HUMAN GENOME RESEARCH CENTER (HGRC)

Departamento de Genética e Biologia Evolutiva
Instituto de Biociências
Universidade de São Paulo

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PART 1 - RESEARCH

GENOME RESEARCH

I- Neuromuscular and Neurodegenerative Disorders

a) Search of new genes: LGMD1G and SPOAN

We have tested several candidate genes but until now we were not able to identify the genes for LGMD1G and SPOAN. We will now try to identify these genes using next generation sequencing.

b) African Ancestry protects against dementia-related neuropathology

This research showed unexpected results, that is, that neuropathological changes were more prevalent in brains from people of European than African ancestry. These findings are in opposition to demographic data from USA that observed that Alzheimer disease is more prevalent among African American than Caucasian. The results were published in the prestigious journal Molecular Psychiatry (Schlesinger et al., 2011).

c) Insights from exceptional cases

A family with Sarcoglycanopathy, and atypical pattern of DNA in the GSG gene was studied through the new Motor chip microarray analysis, for mutations in 245 neuromuscular disorders genes, and was characterized at the molecular level with two new large deletions in the gama-sarcoglycan gene (Piluso et al., 2011).

d) Phenotypic variability

The effect of heterozygous mutations in gene related to the glycosilation of a-DG in the muscle was evaluated both in patients with different NMD as well in mouse model for the dystrophin mutation, heterozygouse for one mutation in the large gene. The preliminary results showed that defect in glycosylation can worst the phenotype in the dystrophic muscle (Martins et al., 2011).

In patients with myotubular myopathy, the association between the presence of necklass fibers and the severity of the phenotype was studied (Gurgel-Giannetti et al., 2012). Our studies in the congenital myopathies resulted in the participation in the Consortium of Standard of Care for Congenital myopathies, and the publication of an international Consensus Statement (Wang et al., 2011, in press).

II- Developmental Disorders: Craniofacial defects, autism, neurodevelopmental disorders, mental retardation associated or not with malformation, obesity and deafness

a) Identification of candidate genes for autism

In two patients with autism spectrum disorders (ASD) and cytogenetic balanced chromosomal rearrangement, we have concluded the characterization of the chromosome breakpoints. In one of them, even though the breakpoints did not disrupt any known gene,

we verified through genomic analysis (arrayCGH) that this patient besides the balanced translocation has maternal unidissomy of chromosome 5 and duplication of part of chromosome 5 of paternal origin. Taken together all these findings with those in the literature, we have suggested that the most likely cause of the ASD phenotype might be due to recessive mutations arisen by the maternal unidissomy of chromosome (Griesi-Oliveira et al., manuscript submitted for publication). In the other patient, we found disruption of the *TRPC6* gene , which is an important protein to control Ca+2 influx in neuron and known to play an important role in neuronal synapses. We have next investigated the functional effect of deficiency of *TRPC6* in neuronal function through the use of iPSC (induced pluripotent stem cells) and in a knockout mouse for *TRPC6*. Our findings suggest that *TRPC6* is a candidate gene for ASD as its deficiency cause altered function in the neurons derived from the iPSC of the patient. Even though we did not find any obvious clinical autism behavior in the mouse knockout model, our data suggest that deficiency of *TRPC6* seems to be a predisposing factor for ASD (Griesi-Oliveira et al, manuscript in preparation). This project has been done in collaboration with Dr. State (University of Yale, USA) and Dr. Muotri (University of California San Diego, USA).

b) Identification of at-risk alleles and signaling pathways in non syndromic cleft lip and palate (NSCLP)

Through the analysis of more than 1000 families with NSCLP, we showed that heritability for NSCLP, although variable, is usually high (>65%), thus indicating the value to conduct genomic studies in our population. Another relevant finding is that heritability seems to be a confounding effect and therefore, pooling samples from different populations are not necessarily the best approach to conduct association studies. These results will have important implications in the experimental design of our future projects (by Brito et al., 2011).

We demonstrated for the first time the usefulness to study mesenchymal stem cells from patients with NSCLP (Bueno DF, Sunaga DY et al., 2011), which will open news perspectives to study the etiology and pathways involved with the origin of this malformation.

c) Craniofacial syndromes

In the year of 2011 we obtained relevant information on the patophysiology of Apert Syndrome (AS) (Yeh et al., 2011), a disorder mainly characterized by excess and premature bone ossification at the cranial sutures.

Through the study of fibroblasts and stem cells from AS patients, in contrast to data on the literature, we are observing that periosteum at the calvaria might play an important role in the resynostosis of the sutures of these patients and a possible pathway involved in this process was identified. These results encourage us to further pursue for the search of the pathways involved in this process as new molecules that facilitate bone regeneration can be identified as well as open perspectives to identify drugs that can decrease the rate of ossification process in these patients.

d) Malformation syndromes

We showed that analysis of chromosomal regions through MLPA have nearly the same chance to detect the most prevalent chromosomal alterations than karyotyping (Jehee et al., 2011). These results opens the perspective to use this approach in large scale as this test is less expensive than karyotyping and allows automation. In this context,

in collaboration with a group of Vitoria, Espirito Santo, it is being conducted a screening through MLPA of all malformed newborns. This will open a perspective to improve health care system in genetics in Brazil.

e) Deafness

Our findings about the role of mitochondrial polymorphisms and family history of deafness in genetic susceptibility to noise induced hearing loss were published (Abreu-Silva et al., 2011). Besides, the mapping of an autosomal dominant gene of deafness to DFNA18 region (Mingroni-Netto et al., 2011a, abstract), was an important finding because this is the second report of linkage to this chromosome region, since the original description of the locus, and the causative gene was never found. The search of the causative gene in the Brazilian family is under way. An interesting report of copy-number variation, including the gene *DOCK4* expressed in cochlea and segregating with deafness in a pedigree, indicated this gene as a novel candidate gene for hereditary deafness. Results were presented in an international conference (Mingroni-Netto et al., 2011c, abstract).

f) Genetic factors associated with hypertension and obesity in Afro-Brazilian partially isolated populations

The results of the genotypes in seven different genes were analysed regarding their role on the development of overweight and obesity in Afro-derived *quilombo* populations. Although significant associations were not detected when each gene was considered individually, we found evidence of interaction between genes, especially between the genes *ADRB2* and *LEPR,* and results are described in Angeli *et al*. (2011). The hypothesis of interaction between the two genes was validated in a population of Japanese ancestry and data were published in Pereira *et al*. (2011). Also, in Pereira and Mingroni-Netto (2011), a method for meta-analysis of association studies involving bi- and tri-allelic polymorphisms was developed and described.

g) Syndromic obesity

After microarray analysis in a series of patients with dysmorphic syndromes that include obesity and developmental delay, we identified several cryptic chromosomal imbalances representing new aetiologies of syndromic obesity and, hence, novel obesity loci for targeted follow-up. We developed a set of custom-designed MLPA probes to rapid screen for regions of copy number gains and losses that might be overrepresented in a cohort selected for syndromic obesity. Most obese patients had normal methylation test for PWS or had other common causes of syndromic obesity, such as 1p36 and 17p11.2 deletions, ruled out. To date, we have found six of over 150 patients carrying chromosomal deletions at 15q11.2 (three), 2q37 (two), and 9q34 (one).

h) Prader-Willi and Angelman syndromes

In this ongoing project, the sample of PWS and AS patients was increased, and we also described a novel chromosome microdeletion at 15q26.1 detected by oligo-array-CGH in a 6-year-old girl presenting features often observed in Angelman syndrome: global development delay, epilepsy, autistic behavior and facial dysmorphisms. The deletion encompasses only 2 genes: CHD2, which is part of a gene family already involved in CHARGE syndrome, and RGMA which exerts a negative control on axon growth. These

results provide a further chromosome region requiring evaluation in patients presenting Angelman features (Capelli, et al., 2011).

i) Silver Russell syndrome

Genetic and epigenetic disturbances are detected in about 50% of the patients with the Silver Russell syndrome of restriction growth. In our cohort of 64 unrelated patients, 4,3% had maternal UPD 7, and 43%, hypomethylation of the telomeric imprinting center (ICR1) on 11p15, the most common causative mechanisms. An ICR2 microduplication was detected in a familial case. In his three-generation family there were four instances of paternal transmissions of the microduplication from a single male uniformly resulting in normal offspring, and five maternal transmissions, via two clinically normal sisters, with all the children exhibiting SRS. A single SRS child had been previously described carrying an ICR2 microduplication. Our finding provided confirmatory evidence that a microduplication restricted to the ICR2 domain results in SRS when maternally transmitted, and showed that no apparent phenotypic change is present when the ICR2 duplication is paternally inherited. Among 19 patients negative for (epi)genetic mutations at 11p15 and mUPD7, three had potentially pathogenic microdeletions of other chromosomes that are under investigation (Bonaldi A, MsSc. Dissertation; Bonaldi et al., 2011)

III- Chromosomal Studies

This study comprised six apparently balanced translocations. Two of them, in patients with acampomelic campomelic dysplasia, involved chromosome 17; the breakpoints, mapped upstream *SOX9*, provided additional information regarding the regulatory region of the gene, and allowed the redefining of the distal breakpoint region. A conserved element was identified as a candidate *SOX9* enhancer for testis development. By mapping the breakpoints of a t(2;16) in a patient with hand and feet defects a putative regulatory element of the *IHH* gene (involved in limb development) was identified. In three other translocations, cryptic duplications/deletions detected in cis to the breakpoints could account for the clinical phenotype of carriers. (Fonseca AS, MSc Dissertation, 2011; Fonseca et al. Reunião Brasileira de Citogenética, 2011).

IV- Interfering in the Human Genome

The main purpose of this part of the project is to analyze cell's responses to DNA damaging agents, and try to understand why certain human syndromes defective in DNA repair (basically nucleotide excision repair, NER, such as xeroderma pigmentosum-XP- and Cockayne syndrome- CS) have so broad and distinct clinical symptoms. Also we are using cells from these patients (and thus with natural genomic instability in the presence of DNA damage) to identify DNA repair pathways responsible for the repair of chemotherapeutic drugs. The idea is that these pathways will reveal potential gene targets for improving the activity of such drugs, helping battle against tumor cells. During this last year, relevant data was obtained with the analysis of oxidatively generated DNA damage (generated by methylene blue and light) effects in XP cells. Basically the main conclusions were that XPC and XPA proteins are involved in the cell protection from these types of lesion, and faulty repair of oxidative damage may in fact be necessary to clinical phenotypes related to progeroide symptoms, however they are not sufficient. This corollary of such observations is that other biological processes are implicated in such severe

symptoms in NER defective syndromes. The identification of two novel missense XPG mutations in Brazilian patients, analyzed also for their responses to oxidative stress confirm this hypothesis. These observations will soon be submitted for publication (Berra et al, and Soltys et al, in preparation). Moreover, a genetic cluster of XP patients were discovered in the middle of Brazil, where more than 20 patients were diagnosed XP within a community of a 1,000 people. The very heterogeneous phenotype and the high frequency of affected patients make this community unique in the work. The mutation is being identified by exome sequencing, and a diagnostic PCR kit is being developed to help that population and look for similar mutations elsewhere in Brazil. The role of DNA repair pathways in cancer avoidance and premature aging were the subject of a review, we recently published (Moraes et al, 2012a) and another review updates the therapeutic strategies for the XP syndrome (Quayle et al, 2011).

Under the search for DNA repair pathways related to chemotherapeutic agents, we identified new roles for NER and translesion synthesis in cell protection from doxorubicin treatment, with a model that indicates which lesions are processed by these pathways (Moraes et al, 2012b). Moreover, we initiated two different strategies that may be useful for many of laboratories of the Human Genome Center, that is the development of nanoparticles containing chemotherapeutic agents (Vieira et al, 2011) and the use of lentivirus vectors expressing shRNA for gene silencing in human cells.

Finally, our expertise on DNA damage induced by UV light led to the development of a DNA based biodosimeter that help to measure the genotoxic potential of sunlight. The use of such dosimeter was recently reported (Schuch et al, 2011) and led to a patent application (Schuch and Menck, 2011). An update on biosensors to measure the intensity and damaging action of the UV component of sunlight was also concluded (Yagura et al, 2011).

STEM CELLS

a) Bone reconstruction

We are investigating 10 different biomaterials to try and identify those most suitable as adjuvants for bone reconstruction. At this point we have two promising biomaterials. We characterized three new sources of stem cells for bone reconstruction from palatal muscle, deciduous tooth, and fallopian tube.

Studying fibroblasts and mesenchymal stem cells (MSC) from Apert syndrome patients, we showed that the calvarial periosteum participates in the resynostosys process of these patients, and we identified a possible signaling pathway. These results may permit the identification of novel molecules contributing to bone regeneration and possibly the development of drugs that may reduce the ossification rate in these patients.

b) Muscular and neuronal regeneration

We have demonstrated that cord blood MSC can improve the clinical outcome of rats with Parkinson's disease, and that injection of fibroblasts can hinder or even efface the effect of the MSC. This research made the cover of FAPESP magazine.

Our results with murine and canine models for muscular dystrophy show that MSC from human adipose tissue seem better than from umbilical cord to regenerate muscle in affected animals and suggest that for clinical trials systemic injections need to be repeated. Our results were published in various journals (see list of publications), and also presented in local and international scientific meetings.

Our studies with murine embryonic and MSC stem cells are in progress, and the Myogenic Potential of Murine Embryonic Stem Cells in the Dmd/mdx Mouse Model for Duchenne Muscular Dystrophy was described in one book chapter (Ayub-Guerrieri et al., 2011), and the Myogenic Differentiation of ES Cells for Therapies in Neuromuscular Diseases: Progress to Date, were documented in a second book chapter (Almeida et al., 2011).

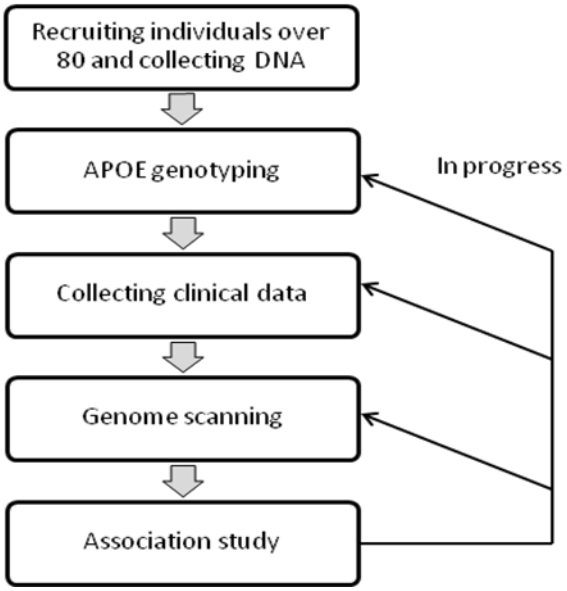
c) Stem-cell bank from families with patients affected by genetic disorders

Collecting samples for the Biobank is a continuous process. At this point, we already have 347 samples originating from a diverse array of tissues. These samples, and their pertinent medical information, are appropriately labeled, stored, and ready for research use. We also identified new sources of mesenchymal stem cells (MSC) from palate muscle and fallopian tube, and we are evaluating these cells therapeutic potential in pre-clinical trials.

PROJECT 80 PLUS

The goal of our Project 80+ is to map the genetic variability of individuals older than 80 whose cognitive function remains within healthy standards. To achieve this, we will recruit 1000 subjects. We have established partnerships with researchers from the Faculdade de Saúde Pública da USP (Profa. Maria Lúcia Lebrão) and from the Escola de Enfermagem da USP (Profa. Yeda A. de Oliveira Duarte). Recruiting has been ongoing in São Paulo since July 2010. All blood samples are sent to the HGRC for genomic DNA extraction.

At the moment, we have collected 932 DNA samples, with 324 of these coming from individuals older than 80 years. The first step in the genetic analysis is genotyping *APOE*, a gene whose variants are associated to the cognitive decline seen in Alzheimer's and other neurological diseases. We are also collecting clinical data on cognitive function, motor dominance, and behavioral and socio-economic aspects. We will then perform complete genome scans and try to identify genetic components associated to the measured clinical characteristics. A simplified flowchart representing the main steps of the project is depicted below.



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40. Vieira DB, Kim V, Petri DSF, Menck CFM, Carmona-Ribeiro AM. Polymer-based delivery vehicle for cisplatine. NSTI-Nanotech 2011, 3, 382-385.
41. Vieira NM, Valadares M, Zucconi E, Secco M, Bueno Junior CR, Brandalise V, Assoni A, Gomes J, Landini V, Andrade T, Lima BL, Caetano HVA, Vainzof M, Zatz M. Human Adipose-Derived Stromal cells injected systemically into GRMD dogs are able to reach the host muscle and express human dystrophin. Cell Transplant. 2011 Oct 14. [Epub ahead of print]
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45. Yeh E, Atique R, Ishiy FA, Fanganiello RD, Alonso N, Matushita H, da Rocha KM, Passos-Bueno MR. FGFR2 mutation confers a less drastic gain of function in mesenchymal stem cells than in fibroblasts. Stem Cell Rev. 2011 Nov 3.
46. Zucconi E, Vieira NM, Bueno Jr CR, Secco M, Jazedje T, Valadares MC, Suzuki MF, Bartolini P, Vainzof M, Zatz M. Pre-clinical studies with umbilical cord mesenchymal stromal-cells in different animal models for muscular dystrophy. J Biomed and Biotechnology 2011, 715251.

b) Books and Chapters in Books

1. Almeida CF, Ayub-Guerrieri D and Vainzof M. Myogenic Differentiation of ES Cells for Therapies in Neuromuscular Diseases: Progress to Date, Embryonic Stem Cells - Differentiation and Pluripotent Alternatives, Michael S. Kallos (Ed.), ISBN: 978-953-307-632-4, InTech, chapter 12, 2011, p. 227-242.
2. Ayub-Guerrieri D, Martins-Machado PCM, Onofre-Oliveira PCG, Pereira LV, Almeida CF, Lopes VF, Vainzof M. Myogenic Potential of Murine Embryonic Stem Cells in the Dmdmdx Mouse Model for Duchenne Muscular Dystrophy, Stem Cells in Clinic and Research, Ali Gholamrezanezhad (Ed.), ISBN: 978-953-307-797-0, 2011, InTech.
3. Okamoto OK. Molecular Biology of Cancer Stem Cells. In: Roberto Scatena; Alvaro Mordente; Bruno Giardina. (Org.). Advances in Cancer Stem Cell Biology. 1 ed. New York: Springer, 2011, p. 33-44.
4. Passos-Bueno MR, Fanganiello RD, Jehee FS. Craniosynostosis and Chromosomal Alterations. In: Muenke M (Bethesda, Md.); Kress W (Würzburg); Collmann H (Würzburg); Solomon BD (Bethesda, Md.). (Org.). Craniosynostoses:

Molecular Genetics, Principles of Diagnosis, and Treatment. 1 ed. Basel: KARGER, 2011, v. 19, p. 152-164.
5. Quayle C, Menck CFM, Lima-Bessa KM. Recombinant viral vectors for investigating DNA damage responses and gene therapy of Xeroderma Pigmentosum. Chapter 6, In: DNA Repair and Health, Ed. Sonya Vengrova, Intech Open Access Publisher, 2011, pp 145-174.
6. Zatz M. GenÉtica: Escolhas que nossos avós não faziam. Ed. Globo, 2011, 207 pages.

c) Abstracts

c.1) International Meetings

International Meetings/Conferences attended: 12
Abstracts presented: 44

1. Brito LA, Silva CBF, Rocha KM, Schlesinger D, Cruz LA, Bárbara LK, Aguena M, Bueno DF, Alonso N, Bertola DR, Meyer D, Passos-Bueno MR. The polymorphism rs642961 (IRF6) contributes differently to nonsyndromic cleft lip/palate susceptibility according to geographic regions of Brazil. In: 12th International Congress of Human Genetics/61st ASHG Annual Meeting, Montreal, Canada, 2011.
2. Bueno CRJ, Pantaleão LC, Zatz M Effect of AMPK/PPAR agonists and exercise training in mdx mice. In: 16th International Congress of the World Muscle Society, Portugal, 2011.
3. Cruz LA, Sunaga DY, Bueno DF, Kobayashi GS, Aguena M, Brito L, Passos-Bueno MR. Mesenchymal stem cells from cleft lip and palate patients have a different transcriptome and show deregulated pathways by comparison to controls. In: I International Meeting on Craniofacial Anomalies: Clinical Phenotype, Genes Related and New Perspectives, Bauru, SP, 2011.
4. da Silva Neto Jr JF, Netto LES. OhrA and OhrR are involved in the response of Chromobacterium violaceum to Organic Hydroperoxide Resistance. In: The 2011 Madison Molecular Genetics of Bacteria and Phages Meeting, Madison, WI, 2011.
5. D'Angelo CS, Koiffmann CP. Comprehensive analysis of novel disease-causing copy number variants. In: 12th International Congress of Human Genetics/61st ASHG Annual Meeting, Montreal, Canada, 2011.
6. del Castillo F, Gandia M, Pollak A, Hoefsloot L, Costa SMS, Batissoco AC, Sartorato EL, Mingroni-Netto RC, Lechowicz U, Mueller-Malesinska M, Skarzynski H, Skarzynsk PH, Moreno-Pelayo MA, Villmar M, Moreno F, Kremer H, Ploski R, Castillo I. A Multiplex Ligation-Dependent Probe Amplification (MLPA) assay specifically developed to detect novel deletions causing non-syndromic hearing impairment at the DFNB1 Locus. In: 8th Molecular Biology of Hearing and Deafness Conference, England, 2011.
7. Forbes J, Genesini T, Riolfi C, Lise L, Macedo E, Rudiger D, Zatz M. The effects of psychoanalysis in neuromuscular disorders. In: 16th International Congress of the World Muscle Society, Portugal, 2011.
8. Freitas E, Otto PA, Rosenberg C. Submicroscopic genomic alterations investigated by array-CGH in Finnish and Brazilian patients with Müllerian Alterations (MA). In: 12th International Congress of Human Genetics/61st ASHG Annual Meeting, Montreal, Canada, 2011.

9. Freitas E, Uehara DT, Mingroni Netto RC, Krepischi ACV, Rosenberg C. Whole-genome array-CGH screening in autosomal dominant sensorineural hearing loss patients detects two chromosomal alterations at 5q32 and 7q31.1. In: The European Society of Human Genetics Conference, Amsterdam, 2011.
10. Gandia M, del Castillo F, Pollak A, Hoefsloot L, Santos SC, Sartorato EL, Batissoco AC, Mingroni-Netto RC, Lechowicz U, Mueller-Malesinska M, Skarzynski H, Skarzynsk PH, Moreno-Pelayo MA, Villamar M, Moreno F, Kremer H, Ploski R, Castillo I. A Multiplex Ligation-Dependent Probe Amplification (MLPA) assay specifically developed to detect novel deletions causing non-syndromic hearing impairment at the DFNB1 Locus. In: The European Human Genetics Conference, Amsterdam, 2011.
11. Griesi-Oliveira K, Sunaga DY, Cruz LA, Vadasz E, Passos-Bueno MR. Expression analysis in dental pulp stem cells of idiopathic autistic patients reveals alterations in important processes for neurogenesis. In: 12th International Congress of Human Genetics/61st ASHG Annual Meeting, Montreal, Canada, 2011.
12. Ishiy FAA, Fanganiello RD, Aguena M, Amaral LBM, Martins MI, Passos-Bueno MR. Differences in osteogenic potential between human dental pulp stem cells and human adipose-derived stem cells *in vitro* and *in vivo.* In: Stem cell Biology Meeting, Cold Spring Harbor Laboratory, USA, 2011.
13. Janjoppi L, Luara M, Cavalheiro EA, Okamoto OK. Genes encoding chromatin remodeling proteins are down regulated in hippocampal cells during the course of epileptogenesis. In: 8th IBRO World Congress of Neuroscience, Florence, Italy, 2011.
14. Jazedje, Tatiana J, Bueno DF, Pimenta B, Caetano H, Czeresnia C E, Perin PM, Halpern S, Maluf M, Evangelista LP, Martins MT, Passos-Bueno MR, Zatz M. Human fallopian tube mesenchymal stem cells enhance bone regeneration in a xenotransplanted model. In: IX Congress of the International Society of Research in Stem Cells (ISSCR) Toronto, 2011.
15. Kobayashi GS, Cruz LA, Sunaga DY, Bueno DF, Ferreira SG, Meire A, Andrade-Lima L, Menck CF, Passos-Bueno MR. Dysregulation of DNA damage repair and cell cycle checkpoint control pathways as a mechanism for cleft lip/palate. In: 12th International Congress of Human Genetics/61st ASHG Annual Meeting, Montreal, Canada, 2011.
16. Lanzotti AA, Onofre-Oliveira PCG, Martins-Machado PCM, Martins-Bach AB, Feitosa LN, Vainzof M. The mdx/SJL mouse: a new double mutant model for neuromuscular disorders with mutations in the dystrophin and dysferlin genes. In: 16th International Congress of the World Muscle Society, Portugal, 2011.
17. Lezirovitz K, Batissoco AC, Lima FT, Auricchio MTBM, Dantas VGL, Oiticica J, Mingroni-Netto RC. Splice Donor site deletion in the *TECTA* gene causing autosomal dominant deafness in a Brazilian family. In: 8th Molecular Biology of Hearing and Deafness Conference, England, 2011.
18. Martins-Bach AB, Bloise AC, Rabani SR, Vainzof M. Application of NMR spectroscopy in the study of Duchene Muscular Dystrophy. In: 16th International Congress of the World Muscle Society, Portugal, 2011.
19. Martins-Machado P, Yamamoto LU, Onofre-Oliveira P, Ayub-Guerrieri D, Vainzof M. Heterozygous mutations in putative glycosiltranferase modulating the severity of the phenotype of muscular dystrophies. In: 16th International Congress of the World Muscle Society, Portugal, 2011.
20. Mingroni-Netto RC, Dantas VGL, Pardono E, Santos S. Autosomal dominant nonsyndromic deafness maps to DFNA18 in a Brazilian family. In: 8th Molecular Biology of Hearing and Deafness Conference, England, 2011a.

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22. Mingroni-Netto RC, Uehara DT, Freitas EL, Mazzeu JF, Auricchio MTBM, Tabith Jr A, Rosenberg C. A duplication in a patient with non-syndromic deafness reveals a novel candidate gene for deafness, *DOCK4*. In: 8th Molecular Biology of Hearing and Deafness Conference, England, 2011c.
23. Mitne Neto M, Machado-Costa M, Marchetto MCA, Bengtson MH, Silva HC, Joazeiro C, Oliveira ASB, Tsuda H, Bellen H, Muotri AR, Zatz, M. Reduced levels of vapb in motor neurons from als8 patients following i*PSC* fibroblasts reprogramming. In: IX Congress of the International Society of Research in Stem Cells (ISSCR) Toronto, 2011.
24. Morris MLM, Medina CN, Freitas E, Rosenberg C, Oliveira SF, Ferrari I, Mazzeu JF. 21,5 Mb Mosaic Pure Inverted Duplication of Chromosome 1q42.13qter. In: 12th International Congress of Human Genetics/61st ASHG Annual Meeting, Montreal, Canada, 2011.
25. Oiticica J, Barboza-Junior LCM, Batissoco AC, Lezirovitz K, Mingroni-Netto RC, Haddad LA, Bento RF. Viable Proliferating progenitor cells. 2011 American Academy of Otolaringology Head and Neck Surgery Annual Meeting & OTO Expo, 2011.
26. Onofre-Oliveira P, Martins-Machado P, Lanzotti A, Almeida C, Nogueira L, Ayub-Guerrieri D, Vainzof M. Myogenic potential of murine mesenchymal stem cells for therapy in progressive muscular dystrophies. In: IX Congress of the International Society of Research in Stem Cells (ISSCR) Toronto, 2011.
27. Pearson PL, Vianna-Morgante AM. Back to family values: Replacing GWAS for mapping menopause genes by a novel family approach. In: 15th International Workshop on Fragile X and other early-onset cognitive disorders, Max Planck Institute for Molecular Genetics, Berlin, Germany, 2011.
28. Pereira MCL, Secco M, Suzuki DE, Janjoppi L, Rodini CO, Torres LB, Araújo BHS, Cavalheiro EA, Zatz M, Okamoto OK. Contamination of mesenchymal stem-cells with fibroblasts accelerates neurodegeneration in an experimental model of parkinson s disease. In: IX Congress of the International Society of Research in Stem Cells (ISSCR) Toronto, 2011.
29. Pimenta MV, Horta BB, Discola KF, Netto LES. Biological characterization of the ahpR system from the phytopathogenic bacteria Xylella fastidiosa. In: International Course Redox Chemistry and Biology of Thiols, Montevideo, Uruguai, 2011.
30. Reydon AFC, Netto LES. Antioxidant Defenses of the peroxisome in Saccharomyces cerevisiae. In: International Course Redox Chemistry and Biology of Thiols, Montevideo, Uruguai, 2011.
31. Ribeiro CM, Moreira DP, Griesi-Oliveira K, Takahashi VNVO, Rosenberg C, Bertola DR, Vasdasz E, Passos-Bueno MR. Characterization of microchromosomal rearrangements in syndromic and nonsyndromic autistic patients using a custom CGH-microarray. In: 41st annual meeting of the Society for Neuroscience, Washington, USA, 2011.
32. Rodrigues TC, Krepischi ACV, Bertola DR, Kok F, Rosenberg C. The logistcs and result of array CGH diagnosis in mental retardation and congenital abnormalities in Brazil. In: 7th International DECIPHER Symposium - Wellcome Trust Sanger Centre, 2011, Hixton, Cambridge.
33. Secco M, Bueno Júnior C, Vieira NM, Jazedje T, Valadares M, Okamoto KO, Zatz M. The effects of igf-1 on the proliferation and myogenic differentiation of stromal

stem cells from human umbilical cord tissue. In: IX Congress of the International Society of Research in Stem Cells (ISSCR) Toronto, 2011.
34. Secco M, Vieira NM, Valadares M, Jazedje T, Okamoto OK, Zatz M. IGF-1 induces proliferation and myogenic differentiation of stromal stem cells from human umbilical cord tissue. In: IX Congress of the International Society of Research in Stem Cells (ISSCR) Toronto, 2011.
35. Shelton GD, Vieira N, Guo LT, Kunkel LM , Zatz M . Dystrophin-deficient muscular dystrophy in a pedigree of labrador retrievers without obvious clinical manifestations. In: 16th International Congress of the World Muscle Society, Portugal, 2011.
36. Tairum-Jr CA, Horta BB, Zara FJ, Oliveira MA, Netto LES. Investigating the Reactivity and structural transitions of the yeast thiol specific antioxidant protein 1. In: International Course Redox Chemistry and Biology of Thiols, Montevideo, Uruguai, 2011.
37. Vainzof M, Gurgel-Gianneti J, Bertola D, Pavanello RCM, Oliveira AB, Rosemberg C, Kok F, Martins-Bach AB, Almeida CF, Zatz M. A new form of myopathy associated with muscle hypertrophy, short stature, macroglossia and brachydactyly. In: 16th International Congress of the World Muscle Society, Portugal, 2011.
38. Velloso FJ, Correa JC, Tempalinni E, Azzi-Nogueira D, Grinberg LT, Chiavegatto S, Vianna-Morgante AM, Haddad LA. FMRP isoforms with an extended variable loop in the KH2 domain are in ribonucleoprotein complexes of rodent and human cerebral cortex. In: 15th International Workshop on Fragile X and other early-onset cognitive disorders, Max Planck Institute for Molecular Genetics, Berlin, Germany, 2011.
39. Vieira NM, Assoni A, Almeida C, Secco M, Brandalise V, Zucconi E, Camargo MM, Zatz M. Immunological properties of injected human adipose-derived mesenchymal stromal cells(hHASCs) undifferentiated and committed to myogenic phenotype. In: IX Congress of the International Society of Research in Stem Cells (ISSCR) Toronto, 2011.
40. Vieira NM, Moreira Y, Zucconi E, Valadares M, Vainzof M, Verjovsky-Almeida S, Zatz M. Microarray analysis of two exceptional Golden Retriever Muscular Dystrophy (GRMD) dogs with no dystrophin and a mild course. In: 16th International Congress of the World Muscle Society, Portugal, 2011.
41. Yeh E, Atique RFT, Alonso N, Matsushita H, Passos-Bueno MR. FGFR2 mutation confers less functional damage in adult human mesenchymal stem cells as compared to fibroblasts. In: I International Meeting on Craniofacial Anomalies: Clinical Phenotype, Genes Related and New Perspectives, Bauru, SP, 2011.
42. Yeh E, Atique RFT, Alonso N, Matsushita H, Passos-Bueno MR. The Effect of FGFR2 S252W mutation (Apert Syndrome) on adult human mesenchimal stem cells compared to fibroblasts. In: IX Congress of the International Society of Research in Stem Cells (ISSCR) Toronto, 2011.
43. Zatz M, Pavanello RC, Lazar M, Vainzof M .How to deal with pathological mutations in healthy persons? In: 16th International Congress of the World Muscle Society, Portugal, 2011.
44. Zatz M,Vieira NM,Valadares M, Zucconi E, Secco M, Bueno Junior CR, Brandalise V, Assoni A, Gomes J, Landini V, Andrade T, Vainzof M, Shelton GD. Pre-clinical studies with human adult mesenchymal stem-cells: what have we learned? In: 16th International Congress of the World Muscle Society, Portugal, 2011.

c.2) National Meetings

National Meetings/Conferences attended: 8
Abstracts presented: 28

1. Alegria TGP, Cussiol JRR, Miyamoto S, Mascio P, Netto LES. Ohr from Xylella fastidiosa Shows Great Affinity for Long Chain Fatty Acids Hydroperoxides. In: XL Reunião Anual da Sociedade Brasileira de Bioquímica e Biologia Molecular (SBBq), Foz do Iguaçu, 2011.
2. Alegria TGP, Discola KF, Cussiol JRR, Netto LES. Structural characterization of Ohr (Organic Hydroperoxide Resistance Protein) from Xylella fastidiosa in oxidized conformation. In: 21ª Reunião anual dos Usuarios do LNLS, 2011.
3. Bagini RH, Tairum-Jr CA, Netto LES, Oliveira MA. Investigation of structural and functional consequences of T44 and R146 substitutions in Saccharomyces cerevisiae Thiol specific Antioxidant 1 protein. In: I São Paulo Advanced School (ESPCA) on Redox Processes in Biomedicine, São Pedro, SP, 2011.
4. Bernardino-Cruz, D. Auricchio, MTBM; Lezirovitz, L e Mingroni-Netto, RC. Estudo de mutações dos genes *OTOF* e *MT-TS2* relacionados à deficiência auditiva. In: 19° Simpósio Internacional de Iniciação Científica da USP
5. Brito LA, Silva CBF, Rocha KM, Schlesinger D, Cruz LA, Barbara LK, Aguena M, Bueno DF, Alonso N, Bertola DR, Meyer D, Passos-Bueno MR. Contribution of marker rs642961 (IRF6) to nonsyndromic cleft lip/palate is weaker in Brazil and varies across geographic region. In: 57° Congresso Brasileiro de Genética, Águas de Lindóia, SP, 2011.
6. Cruz LA, Kobayashi GS, Sunaga DY, Bueno DF, Ferreira SG, Aguena M, Passos-Bueno MR. Gene expression analysis of cleft lip/palate stem cells reveals dysregulation of DNA damage repair pathway. In: 57° Congresso Brasileiro de Genética, Águas de Lindóia, SP, 2011.
7. Cruzeiro-Silva C, Gomes-Neto F, Rodrigues NL, Miyamoto CA, Pinheiro AS, Netto LES, Valente AP, Almeida FCL. Dynamics and Structural Characterization of Trx1 and Trx1 D24N mutant by Nuclear Magnetic Resonance. In: XL Reunião Anual da Sociedade Brasileira de Bioquímica e Biologia Molecular (SBBq), Foz do Iguaçu, 2011.
8. da Silva Neto Jr JF, Netto LES. Regulation of Genes Encoding Organic Hydroperoxide Resistance (Ohr) Proteins from Chromobacterium violaceum. In: I São Paulo Advanced School (ESPCA) on Redox Processes in Biomedicine, São Pedro, SP, 2011.
9. Fonseca ACS, Fonseca SAS, Bonaldi A, Vianna-Morgante AM. The complexity of a familial apparently balanced translocation revealed by combining a-CGH and FISH. 2ª Reunião Brasileira de Citogenética, Águas de Lindóia, SP, 2011.
10. Fontana AG, Yeda FP, Moro AM, Okamoto OK, Smaletz O. Análise de farmacocinética (PK) do anticorpo monoclonal Hu3s193 (rebmab 100) em mais de 4 doses semanais em estudo de fase II para pacientes com câncer avançado de ovário (CO), peritôneo (CP) ou tuba uterina (CTU), resistente/refratário a platina (RRP). In: XVII Congresso Brasileiro de Oncologia Clínica, 2011. Gramado, RS.
11. Kido LY, Santos VF, Oliveira MA, Netto LES. Mixed Disulfide Protein Complexes Between Thioredoxin and Peroxiredoxin. In: 21ª Reunião Anual dos Usuarios do LNLS, 2011.
12. Kobayashi GS, Cruz LA, Sunaga DY, Bueno DF, Ferreira SG, Aguena M, Passos-Bueno MR. Mesenchymal stem cells from cleft lip/palate patients exhibit an expression signature associated with dysregulation of cell cycle progression. In:

57° Congresso Brasileiro de Genética, Águas de Lindóia, SP, 2011.
13. Lezirovitz K, Batissoco AC, Lima FT, Auricchio MTBM, Dantas V, Oiticica J, Mingroni-Netto RC. Splice donor site deletion in the *TECTA* gene causing autosomal dominant deafness in a Brazilian family. In: São Paulo School of Advanced Science: Advanced Topics of Human Molecular Genetics, Campinas, SP, 2011.
14. Martins-Bach AB, Bloise AC, Rabani SR, Vainzof M. Metabolomic study of the mdx mouse. In: 57º Congresso Brasileiro de Genética, 2011. Águas de Lindoia, SP
15. Moreira, DP, Ribeiro, CM, Griesi-Oliveira, K, Lourenço, NCV, Takahashi, VNO, Vadasz, E, Bertola, D, Passos-Bueno, MR. Deletion in individuals with autism spectrum disorders. In: 57° Congresso Brasileiro de Genética, Águas de Lindóia, SP, 2011.
16. Moretti-de-Almeida G, Netto LES, Monteiro G. YMR134W is an essential ORF involved in iron homeostase in Saccharomyces cerevisiae. In: XL Reunião Anual da Sociedade Brasileira de Bioquímica e Biologia Molecular (SBBq), Foz do Iguaçu, 2011.
17. Nakahata AM, Suzuki DE, Okamoto OK. Knock down of e2f2 expression inhibits proliferation of human glioblastoma cells. In: 57º Congresso Brasileiro de Genética, 2011, Águas de Lindóia.
18. Nakamatsu EH, Discola KF, Monteiro G, Murakami MT, Netto LES. Structural characterization of the Thioredoxin reductase 2 protein. In: 21ª Reunião Anual dos Usuarios do LNLS, 2011.
19. Nakamatsu EH, Monteiro G, Discola KF, Murakami MT, Netto LES. Insights on the molecular aspects of species-specificity protein-protein interaction of the mitochondrial thioredoxin system from Saccharomyces cerevisiae. In: I São Paulo Advanced School (ESPCA) on Redox Processes in Biomedicine, São Pedro, SP, 2011.
20. Netto LES, Barros MH, Demasi M. Site-specific mutation of glutahtione - modified cysteines in the catalytic subunit of the proteasome from the yeast Saccharomyces cerevisiae. In: I São Paulo Advanced School (ESPCA) on Redox Processes in Biomedicine, São Pedro, SP, 2011.
21. Nonose RW, Lezirovitz K, Auricchio MTBM, Mingroni-Netto RC. Molecular Studies of *SLC26A4* gene in autosomal recessive deafness. In: 57° Congresso Brasileiro de Genética, Águas de Lindóia, SP, 2011.
22. Oliveira DP, Glezer I, Discola KF, Netto LES. Search for biological targets of grx1 and grx2 from Saccharomyces cerevisiae. In: I São Paulo Advanced School (ESPCA) on Redox Processes in Biomedicine, São Pedro, SP, 2011.
23. Peters MCC, Rezende L, Malvezzi A, Netto LES, Amaral AT. Conformational change of Ohr from Xylella fastidiosa by molecular dynamics simulations and validation. In: I São Paulo Advanced School (ESPCA) on Redox Processes in Biomedicine, São Pedro, SP, 2011.
24. Reydon AFC, Barros MH, Gladyshev VN, Netto LES. Peroxiredoxins and catalases cooperate to protect the yeast Saccharomyces cerevisiae in conditions of high fatty acid intake. In: I São Paulo Advanced School (ESPCA) on Redox Processes in Biomedicine, São Pedro, SP, 2011.
25. Simoes V, Silva GM, Netto LES, Demasi M. Proteasome redx modulation through S-glutathiolation increases proteolysis rates and modifies protein fragmentation. In: I São Paulo Advanced School (ESPCA) on Redox Processes in Biomedicine, São Pedro, SP, 2011.
26. Simoes V, Silva GM, Netto LES, Santos LFA, Gozzo FC, Demasi M. Redox Modulation of the Yeast 20S Proteasome Implies on the Generation of Diverse

Sets of Peptides. In: XL Reunião Anual da Sociedade Brasileira de Bioquímica e Biologia Molecular (SBBq), Foz do Iguaçu, 2011.

27. Suzuki DE, Nakahata AM, Okamoto OK. Suppression of human embryonic stem cell proliferation by RNA interference. In: 57º Congresso Brasileiro de Genética, 2011. Águas de Lindóia, SP.
28. Tairum-Jr CA, Santos VF, Kido LY, Netto LES, Oliveira MA. Catalytic Intermediates and Structural Aspects of Cytosolic Yeast Prx-Trx Protein Complexes. resumo em congresso nacional. In: XL Reunião Anual da Sociedade Brasileira de Bioquímica e Biologia Molecular (SBBq), Foz do Iguaçu, 2011.

d) Theses and Dissertations

1. Adriano Bonaldi. Estudo genético da síndrome de Silver-Russell. 2011. Dissertação (Mestrado em Biologia/Genética). Instituto de Biociências, Universidade de São Paulo
2. Ana Carla Batissoco. A conexina 26 e sua relação com outras proteínas do órgão de Corti. 2011. Tese (Doutorado em Biologia / Genética). Universidade de São Paulo.
3. Ana Carolina Fonseca. Caracterização de rearranjos cromossômicos aparentemente equilibrados associados a quadros clínicos. 2011. Dissertação (Mestrado em Biologia Genética). Instituto de Biociências, Universidade de São Paulo.
4. Carolina de Oliveira Rodini. Expressão de marcadores de células-tronco em meduloblastoma: correlação com prognóstico clínico. 2011. Dissertação (Mestrado em Neurologia / Neurociências). Universidade Federal de São Paulo.
5. Dinorah Zilberztajn. Estudo da expressão da miostatina em modelos murinos de doenças neuromusculares. 2011. Dissertação (Mestrado em Biologia) Programa Interunidades de Biotecnologia.
6. Erika Yeh. Estudo da contribuição molecular e celular do periósteo na craniossinostose da síndrome de Apert. 2011. Tese (Doutorado em Biologia / Genética). Universidade de São Paulo.
7. Estela Mitie Cruvinel. Estudo da expressão do gene UBE3A e do transcrito UBE3A anti-senso em neurônios derivados de células da polpa de dente de pacientes com síndrome de Angelman. 2011. Dissertação (Mestrado em Genética). Universidade de São Paulo.
8. Gerson Kobayashi. Análise do transcriptoma de células-tronco mesenquimais para o estudo da etiologia das fissuras lábio-palatinas não-sindrômicas. 2011. Dissertação (Mestrado em Biologia / Genética). Universidade de São Paulo.
9. Jorge Figueiredo Forbes. Uma nova abordagem psicanalítica no tratamento de famílias com pacientes afetados por doenças neurodegenerativas. 2011. Tese (Doutorado). Faculdade de Medicina da USP.
10. Karen Nogueira Coqueti. O cromossomo X e a deficiência mental no sexo masculino. 2011. Dissertação (Mestrado em Biologia Genética). Instituto de Biociências, Universidade de São Paulo.
11. Karina Oliveira Griese. Identificação de genes e vias associadas aos transtornos do espectro autista. 2011. Tese (Doutorado em Biologia / Genética). Universidade de São Paulo.
12. Larissa Fontes. Análise da expressão do gene FMR1 no ovário. Tese (Doutorado em Biologia Genética). Instituto de Biociências, Universidade de São Paulo.

13. Luciana Janjoppi. Alterações de expressão gênica no hipocampo e envolvimento da via TGFB1 na epileptogênese. 2011. Tese (Doutorado em Neurologia / Neurociências). Universidade Federal de São Paulo.
14. Luciana Licinio. Mapeamento de uma nova forma de Distrofia Muscular Tipo Cinturas de Herança Autosssômica Dominante caracterizada por fraqueza distal, lipodistrofia de face e catarata. 2011. Dissertação (Mestrado em Genética). Instituto de Biociências Universidade de São Paulo.
15. Luciano Abreu Brito. Identificação de Genes de Suscetibilidade às Fissuras Labiopalatinas Não Sindrômicas: Influência da Epidemiologia e da Estratificação Populacional. 2011. Dissertação (Mestrado em Biologia / Genética). Universidade de São Paulo.
16. Mariane Secco. Avaliação do potencial terapêutico de células-tronco mesenquimais do cordão umbilical humano associadas ao IGF-1 para Distrofias Musculares progressivasl. 2011. Tese (Doutorado em Biologia / Genética). Universidade de São Paulo.
17. Miguel Mitne Neto. Estudo de pacientes discordantes em doenças Neuromusculares: paraplegias espásticas e distrofias tipo cinturas. 2011. Tese (Doutorado em Genética). Instituto de Biociências Universidade de São Paulo.
18. Monize Lazar Magalhães. Desvio de segreção (meiotic drive): mutação no gene da calpaina ou em outro gene da região? 2011. Tese (Doutorado em Genética). Instituto de Biociências Universidade de São Paulo.
19. Natassia Moreira da Silva Vieira. Avaliação do potencial terapeutico de celulas tronco mesenquimais de tecido adiposo humano para doenças musculares progressivas.Tese (Doutorado em Genética). Instituto de Biociências Universidade de São Paulo.
20. Renata Soares Thiele de Aguiar. Estudos moleculares em família com defeitos de membros. 2011. Dissertação (Mestrado em Biologia / Genética). Universidade de São Paulo.
21. Vanessa Ferreira Lopes. Estabelecimento de parametros clínicos em modelos murinos de doenças neuromusculares. 2011. Dissertação (Mestrado em Biologia) Programa Interunidades de Biotecnologia.
22. Vanessa Luiza Tavares Romanelli. Identificação de mutações associadas à Sindrome Auriculo-Condilar. 2011. Dissertação (Mestrado em Biologia / Genética). Universidade de São Paulo.

e) Lectures and courses

1. Mingroni-Netto RC. Aula sobre “Surdez Genética Não-Sindrômica” para a disciplina de pós-graduação Genética Médica, BIO-5707, IBUSP, São Paulo, SP.
2. Mingroni-Netto RC. Comunicação oral “A duplication in a patient with non-syndromic deafness reveals a novel candidate gene for deafness, *DOCK4*” durante o 8th Molecular Biology of Hearing and Deafness Conference, Hinxton, Inglaterra., julho 2011
3. Mingroni-Netto RC. Entrevista ao programa “E aí, doutor”, da TV Record, sobre Aconselhamento Genético, apresentado em 4/10/2011.
4. Mingroni-Netto RC. Palestra no II Simpósio de Audiologia Infantil, com o título “A importância da Genética na deficiência auditiva”, em São José dos Campos- SP, em 7/11/2011
5. Mingroni-Netto RC. Palestra sobre “Surdez Hereditária” no Departamento de Fisiologia do IBUSP, São Paulo, SP.

6. Mingroni-Netto RC. Palestrante da mesa redonda “Biologia e Diversidade das Populações Humanas” com o tema “Variabilidade molecular em populações brasileiras” durante o 20º Encontro de Biólogos do Conselho Regional de Biologia, CRBio-1, Corumbá, MS., julho 2011
7. Mingroni-Netto RC. Palestrante no I Congresso Internacional de Surdez, implante coclear, próteses auditivas e cirurgicamente implantáveis e I Simpósio internacional de células-tronco e genética aplicada à surdez da FMUSP, com o título “Genética das Perdas Auditivas” (30/11/11 a 3/12/2011).
8. Mingroni-Netto RC. Palestrante no I Congresso Internacional de Surdez, implante coclear , próteses auditivas e cirurgicamente implantáveis e I Simpósio internacional de células-tronco e genética aplicada à surdez da FMUSP, com o título “Aconselhamento genético da Surdez”. (30/11/11 a 3/12/2011)
9. Mingroni-Netto RC. Palestrante no I Congresso Internacional de Surdez, implante coclear, próteses auditivas e cirurgicamente implantáveis e I Simpósio internacional de células-tronco e genética aplicada à surdez da FMUSP, com o título “Diagnóstico Molecular da Deficiência Auditiva” (30/11/11 a 3/12/2011)
10. Mingroni-Netto RC. Professora do mini-curso “Análises moleculares em doenças genéticas humanas“ durante o 20º Congresso de Biólogos do Conselho Regional de Biologia, CRBio-1, Corumbá, MS., julho 2011
11. Netto LES. Bacteria present distinct pathways to remove peroxides. Structural and biochemical characterization of antioxidant systems from *Xylella fastidiosa*. (26th March) Simposium: Thiol metabolism and redox regulation of cellular functions”. Casapuble Hotel, Punta Ballena, Maldonado, Uruguay.
12. Netto LES. Caracterização de proteínas antioxidantes com potencial aplicação biotecnologica para controle de doenças na agricultura, 2011. Palestra proferida na *III 4 BIOTEC (Four Biotec) Quatro Dias pela Biotecnologia* – UFSCAR. 01 a 04 de agosto de 2011.
13. Netto LES. Oxidative stress and thiol redox pools. Palestra proferida em *Plenary Section: Redox Process and degenerative diseases* durante o congresso *Free Radicals in Brazil 2011*, Hotel colina Verde, São Pedro SP, 13 a 21 de Agosto de 2011.
14. Netto LES. Thiol-containing proteins in antioxidant defense. (22nd March) International course “Redox Chemistry and Biology of Thiols” 21st March to April 1st 2011, Montevideo, Uruguay.
15. Okamoto OK. Stem cell therapy in neurodegenerative disorders: the experience in Parkinson's Disease. 2011.
16. Passos-Bueno MR. Conference Cleft lip and palate: novel signaling pathways. Março 12-15, 2011, Natal, RN, Brazil.
17. Splendore A. Curso de Comunicação Científica. March 31st, April 7th, April 14th, Centro de Estudos do Genoma Humano, SP, 2011.
18. Vainzof M. Animal models and protein studies in neuromuscular disorders. Laboratoire de RMN AIM – CEA. Institut de Myologie, Groupe Hospitalier Pitié-Salpêtrière, Paris, 04/2011.
19. Vainzof M. Modelos Animais para doenças neuromusculares: ajudando a entender mecanismos e testar terapias. Seminários do Depto. de Genética e Biologia Evolutiva, IB-USP, 11/2011.
20. Vianna-Morgante AM. “A Variabilidade Genética Populacional Associada a Doenças”. Mesa Redonda “Biologia e diversidade das populações humanas”. 20º

Congresso de Biólogos do CRBio-01 (20º ConBio), Centro de Convenções do Pantanal e UFMS / Campus do Pantanal, 17 - 20 de julho de 2011.

21. Vianna-Morgante AM. “Aconselhamento Genético” - 20º Congresso de Biólogos do CRBio-01 (20º ConBio), Centro de Convenções do Pantanal e UFMS / Campus do Pantanal, 17 - 20 de julho de 2011.
22. Vianna-Morgante AM. “Publicação de Trabalhos Científicos – Aspectos Éticos”. Mesa Redonda – “Plágio em Ciência”. 14ª Semana Temática da Biologia, Instituto de Biociências, USP, 29 de setembro de 2011.
23. Vianna-Morgante AM. “X-inactivation and X-chromosome mutations in intellectual disability”. Simpósio: “X-Chromosome Inactivation: Celebrating the 50 Year Anniversary of the Lyon Hypothesis” (coordenadora). 57º Congresso Brasileiro de Genética, Águas de Lindóia , SP, setembro 2011,
24. Zatz M. 6º Encontro de Inovação do CGP- Rio de Janeiro, 22 de setembro
25. Zatz M. 7th edition Global Meeting-Deauville-França-13-15 de outubro
26. Zatz M. Ciência e Tecnologia - 4a semana Ab nril do Conhecimento- 13 de setembro
27. Zatz M. International Congress on embryonic Stem Cells- México-, 20-23 de setembro
28. Zatz M. IX ISSCR International Society for Stem Cell Research-Toronto –Canadá 14-18 junho
29. Zatz M. Preclinical studies in muscular dystrophies. In: International Congress on embryonic stem-cells, Mexico, March 14-15. Invited speaker, 2011
30. Zatz M. Pre-clinical studies with human adult mesenchymal stem-cells: from *sjl* mice to grmd dogs. Lille, France, May 12 Invited speaker, 2011
31. Zatz M. Preclinical studies with human adult mesenchymal stem-cells:from SJL mice to GRMD dogs. 5th International Congress of mycology, Lille, France, 9 a 13 de maio
32. Zatz M. Stem cells investigation aiming cell therapy and as a tool to understand gene function in genetic disorders- Washington, FAPESP Week- 26 de outubro
33. Zatz M. Stem-cells in neuromuscular disorders. Mexico, 13 a 15 de Março, 2011.
34. Zatz M. WMS-XVI International Congress on Neuromuscular Disorders- Algavre, Portugal 18-22 de outubro
35. Zatz M. Workshop-Genotype-phenotype correlation in calpainopathies. Santa Mônica California- 27 de outubro
36. Zatz M. X Congress of Mediterranean Society of Myology-Pisa-Italia 28- 30 abril
37. Zatz M. I Simpósio Internacional de Células Tronco e Genética aplicada à Surdez. da FMUSP. “Pesquisas com Celulas-Tronco no Brasil” Desafios Atuais e Futuros – 30 de Novembro de 2011.
38. Zatz M. IV Escola de Verão Eurolatinoamericana de Miologia, na Associação Paulista de Medicina, 10 de Dezembro de 2011.
39. Zatz M. GenÉtica: como ela pode afetar nossas vidas? Seminários do Departamento de Genética e biologia evolutiva, 5 de dezembro de 2011.

AWARDS AND HONORS

Ana Carolina Fonseca. MSc student. Best work in Human Cytogenetics presented at the 2ª REUNIÃO BRASILEIRA DE CITOGENÉTICA (2nd. Brazilian Cytogenetic Meeting), Águas de Lindóia, SP, August 2011.

Mayana Zatz. Doutor Honoris Causa, Universidade autônoma do México, September 2011.

Mayana Zatz. Premio G. Conte - Mediteranean Society of Miology - Italy, April 2011.

PATENTS

1. Andre P Schuch and Carlos FM Menck, “Dosímetro biológico celular, método de dosimetria de uma amostra celular e uso do dosímetro” - protocolo 018110046690.

2. Mariane Secco, Mayana Zatz and Oswaldo K Okamoto, “Processo de diferenciação miogênica, meio de cultura para diferenciação miogênica e uso das células miogênicas” - protocolo 018110046446.

PART 2. EDUCATION/PUBLIC INFORMATION

I. Experimental classes at public schools

We offered two capacitation workshops for Elementary and Middle School teachers to prepare experimental laboratory classes in their schools (02/25 and 02/12/2011). The workshop benefitted 56 Biology teachers from 41 schools belonging to Ensino Norte 2 and Osasco school districts (Annex 1).

We furnished 40 schools with lab equipment for 6 different experimental classes. The equipment remained in each school for three weeks (Annex 2). These experimental classes benefitted around 28,000 students.

II. Instructional material

This year, 40 teachers from 27 schools were trained to use instructional material available at our loan stations at Educational Directories Norte 2, Osasco, and IB-USP (Annex 3).

III. USP goes to your School ("A USP vai a sua Escola")

- Development and implementation of the scientific exposition "A USP vai a sua Escola II – Luz e Vida" (Light and Life).
- Presentation of the exhibit "A USP vai a sua Escola II – Luz e Vida" at USP Science Fair in October 15-16 2011 at EE Álvaro Guião, São Carlos, SP.
- Presentation of the exhibit "A USP vai a sua Escola II – Luz e Vida" at "Semóptica", in October 21 and 22 2011, Shopping Center Iguatemi, São Carlos, SP.

IV. The Giant Cell

An installation of an eukaryotic cell, amplified 130,000 times, allows people to immerse into cell organelles and get an idea of the structure and the function of a living cell. The Giant Cell participated in three events:

- 57º Brazilian Congress of Genetics, in Águas de Lindóia, São Paulo, on September 1-2 2011. Attendance was 500 visitors.
- Goiás Science and Technology Week (1,381 visitors):
    - Pontifícia Universidade de Goiás, Goiânia, October 17-19 2011.
    - Universidade Estadual de Goiás, Palmares de Goiás, October 20-23 2011.
- Osasco Plaza Shopping, September 26 to October 1 2011, sponsored by Diretoria Regional de Educação de Osasco. Attendance was 2,272 visitors.

V. Project Living the University ("Vivendo a USP")

Our Center participated in the project "Vivendo a USP", Promoted by the Pró-reitoria de Cultura e Extensão, in July 11-15 2011, benefitting 100 schoolchildren from the district of Perus (Escola Estadual Florestan Fernandes, Escola Municipal de Ensino Fundamental Jardim da Conquista and Escola Municipal de Ensino Fundamental Candido Portinari).

The students participated in two activities: "Journey to the Center of the Cell" (guided visit to our giant cell), and "Discovering the Microscopic world", a hands-on

workshop in a microscopy laboratory. Three teachers from the aforementioned schools were trained to conduct the activities (07/16/2011).

VI. Other Activities

a) Science Divulgation Articles

1. O discurso do novo ministro de ciência e tecnologia- VEJA.COM. 7 de janeiro
2. Fertilização in vitro, prêmio Novel e células-tronco embrionárias. VEJA.COM. 14 de janeiro
3. Infidelidade e promiscuidade: genético ou ambiental? VEJA.COM. 21 de janeiro
4. Testes genéticos- VEJA.COM. 28 de janeiro
5. Kits de ciência: mais próximos da realidade- VEJA.COM . 4 de fevereiro
6. Obesidade: a culpa é dos genes ou doa hábitos alimentares? VEJA.COM. 11 de fevereiro
7. Exames genéticos e incesto- VEJA.COM. 18 de fevereiro
8. Genética e incesto. Quando a verdade pode ser benéfica? VEJA.COM. 25 de fevereiro
9. Reino Unido lançará kits de testes genéticos pré-concepção VEJA.COM. 4 de março
10. Novo teste pré natal para síndrome de Down- VEJA.COM. 11 de março
11. Incesto:os riscos genéticos dos descendentes- VEJA.COM. 18 de março
12. O que atrai o espermatozóide? VEJA.COM. 25 de março
13. Trissomia do X. VEJA.COM. 1 de abril
14. Fertilização assistida: três é demais. VEJA COM. 8 de abril
15. Viciados em cafeína- VEJA.COM. 15 de abril
16. Esquizofrenia, bullying e células-tronco- VEJA.COM. 22 de abril
17. DNA para todos; estamos preparados? VEJA.COM. 28 de abril
18. Células-tronco e Parkinson:benefícios e riscos- VEJA.COM. 5 de maio
19. Doença de Alzjeimer:identificados novos genes de risco- VEJA.COM. 12 de maio
20. Células-tronco reprogramadas são rejeitadas- VEJA.COM. 20 de maio
21. Leitura “errada” ou programada? VEJA.COM. 27 de maio
22. Telômeros podem realmente prever a sua longevidade?- VEJA.COM. 3 de junho
23. Transdiferenciação: como fazer um neurônio- VEJA.COM. 9 de junho
24. Células-tronco podem consertar um coração partido- VEJA.COM. 16 de junho
25. Células-tronco abrem novos caminhos na esclerose lateral amiotrófica- VEJA.COM. 26 de junho
26. Charles Sabine: o advogado dos pacientes. Veja.com. VEJA.COM. 30 de junho
27. Como é a profissõa de um cientista- VEJA.COM. 7 de julho
28. Autismo: o que mostra um grande estudo de gêmeos- VEJA.COM. 14 de julho
29. Alcoolismo: genético ou ambiental- VEJA.COM. 21 de julho
30. Alzheimer: uma nova abordagem terapêutico- 28 de julho
31. Longevidade; de que depende. VEJA.COM. 4 de agosto
32. Doença de Alzheimer: o que mostra um grande estudo de gêmeos. VEJA.COM. 11 de agosto
33. A busca do filho perfeito: quais são os limites VEJA.COM. 18 de agosto
34. Esclerose lateral amiotrófica- VEJA.com-25/08
35. Bioengenharia de Tecidos: O futuro dos transplantes- VEJA .com-02/09
36. Meu livro ”Gen Ètica: Escolhas que nossos avós não fizeram”-VEJA.com 08/09
37. Cada cabeça uma sentença-VEJA.com-15/09
38. Doutor “Honoris Causa” VEJA.com-22/09

39. Planeta dos Macacos- a Origem-VEJA.com-29/09
40. Prêmio"Para Mulheres na Ciência"-VEJA.com-06/10
41. Camundongo transgênico abre novos caminhos para compreensão do autismo-VEJA.com- 14/10
42. Meu diálogo com Michael Chorost-VEJA.com-21/10
43. Projeto brasileiro estudará DNA de idosos saudáveis para aumentar longevidade da população-VEJA.com 26/10
44. Doença de Alzheimer: como ocorre o depósito de placas amilóides? VEJA.com 27/10
45. Genética da Obesidade e atividade física-VEJA.com 03/11
46. A placenta protege o cérebro do feto-VEJA.com 11/11
47. Mais um passo-VEJA.com -17/11
48. É possível retardar o envelhecimento-VEJA.com-24/11
49. O que revelam nossos cérebros- veja.com. 1/12
50. Célula-tronco da polpa dentária promove regeneração da medula espinhal em ratos. Veja.com. 8/12
51. Hemofilia B é tratada com terapia gênica. Veja.com. 15/12
52. Rir é o melhor remédio. Veja.com. 23/12

b) Event planning:

Koiffmann CP, Yassuda YY, Scheepmaker DS, Vainzof M. Prêmio Oswaldo Frota-Pessoa de Incentivo à Iniciação Científica. 2011.

Vainzof M. International symposium: Ressonância Magnética Nuclear (RMN) e técnicas de imagem para avaliação funcional de modelos animais em protocolos terapêuticos.São Paulo, 8/2011.

Vainzof M. Member of the organizing committee for the Curso Internacional de Miologia - EVELAM2011. São Paulo, 12/2011.

Annex 1

Table 1. Capacitated teachers and partner schools for the project “Experimental Classes in Schools” 2011

School	Teacher	District
E E Alberto Cardoso de Mello Neto	Andrea Valete Machado	Norte 2
E E Albino Cesar	Cristina Marçal da Silva Braga	Norte 2
E E Alfredo Inácio Trindade	Maria Lucia dos Santos	Norte 2
E E Amenaíde Braga de Queiroz	Hosana Correa Luz Pastore	Norte 2
E E Antonio José Leite	Liliana Martins	Norte 2
E E Assis Jose Ambrosio	Fabio Rodrigues Coelho dos santos	Norte 2
E E Prof. Carlos de Laet	Luciana Lucas de Ameida	Norte 2
E E Ministro Dilson Funaro	Regina Aparecida Tellles	Norte 2
E E Elza Saraiva Monteiro	Meire Pereira de França Mariana Gabriela Piaba	Norte 2
E E Eurico Figueiredo	Renato Sinnhofer Sugimotto	Norte 2
E E Francisco Voccio	Cristina Marçal Da Silva Braga	Norte 2
E E João Batista Alves da Silva	Daniela Duarte Costa Silva Vanderley R Santos	Norte 2
E E Johann Gutenberg	Lucineide Pereira da Rocha Silva	Norte 2
E E Doutor Justino Cardoso	Célia Regina Vicentine	Norte 2
E E Pedro Alexandrino	Cleusa Trovão	Norte 2
E E Philomena Baylão	Ana Maria Marcondes de Jesus Ana Paula dos Santos	Norte 2
E E Sebastiao de Souza Bueno	Gianni Messias de Castro	Norte 2
E E Silva Jardim	Elaine Rodrigues Cides	Norte 2
E E Veridiana Camaccho Gomes	Mariana Pereira	Norte II
E E Antonio Raposo Tavares	Regina Cely feres Hadad	Osasco
E E Benedito Caldeira	Cintia Roccini	Osasco
E E Jardim Cipava II A	Cassia Marquioreto Nabarro Kelly Cristina S Belmonte	Osasco
E E Claudinei Garcia	Patrícia Aparecida de Moura Monica Cristina Silva Jesus	Osasco
E E Ernesto Thenn de Barros	Ester Alves Correia Vânia Dias Flauzino de Miranda	Osasco
E E Prof. Fernando Buonaduce	Marli Cunha Alves Eneida De Fernandes	Osasco
E E Francisco Casabona	Maria Tereza da silva G Nazato	Osasco
E E Gastão Ramos	Alessandra Brito Santos Vânia Aparecida Veneruchi	Osasco
E E Jd. Santa Maria	Maria Ângela da Silva Denise da Silva Mendes Izilda Aparecida da Silva	Osasco
E E João Batista de Brito	Cleonice Maria de A. do Nascimento	Osasco
E E José Edson M. Gomes	Andrea Ferreira da Silva Carlos Alberto Ramos	Osasco
E E Jose Geraldo Vieira	Maria Catarina Fernandes	Osasco
E E José Liberatti	Denise da silva Mendes	Osasco
E E José Ribeiro de Souza	Leandro Francisco Couto	Osasco
E E Josué Benedito Mendes	Venancio Lopes da Cruz Filho	Osasco
E E Leonardo Vilas Boas	Sergio Suematsu	Osasco
E E Luci Anna Latorre	Sonia Regina Menezes de Oliveira Kátia Cristina Guerreiro Carraro	Osasco

E E Major Telmo Coelho Filho	Márcia Virginia Mendes Urban Vinicius de Oliveira Almeida	Osasco
E E Neuza de Oliveira Prévide	Marisa Sanches Iracy Vieira de Almeida	Osasco
E E Orlando Geríbola	Vânia Maria Garcia Giane Coração Campos	Osasco
E E Educador Paulo Freire	Aparecida Lira	Osasco
E E Tarsila do Amaral	Roberto Massaru Ueda	Osasco

Annex 2 – Experimental classes in Public Schools

EE Alfredo Inácio

EE Philomena Baylão

Annex 3 – Teacher Capacitation in the usage of instructional material. Osasco Educational Directory.

PART 3. TRANSFER OF TECHNOLOGY/ TECHNOLOGY APPLICATIONS

This section will include the main activities done in the last year regarding Genetic Counseling (GC), genetic testing, sequencing and microsatellite analysis services. These activities involve the participation of all the seven main researchers of the CEPID project, as follows: Angela Vianna-Morgante, Carla Rosenberg, Célia Koiffmann, Maria Rita Passos-Bueno, Mariz Vainzof, Regina C. Mingroni-Netto, and Mayana Zatz. Also, the neurologist Fernando Kok, and clinical geneticists Rita de Cassia Pavanello and Debora Bertola, all collaborators of CEPID, contribute to these activities. Prof. Paulo A. Otto, from the Department of Genetics and Evolutionary Biology, also collaborates to the Genetic Counseling service.

a) Genetic Counseling at CEGH

We offer this service for 6 main groups of disorders: neuromuscular (M Zatz, M Vainzof, F Kok, RC Pavanello), mental retardation - syndromic and non-syndromic forms (A Vianna-Morgante, C Rosenberg, PA Otto), developmental disorders associated with behavior disturbances and/or obesity (C Koiffmann), hearing diseases (RC Mingroni-Netto), craniofacial syndromes (MR Passos-Bueno) and autism (MR Passos-Bueno). In 2011 we counseled approximately 2,000 patients, in accordance to previous years. Genetic testing was offered in all necessary cases, as their results are critical for estimation of genetic recurrence risks, management and follow up of patients, while Genetic Counseling (GC) was offered to all of them.

b) Genetic Counseling at other regions of the country

We have maintained our partnership with Operation Smile, which offers surgery repair for clefting patients in different regions of the country. This year, we have evaluated and offered GC for more than 400 families with cleft lip and palate patients ascertained in 3 different regions of the country (Rio de Janeiro – RJ, Maceio - AL, and Santarém - PA).

c) Database of the Genome Center

Some members of the CEGH are currently using the Laboratory Management Information System (LIMS) software developed by IME-USP to input clinical and laboratory data (http://zen.genoma.ib.usp.br). This system allows genetic tests to be traced in a stepwise fashion, from the patient's first visit (registration) to the final test results. This LIMS software is being developed by Dr. João E. Ferreira and his team, at the Institute of Mathematics/USP in collaboration with CEPID/CEGH-USP.

d) Other activities and interactions

Our Center also interacts with patients/parents associations such as the Brazilian Muscular Dystrophy Association (ABDIM), Fragile X, Prader-Willi, Angelman, and Cleft lip/palate Associations. Through ABDIM we established an important partnership with the Secretaria da Saúde de São Paulo to support genetic tests aiming diagnosis and genetic counseling as well as management to families with affected members by neuromuscular disorders. Through this program we evaluated 500 patients from the state of Sao Paulo, and performed about 300 molecular tests for the diagnosis of NMD.

We established two new partnerships with INCTs (Instituto Nacionais de Ciência e Tecnologia), INCT – Comportamento, Cognição e Ensino and INCT – Instituto Nacional de Pesquisa do Desenvolvimento) which will complement the care offered to our autistic patients. Finally, we have a significant interchange of information about genetic tests and genetic counseling with the general public through e-mails.

e) Sequencing service and diagnostic tests for the general community

In 2011, we had a slight increase in the demand for sequencing reactions (27,952 injections) as compared to last year, whereas the demand for microsatellite analysis (wich includes MLPA reactions) doubled, amounting to 48,694 injections. The income generated from these services were R$ 494,208.90 and R$ 174,464.00, respectively. Genetic tests ordered by Medical Geneticists or other Diagnostic laboratories has remained in the range of 300, the same as last year. The income obtained from our service has been used to pay salaries for technicians and secretarial assistants, and also for equipment maintenance.

We will expand our services with the acquisition of a next-generation sequencing machine and a platform for multiplex genotyping (BeadXpress, Ilumina).

f) Implementation of a Quality System in the DNA Diagnostic Lab

To ensure the quality of our diagnostic facility and meet international standards, we implemented a Quality System in our diagnostic lab. We documented all of our Standard Operational Procedures, appointed a Quality Manager, and implemented test traceability, double checking of results and a system to deal with complaints. We contracted the European Molecular Genetics Quality Network as an external quality control for assessment of test quality and reliability, in addition to our in-house quality control. Our goal is constant improvement of our service. The exams under our Quality System are: Cystic Fibrosis, Spinocerebellar Ataxia (SCA1, SCA2, SCA3 and SCA6), Spinal Muscular Atrophy, Duchenne/Becker Muscular Dystrophy, Rett syndrome and Velocardiofacial syndrome.

g) Partnership for providing Comparative Genomic Hybridization tests

The Human Genome Center has in 2004 implemented in Brazil for the first time the technique of comparative genomic hybridization based on arrays (array-CGH), which has a much higher sensitivity for detecting submicroscopic chromosome deletions and duplications than classical cytogenetics, resulting in much higher diagnostic yield in cases of unexplained developmental delay/intellectual disability (DD/ID), autism spectrum disorders (ASD) or multiple congenital anomalies (MCA) than karyotyping. Albeit in 2010 the consortium from the *International System of Array Nomenclature* (ISCA - https://www.iscaconsortium.org/) has proposed the use of array-CGH as the first-tier cytogenetic diagnostic test for patients with DD/ID, ASD, or MCA, and that already being the standard procedure in most developed countries, no clinical genetic laboratory in Brazil performs the test; the few laboratories that offer the exam either send the material abroad or to our own facility. However, the demand for the test is rapidly increasing, and a structure with more commercial support and high-throughput possibilities is needed. The company Deoxi Biotecnologic, thanks to financing from FINEP, has all the equipment and qualified technical personal necessary for performing the test. As stated in the enclosed letter from Deoxi, we are planning to join them, and provide the specific know-how that will allow us, in a combined effort, to offer the exam to a larger part of the population in need.