

HUMAN GENOME RESEARCH CENTER (HGRC)

**Instituto de Biociências
Universidade de São Paulo
FAPESP/CEPID 98/14254-2
PUBLICAÇÕES 2008**

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Book chapters

- 1- Passos-Bueno, M. R. ; SERTIE, A. L. ; JEHEE, F. S. ; FANGANIELLO, R. ; Yeh E . Genetics of craniosynostosis:genes, syndrome, mutations and genotype-phenotype correlations.. In: D. P. Rice. (Org.). Craniofacial sutures:Development Disease and Treatment. 12 ed. Basel: Karger, 2008, v. 12, p. 107-143.
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- 3- Vainzof M, Kossugue PM. Estudos genéticas na Hipertermia Maligna e Miopatia de Central Core. In. Silva HMC - Hipertermina Maligna, 2008
- 4- Zatz M, Vainzof M. Distrofias Musculares Progressivas. In Beçak W. Genetica Medica. Ed. Manole, 2008, (in press)

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- 1- Gifalli-lughetti C, Koiffmann CP. Microdissection of the Y chromosome of *Brachyteles arachnoides* (Primates, Platyrrhini) using the Eppendorf TransferMan® NK 2. Applications (Eppendorf), Note 191, November 2008.
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PART 3: TRANSFER OF TECHNOLOGY/ TECHNOLOGY APPLICATIONS 2008

We have been offering the following services since 2003: DNA sequencing, microsatellite genotyping, molecular diagnostic tests of genetic disorders, and scanning of cDNA microarrays. The sequencing service is under the responsibility of Vanessa Naomi, while the microsatellite and scanning activities are under the responsibility of Katia Rocha. We currently offer more than 30 genetic tests, which are being conducted at the core laboratory at the Genome Center (Drs. MR Passos-Bueno, Fernanda S. Jehee, Martha Lima and Katia Rocha), and by six research groups (Drs. Angela Vianna-Morgante, Carla Rosenberg, Célia Koiffmann, Mariz Vainzof, Regina C. Mingroni-Netto, and Mayana Zatz). Details of these services are available at http://genoma.ib.usp.br/index_exam_diagnosticos.php. Below we present the summary of main activities of 2008:

Development of a bioinformatic system: Software to evaluate the workflow of DNA analysis is still under development by Dr. João E. Ferreira and his team, at the Institute of Mathematics/USP in collaboration with CEPID/CEGH-USP. We had spent a lot of time during this last year testing the software and a significant progress has been achieved. The software can be divided in three parts: input of data of patients, control of exams, workflow of DNA genetic tests. First part is ready and its use will be started in March/09.

Genetic counseling and diagnostic tests provided to families evaluated at the HGSC: Since 2000, more than 13,000 families were seen and received Genetic Counselling (GC). More than 14,000 genetic tests (DNA, chromosomes and protein analysis) have been performed. Their results are critical in GC, for estimation of genetic risks, identification of at-risk carriers, management and follow-up of patients.

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 and more recently with Petrobrás, for performing diagnostic tests and management of patients with neuromuscular disorders. We also have established collaboration with the Institute of Lacanian psychoanalysis (IPLA-Instituto de Psicoanálise Lacaniana) directed by the psychoanalyst Jorge Forbes, in order to evaluate the effect of psychoanalytic approach in the management of neuromuscular disorders,

New tests: After one year of testing, a new diagnostic tool using MLPA approach has been set up for diagnosis of congenital malformations associated with neurodevelopment delay (information available at <http://genoma.ib.usp.br>). Use and introduction of new tests using MLPA technique are being coordinated by Dr. Fernanda S. Jehee.

Sequencing service and diagnostic tests for the general community: In 2008, we obtained an income of R\$227.000,00 (US\$100.000,0) with the sequencing service, which is comparable to the income of 2007. About 100 genetic tests ordered by clinicians outside the CEGH were performed resulting in an income of R\$50.000 (~US\$25.000,00), a comparable value achieved in 2007.

Audit in the DNA diagnostic lab: A technical audit was performed in our Molecular Genetics Laboratory by Alexander Kneppers (Leiden University, Department of Human Genetics, Netherlands), who is an expert in this field. Dr. Kneppers carried out a 5 days audit. He followed and discussed in detail the complete flow of samples from acceptance of patient material to reporting of the findings to the

referring physician. The general organization, accommodation and environmental conditions, laboratory equipment and its maintenance have been discussed as well. A report has been generated and a top priority list was composed. We have already started the changes in accordance to this list.

Proposal for 2009:

- a) Introduce routinely the use of MLPA tests for congenital malformations;
- b) Introduce part 1 of software CEGH and conclude its part 2.
- c) Introduce rules and organization in the laboratory for genetic tests following the audit report
- d) Courses and other activities to advertise the genetic tests.

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Main education/diffusion activities in 2008

1. High School visiting program -

- a. **USP goes to your School** – A project to reach the students at their school. In a partnership action, the HGRC (Human Genome Research Center) and CEPOF (Group of Optics and Photonic from Physics Institute of São Paulo University, São Carlos) developed an itinerant exhibition focused on Stem Cells and Optics with interactive exhibits and panels. A van was adapted to transport the exhibition to schools from São Paulo, São Carlos and adjacent cities. In São Paulo, 33 Biology and Physics teachers from 19 public schools were instructed on how to use the instructional materials and advised on how to explore the conceptual content of the subjects presented to their classes (annex 1 and 2). A total of 12,600 students in São Paulo were reached by this program in 2008. Two fellows from the program “Ensinar com Pesquisa” carried out an evaluation. The behavior of teachers and students were followed in classes after the exposition had visited the schools; the impression and opinions of more than 1,000 students were collected. The results are being used to improve the visiting program and will be soon published.
- b. **Partnership with Estação Ciência (Science Station)** – The installation of a Cell Biology Laboratory in Estação Ciência was started in 2008 and will be functioning in March 2009. Twenty students will be attended per hour. Two different sets of activities were prepared to attend the public: “Discovering the microscopic world” will be offered to children from 10 to 14 years and the general public and “Observing cell from humans and plants”, a program to high school students. The optical material was financed by HGRC (Fapesp) and the educational/scientific divulgation activities will be oriented by HGRC.
- c. **Participation in the Exposition “Genomic Revolution”** – February 29th to July 13th, 2008, Ibirapuera Park, São Paulo. Eliana Dessen, was one of the Brazilian Exposition Curators, participated in the production of the Educational Material directed to teachers and to students and in the training of the exhibition mediators, with the collaboration of Regina Célia Mingroni Netto,

Other activities

- 51 Science divulgation articles (annex 3)
- 36 Conferences and lectures (annex 4)
- 6 missions of the Program “Sorriso” – Maria Rita Passos participated of Genetic Counseling of nearly 200 families having members affected by cleft lip or cleft palate and gave seminars about the importance of the counseling procedure.
- Fortaleza, Maceió, Barbalha, Curitiba, Santarem and Rio de Janeiro
- 5 Participations in Events of Education/diffusion and 2 symposiums (annex 5)
- 1 Meeting Organization (annex 5)

Annex 1 –Schools Visited by the exposition “A USP vai à sua Escola”

Schools Visited	Date
E E Julio Pestana	Abril 14 th to 16 th 2008
E E Albino Cesar	Abril 16 th to 23 th 2008
E E Carlos de Laet	Abril 23 th to 25 th 2008
E E Paul Hugo	Abril 28 th to 30 th 2008
E E Profa. Raquel Assis Barreiros	June 04 th to 06 th 2008

E E Sebastião de Souza Bueno	June 06 th to 11 th 2008
E E João Ligabue Cônego	June 12 th to 13 th 2008
E E Dr. Alberto Cardoso de Mello Neto	June 16 th to 19 th 2008
E E Leonidas Paiva	June 19 th to 20 th 2008
E E Justino	June 23 th to 24 th 2008
E E Profa. Carmosina Monteiro Vianna	June 25 th to 27 th 2008
E E Antonio José Leite	September 02 nd to 05 th 2008
E E Ministro Dílson Funaro	September 08 th to 09 th 2008
EE Assis Ambrósio	September 10 th to 11 th 2008
E E Castro Alves	September 15 th to 16 th 2008
E E Prof. Sergio da Costa	September 17 th to 18 th 2008
E E Angelo Bortolo	September 22 th to 23 th 2008
E E Amenaíde	September 24 th to 26 th 2008
E E Victor dos Santos Cunha	September 29 th to 30 th 2008

Annex 2 – Teacher capacitated to deal with the Exposition “A USP vai à sua Escola”

Capacitated Teachers	Area
Cristina Ayako Koga Inoue	Física
Luciana Rigamonti	Física
Vanderlei de Lima França	Física
Maria Cecília Leôncio Alves	Física
Paulo Roberto Wandeley	Física
Orlando Araujo de Oliveira	Física
Cristina M da Silva Braga	Biologia
Sandra Pereira Magalhães	Biologia
Alexandra Guedes Carames	Biologia
Karine Morales M Chagas	Biologia
Cristina Tortorelli	Biologia
Maria da Graça Sapage Estácio Gaspar	Biologia
Fátima Solange Riego Lavorente	Biologia
Maria Elizabeth Gasparotto Locci	Biologia
Vera Lúcia Pirre de Castro	Biologia
Luciana Lucas de Almeida	Biologia
Rosa Maria Megna	Física
Marisa Puentes Hamasaki	Física
Sandra Queiroz Gonçalves	Física
Amme Mourad	Física
Emerson Pires Nunes	Física
Sandra Regina Teixeira	Física
Thadeu Pereira do Nascimento	Física
Maria Ines Ferreira Pita Tamborelli	Física e Biologia
Andrea Valet Machado	Biologia
Hosana Correa Luz Pastore	Biologia
Acácio Martins G Jr.	Biologia
Hamilton S. João	Biologia
Cristina Tortorelli	Biologia
Andrea dos Santos Garcia	Biologia
Sandra Aparecida da Silva Correa	Biologia
Paula Galvão Alves	Biologia

Annex 3 - Science Divuligation articles

- Zatz, M. - Grupos sanguíneos e paternidade. Veja.com, January 10th
Zatz, M. - Talassemia e seleção de embriões. Veja.com. January 17th
Zatz, M. - As células embrionárias reprogramadas poderão originar clones humanos? Veja.com. January 24th
Zatz, M. - Ciência, religião e vida. Veja.com. 3 January 31st
Zatz, M. - Religião e ensino de ciência - Veja.com. February 07th
Zatz, M. - Embriões podem doar células sem serem destruídos - Veja.com. February 14th
Zatz, M. - Fertilização “in vitro” - Veja.com. February 21st
Zatz, M. - Em quem acreditar? Veja.com. February 29th
Zatz, M. - Lisa e o STF - Veja. Com. March 5th
Zatz, M. - Os novos pecados e o prêmio Bertha Lutz - Veja. Com. March 12nd
Zatz, M. - O medo da comercialização - Veja. Com. March 19th
Zatz, M. - Clonagem terapêutica e Mal de Parkinson - Veja. Com. March 26th
Zatz, M. - Onde começa a vida? - Veja. Com. April 5th
Zatz, M. - Células-tronco no líquido amniótico - Veja. Com. April 10th 1
Zatz, M. - Experiência de vida - Veja. Com. April 18th
Zatz, M. - Células-tronco embrionárias humanas desvendam os segredos do coração - Veja. Com. April 25th
Zatz, M. - O caso Isabella: a genética explica? Veja. Com. May 2nd
Zatz, M. - O que ocorre com gêmeos univitelinos - Veja. Com. May 09th
Zatz, M. - A teoria da evolução - Veja. Com. May 16th
Zatz, M. - Embriões híbridos e irmãos salvadores - Veja. Com. May 23th
Zatz, M. - A vitória no Supremo - Veja. Com. May 30th
Zatz, M. - Cobaías humanas - Veja.com. June 7th
Zatz, M. - Casamento entre primos - Veja com. June 14th
Zatz, M. - Sobre as cobaías humanas- Veja com. June 21st
Zatz, M. - Técnica de clonagem - Veja com. June 28th
Zatz, M. - Seleção de embriões e fábrica de gênios - Veja com. July 3th
Zatz, M. - Células-tronco e camundongos distróficos - Veja com. July 10th
Zatz, M. - Quando começam os tratamentos? Veja com. July 17th
Zatz, M. - Humanos não são cobaías. Veja com. July 24th
Zatz, M. - O Conceito de vida e de ser humano. Veja com. July 31st
Zatz, M. - Doping genético: ético ou não ético? Veja com. August 7th
Zatz, M. - O que é autismo? Veja com. August 14th
Zatz, M. - Genoma e psicanálise. Veja com. August 21st
Zatz, M. - Crime contra mulher. Veja com. August 28th
Zatz, M. - Fetos anencéfalos não podem doar órgãos- Veja.com. September 4th
Zatz, M. - Drogas podem potencializar os efeitos do exercício- Veja.com. September 11st
Zatz, M. - Injeções de esperança: um negócio da china- Veja.com. September 18th
Zatz, M. - Educação: a usp planta mais uma sementinha- Veja.com. September 25th
Zatz, M. - Não gostei. Veja.com. 02 de outubro
Zatz, M. - Paralisia cerebral. Veja.com. October 09th
Zatz, M. - Você quer saber se ficará calvo? Veja.com. October 16th
Zatz, M. - Diagnóstico genético pré-implantacional para escolha de sexo ou surdez. Veja.com. October 23th
Zatz, M. - Mal de Alzheimer: genético ou adquirido? Veja.com. October 30th
Zatz, M. - Testes em embriões: muita informação e pouco conhecimento. Veja.com. November 06th
Zatz, M. - Células-tronco: toti ou pluripotentes? Veja. Com. November 13th
Zatz, M. - Crime é genético? Veja. Com. November 20th
Zatz, M. - Ainda alzheimer. Veja. Com. November 20th
Zatz, M. - Farmacogenômica: o caminho para medicina personalizada. Veja. Com. November 27th
Zatz, M. - Aspartame de doença de alzheimer . Veja.com. Dezember 04th
Zatz, M. - Células-tronco de gordura e regeneração muscular. Veja.com, Dezember 11st
Zatz, M. - O que é auto-hemoterapia. Veja.com, Dezember 18th
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Annex 4 - Conferences and lectures

- Koifmann, C. - “Diagnóstico e Aconselhamento Genético na Síndrome de Angelman”, Colégio Pentágono –Dezember 06th, 2008.
- Morgante, A. - “Herança genética não-convencional” - 11^a Semana Temática da Biologia, Instituto de Biociências da USP, September, 2008.
- Vainzof M. Células tronco em doenças neuromusculares. Simpósio IV Simpósio do Programa de Pós-graduação em Ciências da Saúde. Criciúma, 25-26 de abril de 2008.
- Vainzof M. Células Tronco em doenças neuromusculares. IX Congresso Médico de Campinas, 14 a 17 de maio de 2008, – Campinas – SP.
- Vainzof M. Modelos Animas para doenças Neuromusculares, Departamento de Biologia Celular e do Desenvolvimento do Instituto de Ciências Biomédicas da Universidade de São Paulo. 22/10/08
- Zatz, M. - Muscular dystrophies and stem-cells- King’s College, London, January 29th,2008.
- Zatz, M. - Clonagem, células-tronco e reprodução assistida :Aspectos éticos- Congresso de fertilização assistida, February 16th,2008
- Zatz, M. - Células-tronco:pesquisas e política. Academia brasileira de Ciências - Rio de Janeiro, March, 2008.
- Zatz, M. - Células-tronco:pesquisas e política.COPEAS, Rio de Janeiro, April, 2008.
- Zatz, M. - Genética e Ética- Curso de Medicina- FMUSP- May, 2008.
- Zatz, M. - Células-tronco embrionárias e adultas: o que podemos aprender com elas? pesquisas e política. Ribeirão Preto, May, 8th, 2008.
- Zatz, M. - Ética e células-tronco: VI encontro de ética, Centro Rebouças, June 2nd,2008.
- Zatz, M. - Células-tronco: religião e ética : Centro de cultura judaica, , S.Paulo, June, 17th, 2008.
- Zatz, M. - O que a genética mudou na nossa vida - Café Filosófico - CPFL Bauru, June, 20th, 2008.
- Zatz, M. - Stem-cells and muscular dystrophies - Newcastle, Reino Unido, July 1st, 2008.
- Zatz, M. - Células-tronco: Política, Ética e Pesquisas- 60^o Reunião da SBPC- Campinas, July, 28th, 2008.
- Zatz, M. - How can bioinformatics help geneticists? Microsoft Research Faculty Summit, Seattle, USA, July 29th,2008.
- Zatz, M. - Integrating exception- Centro de Estudos do Genoma Humano, August, 4th, 2008.
- Zatz, M. - Células-tronco e mídia- Exposição revolução genômica- August, 10th, 2008.

- Zatz, M. - Células-tronco: religião e ética. Espaço K, August, 2008.
- Zatz, M. - Células-tronco em doenças neuromusculares e regeneração óssea : pesquisas e política, Ribeirão Preto, August, 2008.
- Zatz, M. - Genética: passado, presente e futuro. Instituto de Pesquisas Lacanianas, IPLA, August, 2008.
- Zatz, M. - Como a genética beneficiou a vida da mulher? Café Filosófico, Sorocaba, August, 2008.
- Zatz, M. - Como a genética beneficiou a vida da mulher? Café Filosófico, Campinas, September, 2008.
- Zatz, M. - Células-tronco política, ética e pesquisas- Faculdade de Educação, September, 2008.
- Zatz, M. - Células-tronco política, ética e pesquisas- Semana temática, Instituto de Biociências, USP, September, 2008.
- Zatz, M. - Células-tronco: O que poderão representar?Centro de Integração empresa escola. September, 2008.
- Zatz, M. - Gen-ética- CPFL-Café filosófico, Campinas, September, 2008.
- Zatz, M. - SJL dystrophic mice express large amount of human muscle proteins following systemic delivery of human adipose-derived stem cells- XIII Congress of World Muscle Society, Newcastle, September, 2008.
- Zatz, M. - Células-tronco e clonagem- Prêmio Unibanco para escolas públicas, October, 2008.
- Zatz, M. - Universidade de São Paulo: onde estamos e onde queremos chegar? Simpósio USP 20 anos, October, 2008.
- Zatz, M. - Implementation and consolidation of research partnership of public universities- Second international forum of public universities, Beijing, China, November, 2008.
- Zatz, M. - The role of public universities in the development of creativity and training. Second international forum of public universities, Beijing, China, November, 2008.
- Zatz, M. - Neuromuscular disorders: from linkage to preclinical studies. University of Miami, October, 2008.
- Zatz, M. - Células-tronco: o que são e o que podemos esperar delas? Mergulho científico, TV Futura, Rio de Janeiro, Dezember, 5th, 2008.
- Zatz, M. - Perspectivas terapêuticas com células-tronco: Simpósio do Instituto de Ciências biomédicas, Dezember, 11th, 2008.

Annex 5 –Educational and/or Diffusion Activities in Congresses and in The National Science and Technology Week

60th Annual Meeting from Brazilian Society for the Progress of Science, July, 13th to 18th, in Campinas, São Paulo – 1,500 visitors

1^o Meeting of Science Divulcation and Science Teaching (Encontro sobre Divulgação Científica e Ensino de Ciências) – September 10th, Escola de Artes, Ciências e Humanidades (EACH), São Paulo – 100 visitors

Travelling inside the cell - “Science Temple” in Ipameri, Goiás, October, 20th to 23th – 600 visitors

Travelling inside the cell - “Araguaia Shopping Center” in Goiânia, Goiás, October, 23th to 26th – 1,800 visitors

Understanding the cell - “Praça do Relógio” – Universidade de São Paulo – http://www.jornaljovem.com.br/edicao13/semanasp00_00.php - 600 visitors

Symposium and Round Table

Dessen, E. - Os desafios da popularização da ciência – Ibirapuera, São Paulo, April 2008

Koifmann, C. (coordenation) - Conhecendo melhor a síndrome de Angelman passo a passo”. Colégio Pentágono, Dezember 06th, 2008

Meeting Organization

Morgante, A. - Reuniões da Associação X Frágil do Brasil – August 23th to November 23th, 2008.