

## Rumors registered through the CENISO gateway.

| State | Locality   | Phase | Disease  | MIM    | Etiology |
|-------|--|-------|--|--------|----------|
| AL    | Santana do Mundaú  | 2     | Albinism, Oculocutaneous   | 203200 | AR       |
| AL    | Água Branca  | 4     | Aniridia (AN)  | 106210 | AD       |
| AL    | Mata Grande  | 4     | Chondrodysplasia, Blomstrand Type                                    | 215045 | AR       |
| AL    | Batalha  | D     | Chondrodysplasia, Blomstrand Type                                    | 215045 | AR       |
| AL    | Ouro Branco  | D     | Chondrodysplasia, Blomstrand Type                                    | 215045 | AR       |
| AL    | Craibas/ Marruas village   | 3     | Consanguinity and Skeletal disorder                                  |        | NI       |
| AL    | Girau do Ponciano  | 1     | Emery-Dreifuss Muscular Dystrophy 1, X-Linked (EDMD1)                | 310300 | XL       |
| AL    | Feira Grande   | 3     | Huntington Disease   | 143100 | AD       |
| AL    | Girau do Ponciano  | 2     | Mucopolidosis II, Alpha/Beta   | 252500 | AR       |
| AL    | Geographically dispersed   | 1     | Sickle Cell Anemia   | 603903 | AR       |
| AL    | Maravilha  | 3     | Undiagnosed Genodermatosis   | 173650 | AR       |
| AM    | Lábrea   | 1     | Clubfoot Congenital  | 119800 | MF       |
| AM    | Itaituba/ Rio Tapajós  | 1     | Minamata Disease   |        | Env      |
| BA    | Geographically dispersed   | 2     | Albinism, oculocutaneous   | 203200 | AR       |
| BA    | Maraú/ Ilha dos Sapinhos   | 2     | Albinism, oculocutaneous   | 203100 | AR       |
| BA    | Miguel Calmon  | 2     | Albinism, oculocutaneous   | 203100 | AR       |
| BA    | Salvador/ Ilha da Maré   | 2     | Albinism, oculocutaneous   | 203100 | AR       |
| BA    | South of Bahia state   | 4     | Chondrodysplasia, Grebe Type   | 200700 | AR       |
| BA    | Countryside of Bahia state   | D     | Chondrodysplasia, Grebe Type   | 200700 | AR       |
| BA    | Monte Santo  | 4     | Consanguinity: MPS6, PKU, Congenital Hypothyroidism and Hearing Loss |        | AR       |
| BA    | Monte Santo  | 4     | Deafness Autosomal Recessive 1A (DFNB1A)                             | 220290 | AR       |
| BA    | Monte Santo  | D     | Deafness Autosomal Recessive   | 304400 | AR       |
| BA    | Itacaré  | 1     | Dwarfism   |        | NI       |
| BA    | Vitória da Conquista/ Barra da Estiva/ Livramento de Nossa senhora | 3     | Epidermolysis Bullosa  |        | AR       |
| BA    | João Dourada/ Gameleira village                                    | 1     | Lupus  |        | AR       |
| BA    | Itapé  | 1     | Meckel Syndrome, Type 1 (MKS1)                                       | 249000 | AR       |
| BA    | Itapé  | D     | Meckel-Gruber Syndrome, Type 1 (MKS1)                                | 249000 | AR       |

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|--------|---|-------|---|--------|----------|
| BA     | Monte Santo   | 4     | Mucopolysaccharidosis, Type VI (MPS6)   | 253200 | AR       |
| BA     | Eunapolis   | 1     | Netherton Syndrome (NETH)   | 256500 | AR       |
| BA     | Monte Santo   | 2     | Phenylketonuria (PKU)   | 261600 | AR       |
| BA     | Monte Santo   | D     | Phenylketonuria (PKU)   | 261600 | AR       |
| BA     | João Dourada/ Gameleira village   | 1     | Vitiligo  |        | NI       |
| Brasil | Campinas  | 4     | GAPO Syndrome   | 230740 | AR       |
| Brazil | São Paulo   | 4     | Amyotrophic Lateral Sclerosis 8 (ALS8)  |        | AD       |
| Brazil | Geographically dispersed/ Japanese descendants  | 2     | Neuropathy, Hereditary Motor and Sensory, Proximal Type (HMSNP)                             | 604484 | AD       |
| CE     | Icapuí/ Praia de Ponta Grossa   | 1     | Bone changes  |        | NI       |
| CE     | Brejo Santo   | 1     | Cancer Familiar   |        | MF       |
| CE     | Milagres  | 1     | Change in march without ataxia, hypotrophy  |        | NI       |
| CE     | Aracati   | D     | Dermatological disease with incidence of skin cancer whose women are more affected than men |        |          |
| CE     | Fortaleza/ Vila Peri neighborhood   | 1     | Ectodermal Dysplasia 1, Hypohidrotic, X-Linked (XHED)                                       | 305100 | XL       |
| CE     | Fortaleza/ Vila Manoel Sátiro neighborhood  | 1     | Ehlers-Danlos Syndrome, Classic Type I (EDSCL1)   | 130000 | AD       |
| CE     | Tabuleiro do Norte  | 4     | Gaucher Disease, Type I   | 230800 | AR       |
| CE     | Fortaleza/ Bom Jardim neighborhood  | 1     | Hypophosphatemic Rickets, X-Linked Dominant (XLHR)  | 307800 | XL       |
| CE     | Aquiraz   | 2     | Mucopolysaccharidosis, Type II (MPS2)   | 309900 | XL       |
| CE     | Mombaça   | 2     | Mucopolysaccharidosis, Type IVA (MPS4A)   | 253000 | AR       |
| CE     | Quixeré   | 2     | Mucopolysaccharidosis, Type VI (MPS6)   | 253200 | AR       |
| CE     | Cascavel  | 1     | Neuromuscular Dystrophy   |        | NI       |
| CE     | Fortaleza/Jardim Guanabara neighborhood   | D     | Osteogenesis Imperfecta, Type I   | 166200 | AD       |
| CE     | Fortaleza/ José Walter neighborhood   | D     | Osteogenesis Imperfecta, Type I   | 166200 | AD       |
| CE     | Fortaleza/Parque Santa Maria neighborhood   | D     | Osteogenesis Imperfecta, Type I   | 166200 | AD       |
| CE     | Fortaleza/ Messejana, Jardim Guanabara, José Walter, Parque Santa Maria neighborhoods | 1     | Osteogenesis Imperfecta, Type II  | 166200 | AD       |

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|-------|---|-------|--|--------|----------|
| CE    | São Gonçalo do Amarante   | 1     | Osteogenesis Imperfecta, Type II   | 166200 | AD       |
| CE    | Geographically dispersed  | 2     | Pycnodysostosis  | 265800 | AR       |
| CE    | Várzea Alegre   | D     | Seizure with onset at 15 years old , triggered by sound stimulus, change in speech |        | NI       |
| CE    | Fortaleza   | 1     | Spinal and Bulbar Muscular Atrophy, X-Linked (SMA X1)                              | 313200 | AR       |
| CE    | Canindé   | 1     | Spinocerebellar Ataxia 2 (SCA2)  | 183090 | AD       |
| CE    | Crateús   | 3     | Spinocerebellar Ataxia 7 (SCA7)  | 164500 | AD       |
| CE    | Sobral  | 1     | Stargardt Disease  | 600110 | AD       |
| CE    | Aracati   | 3     | Trichoepithelioma, Familial  |        | AD       |
| CE    | Senador Sá  | 1     | Huntington Disease   | 143100 | AD       |
| DF    | Brasília  | 1     | Polydactyly, Preaxial II (PPD2)  | 174500 | AD       |
| ES    | Guarapari   | 1     | Malformations  |        | Env      |
| ES    | Santa Maria de Jetibá   | 1     | Malformations  |        | MF       |
| ES    | Santa Maria de Jetibá   | 1     | Skin Cancer  |        | MF       |
| GO    | Araras/ Faina village   | 4     | Xeroderma Pigmentosum, Complementation Group D (XPD)                               | 278730 | AR       |
| MA    | Cururupu/ Ilha dos Lençóis  | 4     | Albinism, oculocutaneous   | 203200 | AR       |
| MA    | Tutóia  | 1     | Ataxia-Telangiectasia (AT)   | 208900 | AR       |
| MA    | São Luís  | 1     | Myoclonic Epilepsy of Unverricht and Lundborg                                      | 254800 | AR       |
| MA    | Geographically dispersed  | 2     | Short-Rib Thoracic Dysplasia 3 with or without Polydactyly (SRTD3)                 | 613091 | AR       |
| MA    | Cajari/ Regada district   | 4     | Thalidomide Embryopathy  |        | Env      |
| MG    | Minas Gerais  | 4     | Acheiropodia   | 200500 | AR       |
| MG    | Jequitinhonha   | 2     | Cartilage Hair-Hypoplasia (CHH)  | 250250 | AR       |
| MG    | Unai  | 1     | Fragile X Syndrome (FXS)   | 300624 | XL       |
| MG    | Parque Nacional Grande Sertão Veredas, near to Arinos and Januária towns. MG and BA border. | 1     | Hypopigmentation and Mental Retardation  |        | NI       |

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|-------|-----------------------------------|-------|--|--------|----------|
| MG    | CambuÍ                            | 1     | Meckel Syndrome, Type 1 (MKS1)   | 249000 | AR       |
| MG    | Pouso Alegre                      | 1     | Meckel Syndrome, Type 1 (MKS1)   | 249000 | AR       |
| MG    | Extrema                           | 2     | Meckel Syndrome, Type 1 (MKS1)   | 249000 | AR       |
| MG    | Pouso Alegre/ São José do Pântano | 4     | Neu-Laxova Syndrome (NLS)  | 256520 | AR       |
| MG    | Ipatinga                          | 1     | Neural Tube Defects  |        | NI       |
| MG    | Alfenas                           | 4     | Oral clefts  | 119530 | MF       |
| MG    | Bueno Brandão                     | 3     | Osteogenesis Imperfecta, Type VI   | 613982 | AR       |
| MG    | Geographically dispersed          | 2     | Spondyloenchondrodysplasia (SPENCD)  |        | NI       |
| MG    | Goianá                            | 1     | Twinning   |        | MF       |
| MG    | Eralvia                           | 4     | Huntington's disease   | 143100 | AD       |
| MS    | Três Lagoas                       | 2     | Fraser Syndrome 1  | 219000 | AR       |
| MT    | Jangada                           | 2     | Hurler Syndrome  | 607014 | AR       |
| PA    | Maracana/ Fortalezinha            | 2     | Deafness Congenital  | 124480 | AD       |
| PA    | Belém                             | 1     | Diabetes Mellitus  |        | MF       |
| PA    | Itupiranga                        | 2     | Diaphanospondylodysostosis   | 608022 | AR       |
| PA    | Marabá                            | 1     | Early Sarcoma  | 190030 | AD       |
| PA    | Belém                             | D     | Several types of mental illness  |        |          |
| PA    | Belém                             | 1     | Sickle Cell Anemia   | 603903 | AR       |
| PA    | Abaetetuba                        | 1     | Waardenburg Syndrome, Type 1 (WS1)   | 193500 | AD       |
| PB    | Gado Bravo                        | 2     | Adrenal Hyperplasia, Congenital, due to 21-Hydroxylase Deficiency            | 201910 | AR       |
| PB    | São Francisco                     | 2     | Ataxia-Telangiectasia (AT)   | 208900 | AR       |
| PB    | Uiraúna                           | 2     | Ataxia-Telangiectasia (AT)   | 208900 | AR       |
| PB    | Queimadas                         | 2     | Cerebrotendinous Xanthomatosis (CTX)   | 213700 | AR       |
| PB    | Sossêgo                           | 2     | Charcot-Marie-Tooth Disease  | 606482 | AD       |
| PB    | Lagoa                             | 4     | Consanguinity with increased prevalence of disabilities (mental or physical) |        | MF       |
| PB    | Queimadas                         | 2     | Deafness Autosomal Recessive 26 (DFNB26)                                     | 605428 | AR       |
| PB    | Casserengue                       | 1     | Lumps on face and body parts   |        | NI       |
| PB    | Jericó                            | 2     | Machado Joseph Disease (MJD)   | 109150 | AD       |
| PB    | Bom Sucesso                       | D     | Machado Joseph Disease (MJD)   | 109150 | AD       |
| PB    | Lagoa                             | D     | Machado Joseph Disease (MJD)   | 109150 | AD       |
| PB    | Santa Cruz                        | D     | Machado Joseph Disease (MJD)   | 109150 | AD       |
| PB    | Uiraúna                           | D     | Machado Joseph Disease (MJD)   | 109150 | AD       |

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| PB    | Cariri region (Congo, Taperoá, Serra Branca, Coxixola towns)    | 2     | Mucopolysaccharidosis, Type IIIC (MPS3C)          | 252930 | AR       |
| PB    | Campina Grande  | 2     | Mucopolysaccharidosis, Type IVA (MPS4A)           | 253000 | AR       |
| PB    | Cariri region (Serra Branca, Coxixola, Taperoá and Congo towns) | 2     | Mucopolysaccharidosis, Type IVA (MPS4A)           | 253000 | AR       |
| PB    | Conceição   | D     | Mucopolysaccharidosis, Type IVA (MPS4A)           | 253000 | AR       |
| PB    | Congo   | D     | Mucopolysaccharidosis, Type IVA (MPS4A)           | 253000 | AR       |
| PB    | Coxixola  | D     | Mucopolysaccharidosis, Type IVA (MPS4A)           | 253000 | AR       |
| PB    | Ouro Velho  | 2     | Muscular Dystrophy, Becker Type (BMD)             | 300376 | XL       |
| PB    | Jericó  | 2     | Muscular Dystrophy, Limb-Girdle, Type 2B (LGMD2B) | 253601 | AR       |
| PB    | Santa Cruz  | 2     | Myotonic Dystrophy 1 (DM1)                        | 160900 | AD       |
| PB    | Uiraúna   | 2     | Neurofibromatosis, Type I (NF1)                   | 162200 | AD       |
| PB    | Santa Cruz  | 2     | Niemann-Pick Disease, Type C1 (NPC1)              | 257220 | AR       |
| PB    | Campina Grande  | 1     | Obesity   | 601665 | MF       |
| PB    | Vieirópolis   | 2     | Spinal Muscular Atrophy, Type I (SMA1)            | 253300 | AR       |
| PB    | Patos   | 1     | Tay-Sachs Disease (TSD)                           | 272800 | AR       |
| PB    | Gado Bravo  | 3     | Usher Syndrome                                    |        | AR       |
| PB    | Gado Bravo  | D     | Usher Syndrome                                    |        | AR       |
| PE    | Quipapá   | 1     | Albinism, oculocutaneous                          | 203200 | AR       |
| PE    | Fernando de Noronha   | 4     | Alzheimer Disease                                 |        | MF       |
| PE    | Buíque/ Cavallo Farm  | 1     | Ellis-Van Creveld Syndrome (EVC)                  | 225500 | AR       |
| PE    | Orobó   | 4     | Laron Syndrome                                    | 262500 | AR       |
| PE    | Brasil/ Recife  | 4     | Microcephaly by Zika virus                        |        | Env      |
| PE    | Belo Jardim   | 2     | Opsismodysplasia (OPSMO)                          | 258480 | AR       |
| PE    | Recife  | D     | Steinert Myotonic Dystrophy                       |        |          |

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|-------|--------------------------------|-------|---|--------|----------|
| PE    | Dormentes                      | 1     | Suicides and Consanguinity                                      |        | AR       |
| PE    | Gameleira                      | 3     | Verma-Namouff Syndrome  | 613091 | AR       |
| PI    | São Raimundo Nonato            | 1     | Consanguinity with malformations                                |        | NI       |
| PI    | Jaicós/ Várzea Queimada        | 1     | Deafness Congenital   | 124480 | NI       |
| PI    | Betânia do Piauí               | 2     | Fish-Eye Disease (FED)  | 136120 | AD       |
| PI    | Canto do Buriti                | 2     | Fish-Eye Disease (FED)  | 136120 | AD       |
| PI    | Picos                          | 1     | Hyaline Fibromatosis Syndrome (HFS)                             | 228600 | AR       |
| PI    | Esperantina                    | 1     | Porphyria, Acute Intermittent (AIP)                             | 176000 | AD       |
| PR    | Paraná                         | 4     | Adrenocortical Carcinoma, Hereditary (ADCC)                     | 202300 | AD       |
| PR    | Mangueirinha/ Reserva Kaingang | 4     | Rheumatoid Arthritis (RA)                                       | 180300 | MF       |
| RJ    | Rio de Janeiro                 | 4     | Breast Cancer   |        | MF       |
| RJ    | Campos dos Goytacazes          | 1     | Diaphragmatic Hernia, Congenital                                | 142340 | Env      |
| RJ    | Angra dos Reis                 | D     | Malformations   |        | Env      |
| RJ    | Mangaratiba/ Ilha de Marambaia | D     | Malformations   |        | MF       |
| RJ    | Duque de Caxias                | 4     | Periodontitis, Aggressive 1                                     | 170650 | AR       |
| RJ    | Rio de Janeiro                 | 2     | Spinocerebellar Ataxia 7 (SCA7)                                 | 164500 | AD       |
| RN    | São Miguel                     | 2     | Achondroplasia  | 100800 | AD       |
| RN    | Baía Formosa                   | 1     | Albinism, oculocutaneous  | 203200 | AR       |
| RN    | São Miguel                     | 2     | Alpha-Thalassaemia/Mental Retardation Syndrome, X-Linked (ATRX) | 301040 | XL       |
| RN    | São Miguel                     | 2     | Charcot-Marie-Tooth Disease                                     | 606482 | AD       |
| RN    | São Miguel                     | 1     | Clubfoot Congenital   | 119800 | MF       |
| RN    | Serrinha dos Pintos            | 2     | Cystic Fibrosis (CF)  | 219700 | AR       |
| RN    | São Miguel                     | 2     | Deafness Autosomal Dominant 18 (DFNA18)                         | 606012 | AD       |
| RN    | São Miguel                     | 2     | Developmental Dysplasia of the Hip 1 (DDH1)                     | 142700 | MF       |
| RN    | Baraúna                        | 1     | Fragile X Syndrome (FXS)  | 300624 | MF       |
| RN    | Pilões                         | 2     | Friedreich Ataxia 1 (FRDA)                                      | 229300 | AR       |
| RN    | São Miguel                     | 2     | Lesch-Nyhan Syndrome (LNS)                                      | 300322 | XL       |
| RN    | São Miguel                     | 4     | Lipodystrophy, Congenital Generalized, Type 2 (CGL2)            | 269700 | AR       |
| RN    | Pilões                         | 2     | Mucopolysaccharidosis, Type VII (MPS7)                          | 253220 | AR       |

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|--------------|----------------------------|--------------|---|------------|-----------------|
| RN           | São Miguel                 | 2            | Mucopolysaccharidosis, Type VII (MPS7)                      | 253220     | AR              |
| RN           | Pilões                     | 2            | Muscular Dystrophy, Congenital Merosin-Deficient 1A (MDC1A) | 607855     | AR              |
| RN           | São Miguel                 | 2            | Muscular Dystrophy, Duchenne Type (DMD)                     | 310200     | XL              |
| RN           | Ouro Branco                | 2            | Muscular Dystrophy, Limb-Girdle, Type 2B (LGMD2B)           | 253601     | AR              |
| RN           | Macau/ Boa Vista community | 1            | Neurodegenerative Muscular Dystrophy                        |            | NI              |
| RN           | Natal and nearby towns     | 1            | Oral Clefts   |            | MF              |
| RN           | Olho-d'água do Borges      | 2            | Osteogenesis Imperfecta, Type II                            | 166210     | AD              |
| RN           | Riacho de Santana          | 4            | Santos Syndrome   | 613005     | AR              |
| RN           | Serrinha dos Pintos        | 4            | Spastic Paraplegia, Optic Atrophy and Neuropathy (SPOAN)    | 609541     | AR              |
| RN           | Coronel João Pessoa        | D            | Spastic Paraplegia, Optic Atrophy and Neuropathy (SPOAN)    | 609541     | AR              |
| RN           | Doutor Severiano           | D            | Spastic Paraplegia, Optic Atrophy and Neuropathy (SPOAN)    | 609541     | AR              |
| RN           | Encanto                    | D            | Spastic Paraplegia, Optic Atrophy and Neuropathy (SPOAN)    | 609541     | AR              |
| RN           | Pau dos Ferros             | D            | Spastic Paraplegia, Optic Atrophy and Neuropathy (SPOAN)    | 609541     | AR              |
| RN           | São Miguel                 | D            | Spastic Paraplegia, Optic Atrophy and Neuropathy (SPOAN)    | 609541     | AR              |
| RN           | São Miguel                 | 2            | Spatic Paraplegia 35, Autosomal Recessive (SPG35)           | 612319     | AR              |
| RN           | Olho-d'água do Borges      | D            | Spatic Paraplegia 35, Autosomal Recessive (SPG35)           | 612319     | AR              |
| RN           | Pilões                     | D            | Spatic Paraplegia 35, Autosomal Recessive (SPG35)           | 612319     | AR              |
| RN           | Serrinha dos Pintos        | D            | Spatic Paraplegia 35, Autosomal Recessive (SPG35)           | 612319     | AR              |
| RN           | São Miguel                 | 2            | Spinal Muscular Atrophy, Type I (SMA1)                      | 253300     | AR              |
| RN           | Pilões                     | 2            | Spinal Muscular Atrophy, Type II (SMA2)                     | 253550     | AR              |
| RN           | São Miguel                 | 2            | Usher Syndrome  |            | AR              |

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|-------|--|-------|---|--------|----------|
| RS    | Cacique Doble (kaygang Reserv)                     | 2     | Albinism  |        | AR       |
| RS    | Cacique Doble (kaygang Reserv)                     | D     | Albinism  | 203200 | AR       |
| RS    | Interior   | 1     | Albinism, oculocutaneous                              | 203200 | AR       |
| RS    | Montenegro   | D     | Anencephaly   | 206500 | Env      |
| RS    | Triunfo  | D     | Anencephaly   | 206500 | Env      |
| RS    | Vale do Rio dos Sinos region, near to Porto Alegre | D     | Anencephaly   | 206500 | MF       |
| RS    | Santa Maria  | D     | Anencephaly   |        |          |
| RS    | Charqueadas  | D     | Brain Tumour  |        |          |
| RS    | Geographically dispersed                           | 4     | Breast and Ovarian Cancer, Familial                   | 604370 | AD       |
| RS    | Santa Vitoria do Palmar                            | 1     | Charcot-Marie-Tooth Disease                           | 118200 | AD       |
| RS    | Portão   | 1     | Dwarfism  |        | NI       |
| RS    | São Marcos   | 1     | Familial Adenomatous Polyposis 1 (FAP1)               | 175100 | AD       |
| RS    | Charqueadas  | D     | Girl with " mola"                                     |        |          |
| RS    | Caxias do Sul                                      | 2     | Glycogen Storage Disease Ia (GSD1A)                   | 232200 | AR       |
| RS    | Caxias do Sul                                      | D     | Glycogen Storage Disease Ia (GSD1A)                   | 232200 | AR       |
| RS    | Garibaldi  | D     | Glycogen Storage Disease Ia (GSD1A)                   | 232200 | AR       |
| RS    | Grande Porto Alegre                                | 4     | GM1-Gangliosidosis, Type I                            | 230500 | AR       |
| RS    | Humaitá/ Sede Nova                                 | 2     | Ichthyosis, Congenital, Autosomal Recessive 6 (ARCI6) | 612281 | AR       |
| RS    | Fortaleza dos Valos                                | 1     | Lymphedema, Hereditary IA (LMPH1A)                    | 153100 | NI       |
| RS    | Geographically dispersed                           | 3     | Machado Joseph Disease (MJD)                          | 109150 | AD       |
| RS    | General Câmara                                     | D     | Machado Joseph Disease (MJD)                          | 109150 | AD       |
| RS    | Sao Pedro do Sul                                   | D     | Machado Joseph Disease (MJD)                          | 109150 | AD       |
| RS    | Candiota   | D     | Malformations   |        | Env      |
| RS    | Triunfo  | D     | Malformations   |        | Env      |
| RS    | Nova Petrópolis/Picada Café                        | 1     | Oral Clefts   |        |          |
| RS    | Lajeado  | 1     | Orofacial Cleft 1                                     | 119530 |          |
| RS    | Charqueadas  | D     | Parents, cousins and children with strange appearance |        |          |
| RS    | Venâncio Aires                                     | 1     | Tendency to Suicide                                   |        | MF       |
| RS    | Cândido Godoi                                      | 4     | Twinning  |        | MF       |
| SC    | Criciúma   | D     | Anencephaly   | 206500 | Env      |



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|-------|---|-------|--|------------------|----------|
| SC    | Valongo   | D     | Consanguinity  |                  | AR       |
| SC    | Criciúma  | 4     | Growth hormone insensitivity with immunodeficiency   | 245590           | AR       |
| SE    | Tobias Barreto                                  | 1     | Charcot-Marie-Tooth Disease                          | 606482           | AD       |
| SE    | Itabaianinha                                    | D     | Dwarfism   |                  | NI       |
| SE    | Nossa Senhora da Glória                         | 1     | GM1-Gangliosidosis, Type I                           | 230500           | AR       |
| SE    | Itabaianinha                                    | D     | Growth Hormone Deficiency                            |                  |          |
| SE    | Itabaianinha                                    | 4     | Isolated Growth Hormone Deficiency, Type IA (IGHD1A) | 262400           | AR       |
| SE    | Itabaiana                                       | 4     | Spectrum of Pubertal Delay                           |                  | AR       |
| SP    | Luís Antônio                                    | 1     | Albinism, oculocutaneous                             | 203200<br>300419 | AR       |
| SP    | Caçapava  | D     | Anencephaly  | 206500           | Env      |
| SP    | Cubatão   | D     | Anencephaly  | 206500           | Env      |
| SP    | Cubatão/ Santos                                 | D     | Anencephaly  | 206500           | AR       |
| SP    | Paulínia  | 2     | Apert Syndrome                                       | 101200           | AD       |
| SP    | Limeira   | D     | Apert Syndrome                                       | 101200           | AD       |
| SP    | Valinhos  | D     | Apert Syndrome                                       | 101200           | AD       |
| SP    | Piracicaba/Santa Olímpia                        | 1     | Beta-Thalassemia                                     | 613985           | AR       |
| SP    | São Paulo                                       | 4     | Breast and Ovarian Cancer                            |                  | MF       |
| SP    | Campinas  | 2     | Cartilage Hair-Hypoplasia (CHH)                      | 250250           | AR       |
| SP    | Countryside of São Paulo state                  | 1     | Congenital Cataract                                  |                  | AD       |
| SP    | Tupã/Varpa district                             | 1     | Consanguinity  |                  | NI       |
| SP    | Piracicaba/ Santana/ Santa Olímpia              | D     | Consanguinity  |                  | NI       |
| SP    | Vale do Ribeira                                 | D     | Consanguinity  |                  | NI       |
| SP    | Indaiatuba                                      | 4     | Dandy-Walker Syndrome (DWS)                          | 220200           | AR       |
| SP    | Campinas  | 2     | Diaphanospondylodysostosis                           | 608022           | AR       |
| SP    | Coronel Macedo                                  | 1     | Fragile X Syndrome (FXS)                             | 300624           | XL       |
| SP    | Itu   | 2     | Fraser Syndrome 1                                    | 219000           | AR       |
| SP    | Vinhedo   | 4     | Fraser Syndrome 1                                    | 219000           | AR       |
| SP    | Jundiaí   | 2     | GM1-Gangliosidosis, Type I                           | 230500           | AR       |
| SP    | Ribeirão Preto                                  | 4     | Gomez-Lopez-Hernandez Syndrome (GLHS)                | 601853           | AR       |
| SP    | São Paulo                                       | 1     | Hereditary Hemochromatosis                           |                  | AD       |
| SP    | Atibaia/São João da Boa Vista. SP and MG border | 1     | Huntington Disease                                   | 143100           | AD       |

| State                | Locality                      | Phase | Disease  | MIM    | Etiology |
|----------------------|-------------------------------|-------|--|--------|----------|
| SP                   | São Paulo                     | 1     | Hypercholesterolemia                                     | 144010 | AD       |
| SP                   | Vale do Ribeira/ Jacupiranga  | 3     | Hypertension and Consanguinity                           | 145500 | MF       |
| SP                   | São Paulo                     | 4     | Isolated Growth Hormone Deficiency                       |        | AR       |
| SP                   | Vinhedo                       | 2     | Maple Syrup Urine Disease (MSUD)                         | 248600 | AR       |
| SP                   | Sao Paulo                     | D     | Marfan Syndrome  | 154700 | AD       |
| SP                   | Mogi Guaçu                    | 2     | Meckel Syndrome, Type 1 (MKS1)                           | 249000 | AR       |
| SP                   | Piracicaba                    | D     | Meckel Syndrome, Type 1 (MKS1)                           | 249000 | AR       |
| SP                   | Salto                         | D     | Meckel Syndrome, Type 1 (MKS1)                           | 249000 | AR       |
| SP                   | Sumaré                        | D     | Meckel Syndrome, Type 1 (MKS1)                           | 249000 | AR       |
| SP                   | São Paulo                     | D     | Myasthenia   |        |          |
| SP                   | Vale do Ribeira               | 3     | Obesity and Consanguinity                                | 601665 | MF       |
| SP                   | Sumaré                        | 2     | Postaxial Acrofacial Dysostosis (POADS); Miller Syndrome | 263750 | AR       |
| SP                   | Sta. Cruz das Palmeiras       | D     | Postaxial Acrofacial Dysostosis (POADS); Miller Syndrome | 263750 | AR       |
| SP                   | São Paulo                     | 4     | Progressive Muscular Dystrophy                           |        | XL       |
| SP                   | São Paulo                     | 4     | R337H Mutation in TP53 gene in Adrenocortical Tumors     |        | MF       |
| SP                   | Vale do Ribeira               | 2     | Richieri-Costa-Pereira Syndrome                          | 268305 | AR       |
| SP                   | São Paulo                     | 4     | Richieri-Costa-Pereira Syndrome                          | 268305 | AR       |
| SP                   | Santo Antônio de Posse        | 2     | Seckel Syndrome 1 (SCKL1)                                | 210600 | AR       |
| SP                   | Ubatuba/ Ilha dos Porcos      | 1     | Short stature  |        | NI       |
| SP                   | São Paulo                     | 2     | Spinocerebellar Ataxia 1 (SCA1)                          | 164400 | AD       |
| SP                   | Ribeirão Preto                | 4     | Spinocerebellar Ataxia 1 (SCA1)                          | 164400 | AD       |
| SP /<br>MG<br>border | Mococa e Guaxupé              | 4     | Multiple Endocrine Neoplasia type 1 (MEN1)               | 131100 | AD       |
| TO                   | Chapada da Natividade         | 1     | Sickle Cell Anemia                                       | 603903 | AR       |
|                      | South and southeast of Brasil | 4     | Li-Fraumeni Syndrome type 1 (LFS1)                       | 151623 | AD       |
|                      | South of Brazil (RS and PR)   | 4     | Skin cancer in Mennonites communities                    |        | MF       |
|                      | Geographically dispersed      | 4     | p.R337H Mutation in TP53 Locus                           |        | MF       |

States: AL: Alagoas; AM: Amazonia; BA: Bahia; CE: Ceará; DF: Distrito Federal (Federal District); ES: Espírito Santo; GO: Goiás; MA: Maranhão; MG: Minas Gerais; MS: Mato Grosso do Sul; MT: Mato Grosso; PA: Pará; PB: Paraíba; PE: Pernambuco; PI: Piauí; PR: Paraná; RJ: Rio de Janeiro; RN: Rio Grande do Norte; RS: Rio Grande do Sul; SC: Santa Catarina; SE: Sergipe; SP: São Paulo; TO: Tocantins.

Phases: Phase 1. Registration of the rumors; Phase 2: Validation process of the rumors (investigation of the rumors to validate if they are true or not. If after that rumors are not true they will be discarded, if they are true will enter in phase 3 and will be classified as a cluster); Phase 3: Community cluster being studied by research group; Phase 4: Publication of results; D: Discarded.

Etiology: AD - Autosomal Dominant il; AR - Autosomal Recessive; XL - X-Linked; MF – Multifactorial; Env - Environmental; NI - Not Identified.