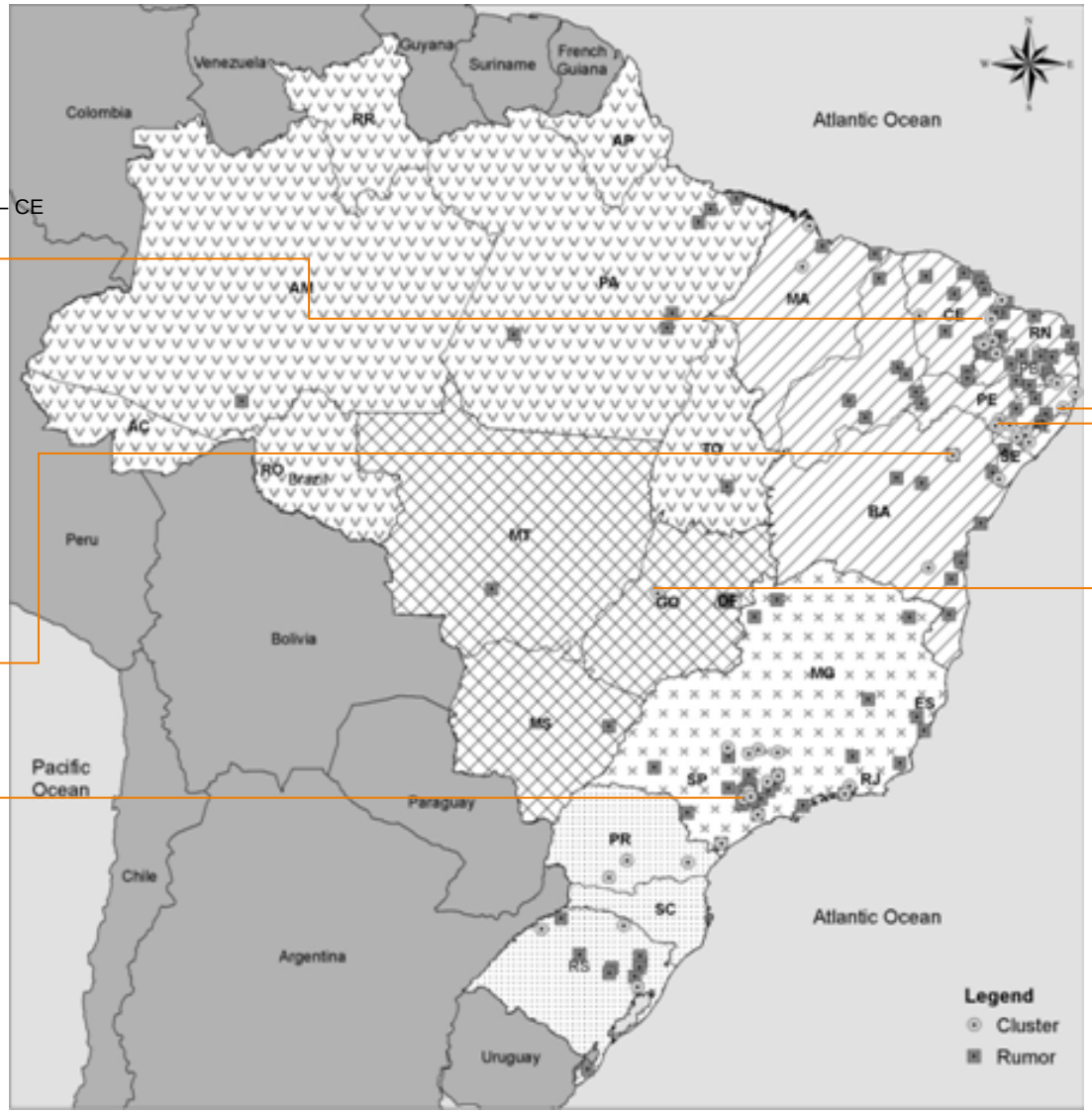
Gaucher Disease, Tabuleiro do Norte – CE
DOI: 10.1007/8904_2011_19

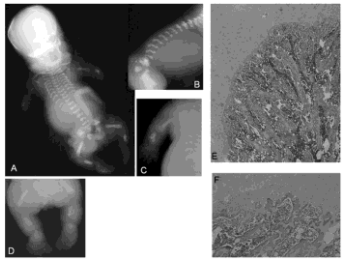


Acheiropodia, Minas Gerais
PMCID: PMC1762812

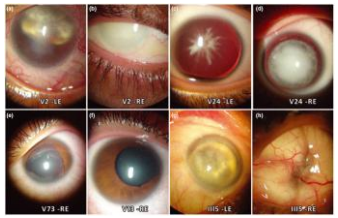


Fraser Syndrome, Vinhedo - SP
DOI: 10.1002/ajmg.a.31426





Short Rib Polidactily, Gameleira – PE
DOI: 10.1136/jmg.2009.069468



Aniridia, Água Branca – AL
DOI: 10.1111/cge.12329

Xeroderma Pigmentosum, Araras - GO
http://g1.globo.com/goias/noticia/2014/05/povoado-em-goias-tem-maior-taxa-mundial-de-doenca-rara-de-pele.html

Fig 1. Rumors and clusters identified by CENISO.

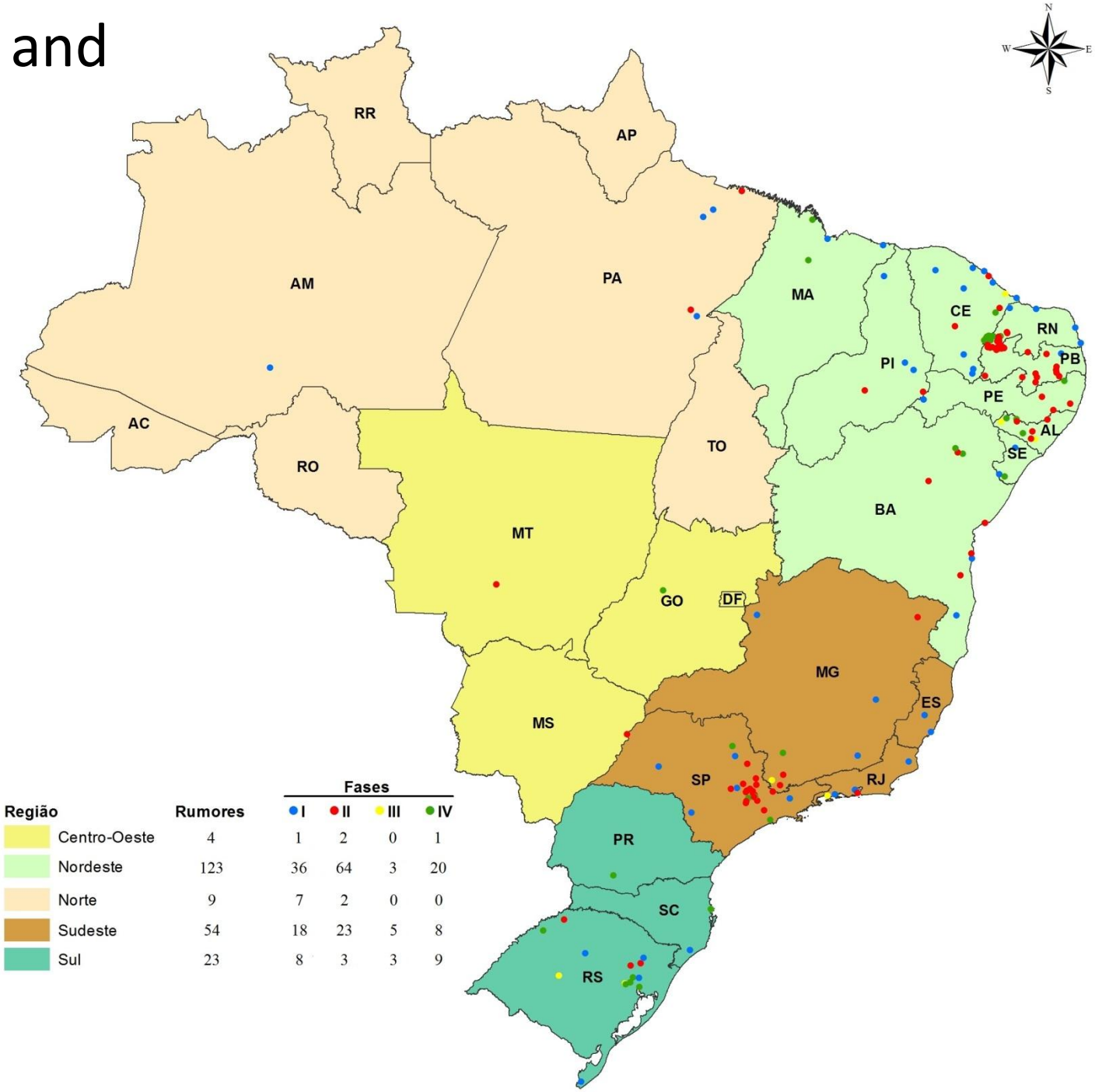
www.inagemp.bio.br

www.inagemp.bio.br

www.inagemp.bio.br/sis/produtos/ceniso_planilha.pdf#

ID	DOENÇA	MIM	LOCALIDADE	ESTADO	LAT S	LONG W	REFERÊNCIA BIBLIOGRaFICA
1	Doença de Stargardt	600110#	Sobral	CE	3° 41′	40° 21′	
2	Li-Fraumeni	151623#	Sul e Sudeste do Brasil				Achatz, MI et al., Highly prevalent TP53 mutation predisposing to many cancers in the Brazilian populations: case for newborn screening?, Lancet Oncol., 2009, 10:920-5.
3	Fendas orais	11953%	Alfenas	MG	21° 25′	45° 56′	Aquino, S et al., Estudo de pacientes com fendas labio palatinas com pais consanguíneos., Braz. j. otorhynolaryngol., 2011, 77:19-23.
4	Sarcoma precoce	190030*	Marabá	PA	5° 22′	49° 7′	
5	SPOAN	609541%	Serrinha dos Pintos	RN	06°06′	37°57′	Macedo-Souza, LI et al., Spastic paraplegia, optic atrophy, and neuropathy is linked to chromosome 11q13., Ann Neurol., 2005, 57(5): 730-7. / Macedo-Souza, LI et al., Spastic paraplegia, optic atrophy, and neuropathy: new observationslocus refinement, and exclusion of candidates genes., Ann Hum Genet., 2009, 73(Pt 3): 382-7.
6	Huntington	143100#	Feira Grande	AL	9° 54′	36° 40′	
7	Albinismo cutâneo, sem problemas visuais	203200#	Santana do Mundaú	AL	9° 10′	36° 13′	
8	Doença de Minamata		Rio Tapajós	AM	2° 26′	54° 42′	
9	Malformações		Angra dos Reis	RJ	23° 0′	44° 19′	
10	Acondrogênese de Grebe	200700#	Sul da Bahia	BA			Quelce-Salgado, A et al., A new type of dwarfism with various bone aplasias and hypoplasias of the extremities., Acta Genet Stat Med., 1964, 14:63-6.
11	Gaucher I	230800#	Tabuleiro do Norte	CE	5°15'	38°07'	
12	Tricoepitelioma familiar	601606#	Aracati	CE	4° 33′	37° 46′	
13	Malformações		Jetibá	ES	20° 1′	40° 44′	

Rumors and clusters



Região	Rumores	Fases I	II	III	IV
Centro-Oeste	4	1	2	0	1
Nordeste	123	36	64	3	20
Norte	9	7	2	0	0
Sudeste	54	18	23	5	8
Sul	23	8	3	3	9

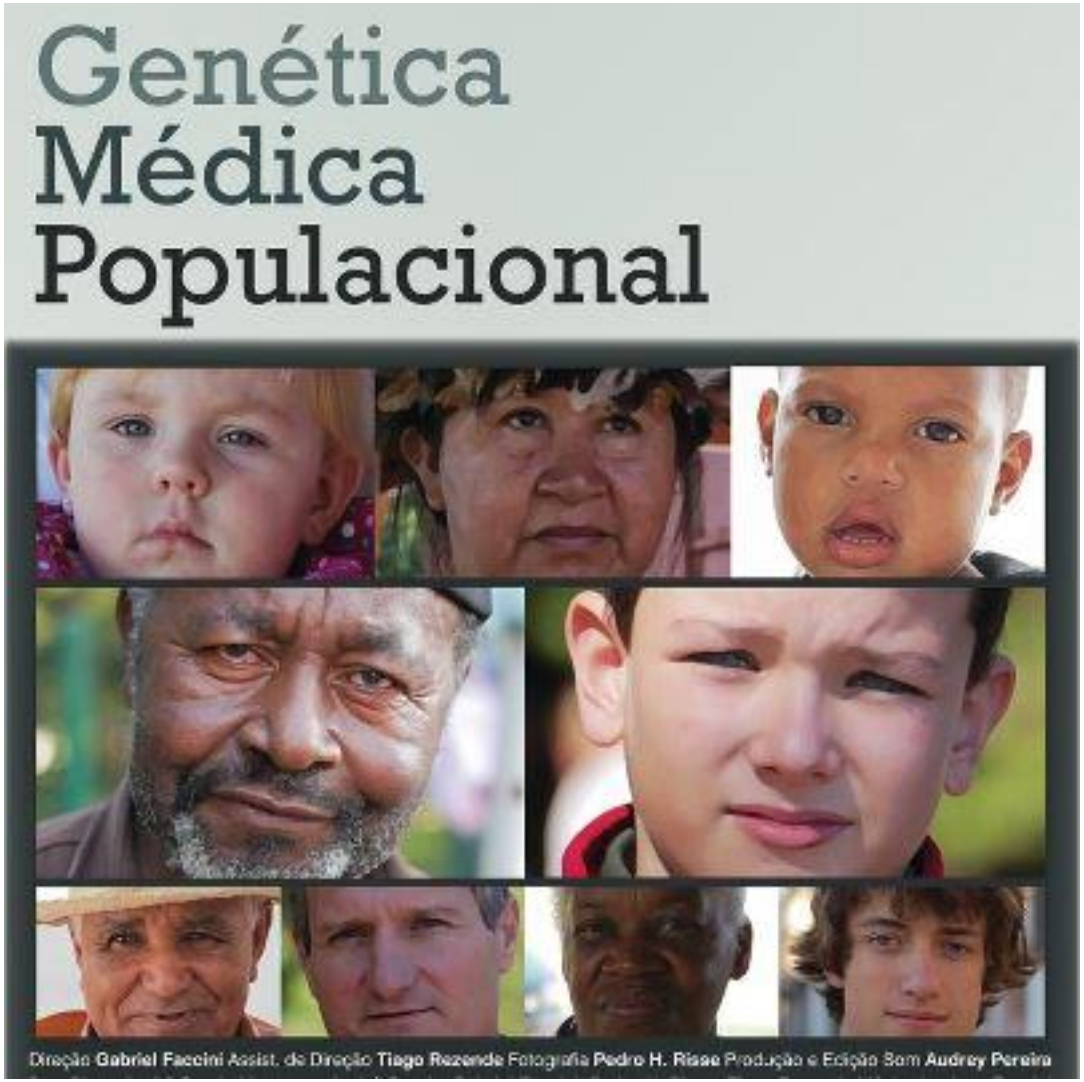
CODIGO	ENFERMEDAD	DONDE	Estado/Região/Província	PAÍS
ARG001	Albinismo	Acuña	La Rioja	Argentina
ARG001	Albinismo	Acuña	La Rioja	Argentina
ARG002	Ellis van Creveld	San Luis del Palmar.	Corrientes	Argentina
ARG002	Ellis van Creveld	San Luis Del Palmar	Corrientes	Argentina
ARG002	Ellis van Creveld	San Luis Del Palmar	Corrientes	Argentina
ARG003	Gemelaridade (gemelos)	-	Tucumán	Argentina
ARG004	Doença de Huntington	San Lorenzo e Rosario	Santa Fé	Argentina
ARG005	Doença de Sandhoff	Córdoba	Córdoba	Argentina
ARG006	Retardo metal/ autismo	Santa Fè	Santa Fé	Argentina
ARG007	Fissura palatina e anomalias urogenitais	Posadas	Misiones	Argentina
ARG008	anomalias urogenitales		Misiones	Argentina
ARG009	Alta frequência de doenças raras		Misiones	Argentina
ARG010	metabolopatias, anomalias lisossômicas		Misiones	Argentina
ARG011	neuromusculares e cerebrais com defeitos associados		Misiones	Argentina
ARG012	deficit familiar de OC		Chaco	Argentina
ARG013	Distrofia muscular de Duchenne		Chaco	Argentina
ARG014	Síndrome de Bloom	San Luis del Palmar.	Corrientes	Argentina
ARG015	Endogamia	Guatrache	La Pampa	Argentina
ARG016	Doença de Gaucher	Recreo	Santa Fe	Argentina
ARG017	Fissura oral		El Bolson	Argentina

CODIGO	ENFERMEDAD	DONDE	Estado/Região/Província	PAÍS
CHL001	Síndrome de Nijmejn	Arca Maule (Maule)	Talca	Chile
CHL002	Acromatopsia			Chile
CHL002	Acromatopsia	-	Rigião de Valparaiso	Chile
CHL003	MSUD	-	Região O'Higgins	Chile
CHL004	Colelitíase	Isla de Chiloé		Chile
CHL005	MPS	Isla de Chiloé		Chile

CODIGO	ENFERMEDAD	DONDE	Estado/Região/Província	PAÍS
COL001	Albinismo	-	Antioquia	Colombia
COL002	Alzheimer	-	Antioquia	Colombia
COL005	Doença de Huntington	-	-	Colombia
COL005	Doença de Huntington (Corea de Huntington)	Juan de Acosta	Atlántico	Colombia
COL006	MPS		Cauca	Colombia
COL006	MPS IV (Morquio)	-	Boyacá	Colombia
COL006	MPS	-	Cauca	Colombia
COL006	MPS IV (Morquio)	-	Boyacá	Colombia
COL007	MPS IV	-	Cauca	Colombia
COL008	MPS VI	Resguardo, Totoro	Cauca	Colombia
COL008	MPS VI	-	Cauca	Colombia
COL009	Fenilcetonuria (PKU)	-	Boyacá	Colombia
COL010	MPS IV A	Pereira, Manizales e Armenia	Departamentos de Risaralda, Caldas e Quindió. Região de Eje Cafeteno.	Colombia
COL011	X Frágil	Ricaurte e Bolivar	Cudinamarca(Departamento)	Colombia
COL012	Neurofibromatose tipo 1		Atlantico (departamento)	Colombia
COL013	Síndrome de Lesch Nyhan)	Meta	região Orinoquia	Colombia
COL014	Fissura oral: labio e palato	Meta	região Orinoquia	Colombia
COL015	câncer de estomago	Tuquerres	Nariño	Colombia
COL016	X Frágil	Ricaurte		Colombia
COL016	X Frágil	Ricaurte	Vale del Cauca	Colombia
COL017	Endogamia	Eje Cafetero (RIsaralda, Quindio y Caldas)	Risaralda	Colombia
COL018	Transtorno Bipolar		Antioquia	Colombia
COL019	Acidose Renal Tubular Distal		Antioquia	Colombia
COL020	Parkinson de inicio precoce		Antioquia	Colombia
COL021	Sickle Cell Disease		Antioquia	Colombia

CODIGO	ENFERMEDAD	DONDE	Estado/Região/Província	PAÍS
CRC001	MSUD	Turrialba		Costa Rica
EQU001	Sìndrome de Laron	Loja	Manabí (provincia)	Equador
EQU002	Fissura oral	região central de la serraria	Cotopaxi, Tungurahua e Chimborazo	Equador
EQU003	Ictiosis		Manabí (provincia)	Equador
EQU004	Labio leporino		sierra equatoriana	Equador
GUA001	Surdez		Huehuetenango	Guatemala
GUA002	Inflamações cutâneas		Huehuetenango	Guatemala
GUA003	Xeroderma Pigmentoso		Huehuetenango	Guatemala
MEX001	Coloboma de Iris (visão preserrada, pode ser uni ou bilateral)	Emilio Carranza	centro de Veracruz	México
MEX002	Síndrome de Van der Woude	Cintalapa	Chiapas	México
MEX003	Fibrose cistica	Base en Monterrey	Nuevo Leòn	México
NIC001	Albinismo	Matagalpa		Nicaragua
PAR001	Problemas mentais e convulsão		Chaco	Paraguay
PAR002	Ellis-van-Creveld		Gravidas paraguaias na Argentina	Paraguay
PER001	Consanguinidade	Pirua		Peru
PER002	Problemas de visão na infância	Serra do Peru		Peru
PER003	Doença de Huntington	Cañete - Lima		Peru
PER004	Lipodistrofia congénita de Berardinelli Seip	Piura		Peru
VZL001	Doença de Huntington	Rosario de Perija (Barranquita)	Lago Maracaibo	Venezuela

http://www.inagemp.bio.br/videos/quatro-herancas-genetica-medica-populacional/



Obrigada!

A autora declara não ter conflitos de interesse

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CENTRO COLABORADOR DA ORGANIZAÇÃO MUNDIAL DA SAÚDE PARA O DESENVOLVIMENTO DE SERVIÇOS DE GENÉTICA MÉDICA NA AMÉRICA LATINA