INTERNATIONAL REGIONAL CONFERENCE ON MEDICAL GENETICS
BELGRADE, MARCH 6-7, 2009

Genetics in Croatia
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Genetics in Croatia

- 1959 - First karyotype
- 1964 - Institute for Human Genetics
- 1969 - Prenatal diagnosis (amniocentesis)
- 1972 – Croatian Society for Human Genetics
- 1978 – Newborn screening for phenylketonuria
- 1992 - Molecular testing for cystic fibrosis
- 2008 – Croatian Society for Rare Diseases
Medical Genetics

- CLINICAL GENETIC SERVICES

- RESEARCH
Clinical Genetic Services and Genetic Counselling Clinics

- Department of Paediatrics, Children’s University Hospital Zagreb
  - Referral Centre for Birth Defects of the Ministry of Health and Social Affairs

- Department of Paediatrics, Clinical Hospital Centre Zagreb
  - Referral Centre for Genetic Metabolic Disorders of the Ministry of Health and Social Affairs

- Department of Paediatrics, Clinical Hospital Centre Split
Types of available genetic tests in Croatia

- **Cytogenetic tests**
- **Molecular karyotyping** (FISH, MLPA)
- **Molecular/biochemical tests**

- **Newborn screening**
- **Diagnostic testing**
  - Individuals with rare inherited disorders
  - Diagnosis of infectious diseases
  - Characterising leukaemias and tumours by analysing acquired genetic changes;

- **Carrier testing**
- **Predictive and presymptomatic testing**
- **Prenatal testing/screening**
- **Preimplantation testing**
- **Forensic testing**
Cytogenetic tests

- Standard karyotyping
- High resolution
- SCE etc
- QF-PCR/FISH for PND
  - blood
  - fibroblasts
  - chorionic villi
  - amniocytes
  - fetal blood
- Clinical Hospital Centre Zagreb
- Children’s University Hospital Zagreb
- General Hospital Holy Spirit, Zagreb
- Clinical Hospital Mercur, Zagreb
- Clinical Hospital Centre Rijeka
- Clinical Hospital Centre Split
FISH and MLPA

- Microdeletion syndromes
- Constitutional aneuploidies and structural chromosomal aberrations
- Cancer genetics
  - Leukemias and Lymphomas
  - Solid Tumors

- Microdeletion syndromes
- Constitutional aneuploidies and structural chromosomal aberrations
- Subtelomere screening by FISH and MLPA

- Clinical Hospital Centre Zagreb
- Children’s University Hospital Zagreb
Molecular tests - Zagreb area
Monogenic disorders

- Achondroplasia/hypochondroplasia (FGFR3)
- Alpha-1-antitrypsin deficiency
- Apolipoprotein E deficiency
- Charcot Marie Tooth (CMT1A)
- Cystic fibrosis
- Congenital adrenal hyperplasia
- Deafness
- Duchenne/Becker muscular dystrophy
- Fragile X syndrome
- Friedreich ataxia
- Gilbert syndrome
- Gonadal dysgenesis (SRY)
- Hemochromatosis
- Huntington chorea
- Hypercholesterolaemia
- MERF
- MELAS
- Myotonic dystrophy
- NARP
- Neurofibromatosis type I
- Polycystic Kidney Disease
- Rett syndrome
- Spinal muscular atrophy
- Spinocerebellar ataxia (type 1, 2, 3, 6)
- Wilson disease
Cancer genetics

Leukemia and Lymphomas & Solid Tumors

- Fusion products
  - t(4;11) - MLL/AF4
  - t(8;21) – AML1/ETO
  - t(9;22) – BCR/ABL
  - t(12;21) – TEL/AML1
  - t(15;17) – PML/RARA
  - inv(16)(p13;q22)-CBFb/MYH11

- Genes
  - JAK2, FLT3, BCL-2 , IgH, TCR, BRAF, Rett, p53, K-RAS, C-kit, HNPPCC (MLH1, MSH2 and MSH6 )
Pharmacogenetics

- CYP2C9
- CYP219
- CYP2D6
- CYP3A4
- DRD2
- NAT2
- MDR1
- MRP2
- UGT1A1
- DPYD, 5-FU
- TS, 5-FU
Genetic risk factors

- ACE (Ins/Del) (hypertension, deep venous trombosis)
- APOE (Alzheimer, coronary heart disease)
- APOB (hypobetalipoproteinaemia)
- CARD15 (NOD2) (Crohn disease)
- DQA1,DQB1,DRB1 (Celiac disease)
- eNOS (coronary spasm, hypertension)
- ESR1 (atherosclerosis, coronary thrombosis and myocardial infraction)
- factor V (deep venous thrombosis, preeclampsia)
- factor II (deep venous thrombosis)
- Interleukin-6 (type 2 diabetes)
- LPL (lipoprotein lipase gene) (Alzheimer)
- MTHFR (deep venous thrombosis)
- PAI-1 (del/bs, 4G/5G) (coronary artery disease)
- PDGFRB (medulloblastoma)
- SERT (anxiety, bipolar disorders)
- 5-HTR2C (behavioral and psychologic symptoms, obesity)
- TPMT (folate and homocysteine levels)
Other

- UDP
- HLA typing tests
- Paternity testing
- Diagnosis of infectious diseases (Chlamydia, CMV, Hepatitis, HIV, HPV, etc)
- Forensics
The cost of genetic testing range from 200 Eur to 1000 Eur, depending on the nature and complexity of the test.

Public health care system covers the costs of genetic testing when it is recommended by a person’s doctor:

- In all children (medical benefits to the child through early diagnosis or preventive measures and therapies must justify genetic testing and screening)

- In adults with additional health insurance (by private or government health insurance provider)
Ethical and Legal issues

- Genetic Counselling (pre- post test)
  - Privacy
  - Informed consent
  - Confidentiality
  - Autonomy

- No specific legal provisions on genetic counselling and testing

- Currently no regulations are in place for evaluating the accuracy and reliability of genetic testing. Most genetic tests developed by laboratories are categorized as services, approved/licensed by Ministry of Health and Social Affairs

- Professional societies are involved in the development of guidelines for genetic tests
- Institute Rudjer Boskovic
- Institute for Anthropology
- Medical School University of Zagreb, Split, and Rijeka
- Dental School University of Zagreb
- MEDILS (Mediterranean Institute for Life Sciences), Split
Upcomming meetings

ISABS Conference on Human Genome Project Based Applications in Forensic Science, Anthropology and Individualized Medicine

JUNE 1-5, 2009

HOTEL MERIDIEN LAV, SPLIT

www.isabs.hr
Upcoming meetings

8th Balkan Meeting on Human Genetics
May 14-17, 2009
Cavtat-Dubrovnik, Croatia

Organisers:
- Croatian Society of Human Genetics
- Croatian Society of Rare Diseases
- Croatian Medical Association

www.studiohrg.hr/human-genetics2009
http://hdhg.me/hr
Thank you for your attention.