Curriculum Vitae

Saeed Reza Ghaffari MSc MD PhD

Cell: 0098-912-1399810
Tel: 0098-021-66935655
E-mail:
saeed@ghaffari.org

2017-5

PERSONAL INFORMATION:

Date and Place of Birth	23 July 1962, Tehran, Iran
Nationality	Iranian
Marital Status	Married

EDUCATIONAL BACKGROUND:

1995-1999	PhD, Medical Genetics Department of Medical Genetics, Faculty of Medicine, University of Glasgow, Glasgow, UK
1994-1995	MSC, Medical Genetics Department of Medical Genetics, Faculty of Medicine, University of Glasgow, Glasgow, UK"
1983-1990	MD, Faculty of Medicine, Tehran University of Medical Sciences, Tehran, Iran

THESIS:

PhD (1995-1999)	“Development and application of comparative genomic hybridization”
MSc (1994-1995)	"Approaches to rapid cytogenetic analysis in newborns”

ACADEMIC and PROFESSIONAL EXPERIENCE

2016-present	Head and Founder Hope Generation Fetal Health Research Institute, Tehran, Iran
2012-Present	Head and Founder Maternal, Fetal and Neonatal Health Clinic, Avicenna Infertility Clinic, Avicenna Biotechnology Research Institute, Tehran, Iran
2012-Present	Member of Board of Directors Avicenna Infertility Clinic, Avicenna Biotechnology Research Institute
2006-present	Head and Founder Center for Control of Birth Defects and Developmental Delay, Hope Generation Foundation, Tehran, Iran Iranian Fetal Medicine Foundation (FMF Iran), Tehran, Iran
2002-Present	Head and Founder Gene Clinic, Genetic Counseling Center and Genetic Laboratory, Tehran, Iran
2002-Present	Scientific Secretariat, Seminars in Clinical Genetics, 103 seminars
2001-2013	Assistant Professor, Department of Medical Genetics, Tehran University of Medical Sciences, Tehran, Iran
2008-2011	Head and Founder Comprehensive Genetic Center, Imam Hospital Complex, Tehran University of Medical Sciences, Tehran, Iran
2007-2011	Head "Iranian Medical Genetics Society"
2006-2008	Head and Founder Department of Reproductive Genetics, Vali-e-Asr Reproductive Health Research Center, Imam Hospital, Tehran University of Medical Sciences, Tehran, Iran
2001-2005	Head and Founder

	DNA Bank, Iranian Cancer Institute, Tehran University of Medical Sciences, Tehran, Iran
2001-2005	Head and Founder Molecular Cytogenetics Laboratory, Imam Hospital Complex, Tehran University of Medical Sciences, Tehran, Iran
2001-2005	Head and Founder Molecular Genetics Laboratory, Imam Hospital Complex, Tehran University of Medical Sciences, Tehran, Iran
2001-2003	Head and Founder Section of Medical Genetics, Shariati Hospital, Tehran University of Medical Sciences, Tehran, Iran
2001-2012	**Designer and manager,** Tehran University of Medical Sciences **Website**
1999-2000	Head Department of Medical Genetics, Zahedan Medical School, Zahedan, Iran
1994-1998	Graduate Student Researcher, Department of Medical Genetics, University of Glasgow, Glasgow, UK
1990-1994	Hospital Based Clinical Practice, National Health Service, Iran

MEMBERSHIP HISTORY:

2002-present, Member:

Scientific board, National committee for prenatal diagnosis, ministry of health

Scientific board, National committee for genetic counseling, ministry of health

Scientific board, Iranian Cancer Institute
Editorial board, Tehran Medical Journal (2000- 2002)

Scientific board, National Genetics committee, Welfare and rehabilitation organization, Iran

Scientific board, Iranian Network for Molecular Medicine
Steering Committee, Iranian Society for Medical Genetics
Scientific Director, Seminars in clinical genetics (so far 96 seminars)

WORKSHOPS

2015, 23-25 Nov
Scientific Secretariat and Instructor

2nd Workshop:"National Program on Education of Genetic Counseling: Practical Course"
Expert Panel and Case Discussion on:

- Drawing A Standard Pedigree
- Risk assessment
- Pedigree analysis
- Psychological Aspects of Genetic Counseling
- Ethical Aspects of Genetic Counseling
- Legal Aspects of Genetic Counseling

Ministry of Health and Medical Education
Tehran, Iran

2015, 19-21 Aug
Scientific Secretariat and Instructor

Pitfalls in Prenatal and Postnatal Diagnosis of Thalassemias
Workshop on "Improving the Technical Skills of Genetic Diagnosis: Lessons from Thalassemia as a Model" (for members of National Genetic Diagnosis Network)

Ministry of Health and Medical Education
Mashhad, Iran

2014, June 23-24
Scientific Secretariat and Instructor

1st Workshop:"National Program on Education of Genetic Counseling: Practical Course",
Expert Panel and Case Discussion on:

- Drawing A Standard Pedigree
- Psychological Aspects of Genetic Counseling
- Ethical Aspects of Genetic Counseling
- Legal Aspects of Genetic Counseling

Ministry of Health and Medical Education
Tehran, Iran

2014, June 20-21

Scientific Secretariat and Instructor

Workshop on "Practical Course on Genetic Counseling"
Expert Panel and Case Discussion on:

- Draw A Standard Pedigree
- Psychological Aspects of Genetic Counseling
- Ethical Aspects of Genetic Counseling
- Legal Aspects of Genetic Counseling

Ministry of Health and Medical Education
Tehran, Iran

2014, June 16-17

Scientific Secretariat and Instructor

Workshop on "Practical Course on Genetic Counseling"
Expert Panel and Case Discussion on:

- Draw A Standard Pedigree
- Psychological Aspects of Genetic Counseling
- Ethical Aspects of Genetic Counseling
- Legal Aspects of Genetic Counseling

Ministry of Health and Medical Education
Tehran, Iran

2011

Scientific Secretariat and Instructor

Workshop on "Sample Treatment, Technical Aspects, Processing and Data Analysis of MLPA"
2nd Medical Genetic Congress, Comprehensive Genetic Center, Imam Khomeini Hospital, Tehran University of Medical Sciences

2011

Scientific Secretariat and Instructor

Workshop on "Sample Treatment, Technical Aspects, Processing and Data Analysis of DNA Sequencing"
2nd Medical Genetic Congress, Comprehensive Genetic Center, Imam Khomeini Hospital, Tehran University of Medical Sciences

2010

Scientific Secretariat and Instructor **Workshop on "Progeny Software: pedigree drawing, design database fields and data management"**
Comprehensive Genetic Center, Imam Khomeini Hospital, Tehran University of Medical Sciences
Iranian Molecular Medicine Network

2008
Scientific Secretariat and Instructor Workshop on "**Sperm Preparation in ART: Fluorescence In Situ Hybridization (FISH) on sperms"**
Vali-E-Asr Reproductive Health Research Center, Tehran University of Medical Sciences

SUPERVISED THESES AND DISSERTATIONS

2014-Present, Supervisor
PhD Student of Medical Genetics
Maternal, Fetal and Neonatal Health Clinic, Avicenna Biotechnology Research Institute
Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences

Determining the Distribution of Genetic Causes among Iranian Patients with Premature Ovarian Insufficiency using a Comprehensive Algorithm with a Specific Approach to Single Gene Disorders by Whole Exome Sequencing

2015-Present, Supervisor
MSc Student of Medical Genetics
Hope generation foundation
Payam-e-Noor University

Application of A Comprehensive Approach to Mutation Analysis of Hearing Loss using Next Generation and Sanger Sequencing Techniques

2015-2016, Supervisor
MSc Student of Medical Genetics
Maternal, Fetal and Neonatal Health Clinic, Avicenna Biotechnology Research Institute
Department of Biology, Karaj Branch, Islamic Azad University, Karaj, Iran

Determining the association of the variants linked to PLK4 gene in mother with fetal aneuploidy, aneuploidy and mother's genes

2014-2016, Supervisor
MSc Student of Developmental Genetics
Department of Biology, Karaj Branch, Islamic Azad University, Karaj, Iran
Gene Clinic

Mutation Analysis of RP2 Gene in X-Linked Retinitis Pigmentosa Patients

2011-2013, Supervisor
MSc Student of Medical Bioinformatics
Joint project:
Hope Generation Foundation
Department of Medical Bioinformatics, Tehran University of Medical Sciences

Design an Algorithm for Automatic Measurement of Ultrasound Markers Used in Prenatal Screening of Down syndrome

2007-2012, Supervisor
PhD Student of Medical Genetics
Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences

Feasibility study of Preimplantation Genetic Diagnosis (PGD) using Whole Genome Amplification Technique and Multiplex PCR

2011-2012, Supervisor
MSc Student of Medical Genetics
International Faculty of Gilan University

Genetic Investigation of Products of Conception using MLPA Technique

2006-2012, Supervisor
PhD Student of Medical Genetics
Joint project:
Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences
State Welfare Organization of Iran

Design A Comprehensive Stepwise Diagnostic Approach to Mental Retardation by Determining the Molecular Profile of DNA Copy Number Abnormalities

2009-2011, Supervisor
MSc Student of Medical Genetics
Science and Research Branch, Islamic Azad University

Assessment of Common Microdeletion/Microduplication Syndromes in Iranian Patients with Familial Intellectual Disability using MLPA Technique

2009-2011, Supervisor
MSc Student of Medical Genetics
Joint project:
Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences
Department of haematology, oncology, Society of Iranian Hemophilia Patient, School of Medicine, Tehran University of Medical Sciences
Science and Research Branch, Islamic Azad University

Deletion/duplication Analysis of F8 Gene in Patients with Severe Haemophilia a and Inhibitor Development Using MLPA Technique

2005-2009, Supervisor
PhD Student of Medical Genetics
Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences
Heterozygosity Assessment of 15 STRs Located on Chromosomes 21, 18, 13, X and Y in Fetal Samples Used in Prenatal Diagnosis

2007-2009, Supervisor
MSc Student of Medical Genetics
Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences
Feasibility Study of Rapid Aneuploidy Detection in Uncultured Amniotic Fluid by MLPA Technique

2007-2009, Supervisor
MSc Student of Medical Genetics
Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences
Correlation of Embryo Morphology with Aneuploidy: Preimplantation Genetic Screening of Aneuploidies Using FISH Technique

2007-2008, Supervisor
Medical student
Joint project:
Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences
Department of Paediatrics and Neonatology, School of Medicine, Tehran University of Medical Sciences
Prevalence of 22q11 Microdeletion in Children Affected with Congenital Heart Defects, Referred to Hospitals Affiliated with Tehran University of Medical Sciences

2006-2007, Supervisor
Medical Student
Joint project:
Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences
Department of Urology, School of Medicine, Tehran University of Medical Sciences
Set-Up and Optimization of Chromosome Y Microdeletion Study in Men eith Idiopathic Non-Obstructive Oligospermia using 6 Primer Sets Recommended by European Academy of Andrology

2005-2007, Supervisor
Joint project:
Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences
School of Dentistry, Tehran University of Medical Sciences

Assessment of viral load of Human Papillomavirus in Patients saliva And Fresh Tumoral Tissues of Squamous Cell Carcinoma of the Mouth using PCR

2004-2006, Supervisor
MSc Student of Medical Genetics

Assessment of Chromosomal Aneuploidies in Oesophageal Squamous Cell Carcinoma

2006, Supervisor
Medical student
School of Medicine, Tehran University of Medical Sciences

Prevalence of Congenital Heart Defect and Other Congenital Anomalies in First and Second Degree Relatives of 110 Patients with Conotruncal Abnormalties

2004-2006, , Supervisor
Urology Resident
Joint project:
Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences
Department of Urology, School of Medicine, Tehran University of Medical Sciences

Non-invasive Diagnosis of Bladder Cancer by Investigation of Chromosome Abnormalities in Urine Sediment Cells Using FISH Technique

2003-2005, , Supervisor
Pathology Resident
Joint project:
Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences
Department of Pathology, School of Medicine, Tehran University of Medical Sciences

Analysis of Chromosome Abnormalities in Invasive Ductal Carcinoma of Breast with and without Metastasis to Lymph Nodes Using Comparative Genomic Hybridization (CGH)

2001-2003, , Supervisor
MSc Student of electronic engineering
Joint project:
Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences
School of Electrical and Computer Engineering, College of Engineering, University of Tehran

Automatic Landmark Detection and Locating Specific Regions of Human Chromosomes using Computational Intelligence

CONTRIBUTION to NATIONAL PROGRAMS

Collaborative Projects with "Ministry of Health and Medical Education"

2011-present

Program on "Development of National Genetic Counseling Network: Education of General Practitioners in Primary Healthcare Network"

Types of Partnership:
(Design the program, management, teaching, etc.)

A) Providing Educational Contents **Making Educational Videos**

- Basic Principles of Genetic Counseling
- Pedigree Analysis and Risk Assessment
- Genetic Counseling in Chromosomal Abnormalities
- Genetic Counseling and Risk Assessment in CFTR Related Disorders
- Genetic Counseling and Risk Assessment in Alpha Thalassemia
- Genetic Counseling and Risk Assessment in Autosomal Recessive Polycystic Kidney Disease
- Genetic Counseling and Risk Assessment in Phenylketonuria
- Genetic Counseling and Risk Assessment in Common Multifactorial Disorders

Publishing 20 books on the subject of genetic counseling
(Listed in "BOOKS" section)

B) Development of "Comprehensive Genetic Counseling" Software

- Pedigree Structure, Field Design, …
- Running the Software in the Pilot Phase

C) Workshops (Three Practical Courses)

Expert Panel and Case Discussion on:

- Draw A Standard Pedigree
- Psychological Aspects of Genetic Counseling
- Ethical Aspects of Genetic Counseling
- Legal Aspects of Genetic Counseling

(More details in "WORKSHOPS" Section)

D) Evaluations and Exams **Design Questions for :**

Multiple-Choice Online Exams

First Training Course: Three exams, **2012-2014**
Second Training Course: Three exams, **2014-2015**

Practical Online Exams (case discussion)

First Training Course: **2014-2015**

48 Online Exams focusing on Cases with:

- Alpha thalassemia (3 families)
- Beta thalassemia (3 families)
- hemophilia
- Spinal muscular atrophy (5 families)
- Duchenne muscular dystrophy (4 families)
- Intellectual disability (4 families)
- Hearing loss (5 families)
- Visual Loss
- Autosomal Recessive Polycystic Kidney
- Autosomal Dominant Polycystic Kidney
- Inherited Metabolic Disorders
- Hereditary Breast and Ovarian Cancer
- Hereditary Colorectal Cancer

Second Training Course: **2015-2016**

48 Online Exams focusing on Cases with:

- Alpha thalassemia (3 families)
- Beta thalassemia (3 families)
- hemophilia
- Spinal muscular atrophy (5 families)

- Duchenne muscular dystrophy (4 families)
- Intellectual disability (4 families)
- Hearing loss (5 families)
- Visual Loss
- Autosomal Recessive Polycystic Kidney
- Autosomal Dominant Polycystic Kidney
- Inherited Metabolic Disorders
- Hereditary Breast and Ovarian Cancer
- Hereditary Colorectal Cancer

E) Practical Courses **Education of Genetic Counseling in a Clinical Setting and Evaluation of Logbooks**

First Training Course: **2015**

2015, February 21-23	1st Group, 3 Students
2015, February 24-26	2nd Group, 3 Students
2015, February 28-March 2	3rd Group, 4 Students
2015, March 3-5	4th Group, 2 Students
2015, March 7-9	5th Group, 3 Students
2015, March 10-12	6th Group, 2 Students
2015, March 14-16	7th Group, 2 Students
2015, April 13-15	8th Group, 2 Students
2015, June 27-29	9th Group, 1 Student
2015, July 4-6	10th Group, 1 Student
2015, July 11-13	11th Group, 1 Student
2015, July 20-22	12th Group, 1 Student
2015, July 25-27	13th Group, 3 Students
2015, July 28-30	14th Group, 2 Students
2015, August 1-3	15th Group, 3 Students
2015, August 4-6	16th Group, 3 Students
2015, August 8-10	17th Group, 3 Students
2015, August 15-17	18th Group, 3 Students
2015, August 18-20	19th Group, 3 Students
2015, August 22-24	20th Group, 3 Students
2015, August 25-27	21th Group, 3 Students
2015, August 29-31	22th Group, 4 Students
2015, September 5-7	23th Group, 4 Students
2015, September 8-10	24th Group, 4 Students
2015, September 12-14	25th Group, 3 Students
2015, September 15-17	26th Group, 3 Students
2015, September 19-21	27th Group, 3 Students
2015, September 26-28	28th Group, 5 Students

Second Training Course: **2016**

2016, July 23-28	1st Group, 5 Students
2016, July 31-August 4	2nd Group, 3 Students
2016, August 6-11	3rd Group, 4 Students
2016August 13-18	4th Group, 6 Students
2016August 20-25	5th Group, 8 Students
2016August 27-September 1	6th Group, 7 Students
2016 September 3-8	7th Group, 6 Students
2016 September 24-29	8th Group, 7 Students
2016 October 1-6	9th Group, 5 Students
2016 October 15-20	10th Group, 8 Students
2016 October 22-27	11th Group, 1 Student
2016 October 29- November 3	12th Group, 3 Students

2013-Present	
Principal Investigator	**National Program on "Evaluation of Commercial Risk Assessment Software used in Prenatal Screening of Chromosomal Abnormalities", Reference Lab, Ministry of Health and Medical Education** **Hope Generation Foundation**

TEACHING EXPERIENCE

2016

PhD Students of Reproductive Biology

Reproductive Genetics:
Preimplantation Genetic Diagnosis (PGD) of single gene disorders using single cell whole genome amplification
Prenatal Dignosis (PND)
Molecular Cytogenetics: QF-PCR, MLPA, Array CGH
Chromosme abnormalities
Department of Anatomy, School of Medicine, Tehran University of Medical Sciences

2012-Present

PhD students of Reproductive Health (5 Semesters)

Genetics in Infertility: Preimplantation Genetic Screening and Diagnosis
School of Medicine, Tehran University of Medical Sciences
Avicenna Research Institute

2001-2012
PhD Students of Medical Genetics, (11 academic years, 21 Semesters)

Medical Genetics 1
Department of Medical Genetics,
School of Medicine, Tehran University of Medical Sciences

2005-2012
PhD Students of Medical Genetics, (7 academic years, 14 Semesters)

Medical Genetics 2 (Internship)
Department of Medical Genetics,
School of Medicine, Tehran University of Medical Sciences

2001-2012
PhD Students of Medical Genetics, (11 academic years, 21 Semesters)

Molecular Cytogenetics,
Department of Medical Genetics,
School of Medicine, Tehran University of Medical Sciences

2001-2012
PhD Students of Medical Genetics, (11 academic years, 21 Semesters)

Advanced Molecular Genetics,
Department of Medical Genetics,
School of Medicine, Tehran University of Medical Sciences

2001-2012
PhD Students of Medical Genetics,

Advanced Cancer Genetics
Department of Medical Genetics,

(11 academic years, 21 Semesters)	School of Medicine, Tehran University of Medical Sciences
2001-2012 **PhD Students of Medical Genetics,** **(11 academic years, 21 Semesters)**	**Advanced Cytogenetics,** Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences
2010-2012 **PhD Students of Medical Genetics,** **(2 academic years, 4 Semesters)**	**Behavioural Genetics** Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences
2001-2007 **PhD Students of Medical Genetics,** **(8 Semesters)**	**Population Genetics** Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences
2009-2011 **Fellowship in Maternal and Fetal Medicine** **(3 Semesters)**	**Medical Genetics Rotation** Department of Obstetrics and Gynecology, School of Medicine, Tehran University of Medical Sciences
2011 **Neonatalogy fellowship**	**Medical Genetics Rotation** Department of Paediatrics And Neonatalogy, School of Medicine, Tehran University of Medical Sciences
2005-Present **General Practitioners (MD)** **(15 Courses)**	**Genetic Counseling in Intellectual Disability** State Welfare Organization of Iran
2005-Present **Medical Students** **(11 academic years, 22 semesters)**	**Genetic Counseling in Intellectual Disability** School of Medicine, Tehran University of Medical Sciences
2001-2012 **MSc Students of Medical Genetics,** **(11 academic years, 21 Semesters)**	**Medical Genetics 1** Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences
2001-2012 **MSc Students of Medical Genetics,**	**Cytogenetics 1** Department of Medical Genetics,

(11 academic years, 21 Semesters)	School of Medicine, Tehran University of Medical Sciences

2001-2012	**Cancer Genetics**
MSc Students of Medical Genetics,	Department of Medical Genetics,
(11 academic years, 21 Semesters)	School of Medicine, Tehran University of Medical Sciences

RESEARCH ACHIEVEMENTS

2016

The First Iranian Child Born after Preimplantation Genetic Diagnosis (PGD) of Cystic Fibrosis

- Avicenna Infertility Clinic, Avicenna Research Institute

2015
Principal Scientist

Optimization, and Set up of A "Next Generation Sequencing" Lab for the First Time in Iran

- Hope Generation Foundation

2013-2014
Principal Scientist

Optimization, and Set up of A "Preimplantation Genetic Diagnosis (PGD) Lab" for PGD of a variety of Single Gene Disorders

- Avicenna Infertility Clinic, Avicenna Research Institute

2013
Principal Scientist

Optimization and Clinical Application of Single Cell Whole Genome Amplification Technique

- Avicenna Infertility Clinic, Avicenna Research Institute

2010-2014
Principal Scientist

Optimization, and Set up of A "Prenatal Diagnosis (PND) Lab" for PND of a variety of Genetic Disorders

- Hope Generation Foundation
- Comprehensive Genetic Center, Imam Hospital Complex, Tehran University of Medical Sciences
- "Gene" clinic
- Avicenna Infertility Clinic, Avicenna Research Institute

2011-2014
Principal Scientist

Optimization and Clinical Application of "Rapid Prenatal Aneuploidy Detection "Techniques including MLPA and QF-PCR

- Hope Generation Foundation
- Comprehensive Genetic Center, Imam Hospital Complex, Tehran University of Medical Sciences
- "Gene" clinic
- Avicenna Infertility Clinic, Avicenna Research Institute

2010-2012
Principal Scientist

Optimization and Clinical Application of Genetic Investigation of Products of Conception using Molecular Cytogenetic Techniques

- Hope Generation Foundation
- Comprehensive Genetic Center, Imam Hospital Complex, Tehran University of Medical Sciences
- "Gene" clinic
- Avicenna Infertility Clinic, Avicenna Research Institute

2009-2012
Principal Scientist

Optimization and Clinical Application of Multiplex Ligation-Dependent Probe Amplification (MLPA) Technique

- Hope Generation Foundation
- Comprehensive Genetic Center, Imam Hospital Complex, Tehran University of Medical Sciences
- "Gene" clinic
- Avicenna Infertility Clinic, Avicenna Research Institute

2009-2011
Principal Scientist **Design and Establishment of "Comprehensive Genetic Center" and "clinical genetics laboratories"**

- Imam Hospital Complex, Tehran University of Medical Sciences

Before 2009:

- Development of rapid approaches to cytogenetics analysis in newborns, using direct lymphocyte preparations, overnight un-stimulated lymphocyte cultures and rapid multi-color FISH

  (MSc project).

- Optimization and modification of CGH for a clinical cytogenetics laboratory
  - Glasgow, UK
  - "GENE" Institute, Iran.
  - Tehran University hospitals, Iran
- Application of CGH for detection of complex chromosomal abnormalities in patients with hematological malignancies, solid tumours, double minutes, low percentage mosaisicm marker chromosomes, and common chromosomal abnormalities,
  - Glasgow, UK
  - "GENE" Institute, Iran.
- Development of a new strategy for CGH image analysis in patients with hyperploidies.
  - Glasgow, UK
  - "GENE" Institute, Iran.
- Development of a new strategy for cryptic chromosome translocation screening in patients with idiopathic mental retardation, using CGH.
  - Glasgow, UK
  - "GENE" Institute, Iran.
- Improvement of the CGH technique resolution.
  - Glasgow, UK

- Development of overnight CGH.
    - Glasgow, UK
- Development of CGH to DNA targets (single DNA fibers, Arrays of DNA fibers and DNA microarrays) (feasibility study)
    - Glasgow, UK
    - "GENE" Institute, Iran.
- Optimization and application of single cell CGH
    - Glasgow, UK
    -
    - "GENE" Institute, Iran.
- Application of PCR and FISH for the preimplantation diagnosis of molecular and numerical chromosomal abnormalities
    - "GENE" Institute Iran. In collaboration with Institute of assisted reproduction, Tehran, Iran
- Optimization of techniques for First trimester screening of chromosomal abnormalities using serum markers in pregnant women
    - Imam Hospital Complex, Biochemical Genetics Laboratory
- Optimization and application of multiplex PCR for the molecular diagnosis of of Alpha Thalassemia.
    - Imam Hospital Complex, Biochemical Genetics Laboratory
- Optimization and set up of a FISH lab for detection of numerical, structural abnormalities and cryptic chromosome rearrangements (including all known microdeletion syndromes and 42 telomeric probes).
    - "GENE" laboratories, Molecular cytogenetics, Iran (online details at: http://medical-genetics.com)
- Design and establishment of "Iranian Genetic Core Facility" (Part of CPCA)

Design and establishment of "Centre for prevention of congenital abnormalities(CPCA)", approved by ministry of health

RESEARCH EXPERIENCE:

Principal Investigator:

2015-2016	**Feasibility Study of Noninvasive Prenatal Testing of Aneuploidies on Cell-Free Fetal DNA using SNP-Based Methods**
2015-2016	**Development and Optimization of PGS-NGS: Preimplantation Genetic Screening of All Chromosomal Aneuploidies using Next Generation Sequencing (Single Cell Whole Genome Amplification Followed by Low-Pass NGS Aneuploidy Detection)** Hope Generation Foundation Avicenna Research Institute
2015-2016	**Development and Optimization of HLA Typing using Next Generation Sequencing** Hope Generation Foundation Iranian Blood Transfusion Organization
2015	**Design the Specific Gene Sets for Targeted Next Generation Sequencing in Heterogenous Disorders (Intellectual Disability, Hearing Impairment, Retinitis Pigmentosa, Neuromuscular Disorders, Genodermatoses, …)** Hope Generation Foundation
2015	**Optimization and Clinical Application of Next Generation Sequencing Technique** Hope Generation Foundation
2013-2015	**Construction of Repeat-Free Fluorescence In Situ Hybridization Probes (High-Definition DNA FISH)** Avicenna Infertility Clinic, Avicenna Research Institute
2012-2015	**Optimization and Clinical Application of Preimplantation Genetic Diagnosis (PGD) in a Wide Range of Single Gene Disorders**

Avicenna Infertility Clinic, Avicenna Research Institute

2012-2013 **Optimization of Single Cell Whole Genome Amplification Technique**
Avicenna Infertility Clinic, Avicenna Research Institute

2012-2015 **Mutation Analysis of RP2 Gene in X-linked Retinitis Pigmentosa patients**
Gene Clinic
Iranian RP Society

2012-2014 **Mutation Analysis of RHO Gene in Autosomal Dominant Retinitis Pigmentosa patients**
Hope Generation Foundation
Iranian RP Society

2010-2013 **A Comprehensive Genomic Approach to Autosomal Recessive Retinitis Pigmentosa using Next Generation Sequencing Technique**
Comprehensive Genetic Center, Faculty of Medicine, Tehran University of Medical Sciences
Iranian RP Society

2011-present **Investigation of the Pregnancy Outcome of More than 22000 Women Referred for First/SecondTrimester Screening and/or ultrasound study**
Hope Generation Foundation

2012-present **Investigation of the Clinical Significance of Prenatally Detected Ultrasound Abnormalities by Active Follow up of the Pregnancies with Affected Fetuses**
Hope Generation Foundation

2010- Present **Design and Implementation of Integrated Data Mining and Analysis of More than 101000 Pregnant**

Women Referred for First/SecondTrimester Screening and/or ultrasound study
Hope Generation Foundation

2010-2013 **Investigation of Aneuploidies and Cryptic Chromosomal Abnormalities in Products of Conception**
Hope Generation Foundation

2010-2011 **Determining the Deletions and Duplications of F8 Gene in Severe Haemophilia A Patients with Inhibitor Development**
Comprehensive Genetic Center, Faculty of Medicine, Tehran University of Medical Sciences

2010-2011 **Rapid Prenatal Diagnosis of Chromosomal Aneuploidies in Amniotic Fluid or Chorionic Villus Samples using MLPA Technique**
Comprehensive Genetic Center, Faculty of Medicine, Tehran University of Medical Sciences

2009-2011 **Determining the Distribution of Different Modes of Inheritance among Iranian Patients with Retinitis Pigmentosa by Pedigree Analysis of 3950 Iranian RP Families registered in Iranian RP Society**,
Comprehensive Genetic Center, Faculty of Medicine, Tehran University of Medical Sciences
Iranian RP Society

2009-2010 **Investigation of Deletions and Duplications of F8 Gene in Iranian Severe Haemophilia A Patients Using MLPA Technique**, Comprehensive Genetic Center, Faculty of Medicine, Tehran University of Medical Sciences

2008-2011 **Design A Comprehensive Stepwise Diagnostic Approach to Mental Retardation by Determining the Molecular Profile of DNA Copy Number Abnormalities,** Comprehensive Genetic Center, Faculty of Medicine, Tehran University of Medical Sciences

2010-2012	**Preimplantation Genetic Diagnosis of Spinal Muscular Atrophy**, Comprehensive Genetic Center, Faculty of Medicine, Tehran University of Medical Sciences
2010-2011	**Investigation of Chromosomal Aneuploidies in Oral SCC Tumors**, Comprehensive Genetic Center, Faculty of Medicine, Tehran University of Medical Sciences
2009-2011	**Assessment of Her-2 Amplification Status in Iranian Breast Cancer Patients**, Comparison of IHC and FISH, Gene Clinic

Before 2009:

- Preimlantation genetic diagnosis using FISH technique, a Collaborative study, *Reproductive health research centre, Reproductive Health Research Center, Assisted Reproductive Research Centre, GENE institute*
- Application of all (42) telomeric probes in the patients with idiopathic mental retardation., *GENE institute, Children's Central Hospital, Iranian Molecular Medicine Network, Cancer Research center*
- Application of all (42) telomeric probes in the patients with recurrent abortions., *GENE institute, Reproductive Health Research Center, Iranian Molecular medicine network, Assisted reproductive research centre, Cancer Research center*
- Application of FISH in the detection of microdeletion syndromes. *GENE institute, Iranian Molecular medicine network, Cancer Research center*
- Application of FISH for the detection of chromosomal deletions in patients with congenital heart disease, *GENE institute, Children's Central Hospital, Iranian Molecular Medicine, Department of Medical Genetics, Tehran Medical School, Cancer Research center*
- Radiation Induced Chromosomal Breakage Assay of Peripheral Lymphocytes in Patients with Familial and Sporadic Breast Cancer, a Comparative Study, *Cancer Research centre, Asthma and Immunology research centre*
- Chromosomal breakage study in the patients with Ataxia Telangectasia, a Comparative Study, *Cancer Research Center, Asthma and Immunology Research Center*

- Early-detection of micrometastases in lymph nodes from patients with breast cancer by RT-PCR screening of axillary nodes, compared to routine histology. , *Cancer Research Center*

- Chromosomal aberrations detected by Comparative genomic hybridization (CGH) in invasive ductal carcinoma and peritumoral ductal carcinoma insitu (DCIS) and usual ductal hyperplasia. *Cancer Research Center*

- Comparison of chromosomal aberration in two groups of invasive ductal carcinoma of breast with and without axillary lymph nodes metastasis by comparative genomic hybridization (CGH). *Cancer Research Center*

- Molecular and molecular cytogenetic diagnosis of Hematological Malignancies, *Gene Institute*

- Detection of HPV infection using molecular techniques in 130 families, *Cancer research center, Reproductive Health Research Center*

- First Trimester Screening of fetal chromosomal abnormalities using maternal age, ultrasound and biochemical markers, a comprehensive study, *Iranian Genetic Core Facility, FMF Iran*

- Determination of RBC Indices in the parents of Major Thalassemia Patients and Mutation Detection in Those Who Are Not Detectable by National Carrier Screening programme, *Cancer Institute, Central Children Hospital, Pasteur Institute, Genetic Office*

- Determination of RBC Indices and Mutation Detection in Infants with the Haemoglobin Bart's Hydrops Fetalis Syndrom and their parents , *Reproductive health research centre, Cancer research Center*

- Comparative Genomic Hybridization Analysis of Esophageal tumors, *Tarbiat Modares University, Cancer research Center, Iranian Genetic core Facility*

CURRENT RESEARCH PROJECTS:

2016 **Copy Number Variation Analysis on Data of Exome Sequencing**
Hope Generation Foundation

2016 **Preimplantation Genetic Diagnosis of embryo HLA using Next Generation Sequencing (PGD-HLA-NGS)**
Hope Generation Foundation
Avicenna Research Institute

2016 **Validation Study of Clinical Application of Noninvasive Prenatal Testing of Aneuploidies on Cell-Free Fetal DNA Using SNP-Based Methods**
Hope Generation Foundation

2015 **Clinical Application of Next Generation Sequencing in Personalized Medicine: Multi-Gene Panel Cancer Genetics (Germline and Somatic Mutation Detection)**
Hope Generation Foundation

2015 **Clinical Application of Next Generation Sequencing in Personalized Medicine: Pre-symptomatic Diagnosis of Preventable Lethal Disorders**
Hope Generation Foundation

2015 **Determining the Distribution of Genetic Causes among Iranian Patients with Premature Ovarian Insufficiency using a Comprehensive Algorithm with a Specific Approach to Single Gene Disorders by Whole Exome Sequencing**
Hope Generation Foundation
Avicenna Research Institute

2015 **Determining the Genetic Causes of Familial Cerebral Palsy using Whole Exome Sequencing**
Hope Generation Foundation
Vali E-Asr Rehabilitation Foundation

2015 **Assessment of Single Gene Disorders in Familial Intellectual Disability by Whole Exome Sequencing**
Hope Generation Foundation

2015 **Clinical Application of Next Generation Sequencing in Personalized Medicine: targeted Cancer Treatment**
Hope Generation Foundation

2015 **Clinical Application of Next Generation Sequencing in Personalized Medicine: Presymptomatic Diagnosis of Preventable Lethal Disorders**

Hope Generation Foundation

PAPERS

Published Papers:

1- Primary ovarian insufficiency with t(5;13): a case report and literature review on disrupted genes.
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38- Hematological Indices of Parents in Non-Immune Hydrops Fetalis Pregnancie.**Saeed Reza Ghaffari**, M.D., Ph.D.;1,2,3,4 , Farzaneh Larti, M.D.; Tayebeh Sabokbar, M.S.c.; Maryam Rafati, M.D.; Jila Dastan M.S.c, M.D.; Laleh Eslamian, M.D.; Fatemeh Rahimi, M.D.; SedigheBorna, M.D. Journal of Family and Reproductive Health2008;2(1) : 33-36

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BOOKS

- **"Lecture Notes in Genetic Counseling" series**
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    Published Online: Official Website of "Ministry of Health and Medical Education", 2014

19. **Lecture Notes in Genetic Counseling, Hereditary Colorectal Cancer, Co-author**
    Published Online: Official Website of "Ministry of Health and Medical Education", 2014

20. **Genetics: Law, Ethics and Psychology, chapter 4: Human Enhancement Technology, Co-author**
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21. **Lecture Notes in Genetics for Health Care Providers, Author**
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REGISTRATION of NEW MUTATIONS IN GenBank

Link to released novel mutations: https://www.ncbi.nlm.nih.gov/nuccore/?term=ghaffari+sr

No	GenBank Accession Number	Gene	Mutation	Title	Date of Accession Number Assignment	Released
1	KU951140	PCCB	1501GT	A novel mutation detected by whole exome sequencing in an Iranian patient with intellectual disability	22 March 2016	*
2	KU951141	PYCR1	H243R	Whole exome sequencing detected compound heterozygous mutations in an Iranian patient with syndromic intellectual disability, making the diagnosis of cutis laxa type IIB	22 March 2016	*
3	KU951142	ST3GAL3	R196G	Whole exome sequencing revealed a novel mutation in an Iranian patient with non-syndromic intellectual disability	22 March 2016	*
4	KU951143	NPHS1	4bp duplication	A novel truncating mutation in NPHS1 detected in an Iranian patient with congenital nephritic syndrome	22 March 2016	*
5	KU726869	CYP4F22	1346CT	Targeted next generation sequencing detected a novel mutation in an Iranian patient with congenital ichthyosis	18 Feb 2016	*
6	KU695564	NPC1	1415TC	Whole exome sequencing detected a novel mutation in an Iranian patient with inborn errors of metabolism		*
7	KP742956	EDARADD	G73R	Targeted Next Generation Sequencing Revealed a Novel Mutation in EDARADD gene in an Iranian Patient with Hypohidrotic Ectodermal Dysplasia	5 Feb 2015	*
8	KP734176.1	RHO	L95P	Identification of a Novel Mutation in RHO Gene in an Iranian Patient with Autosomal Dominant Retinitis Pigmentosa	4 Feb 2015	*
9	KP750178.1	MUT	360dupT	A Novel Truncating Mutation in MUT gene in an Iranian patient with Methyl Malonic Acidemia which is Identified by Next Generation Sequencing	7 Feb 2015	*
10	KT001453	AGL	2681+4_+5insA	Identification of a Novel Mutation in AGL Gene in an Iranian Family with Glycogen Storage Disease Type III using	3 June 2015	*

				Next-Generation Sequencing"		
11	KT001454	NPC1	2683GT	Next-Generation Sequencing Revealed a Novel Mutation in NPC1 Gene in an Iranian Family with Niemann-Pick Disease	3 June 2015	*
12	KR052806.1	NR2E3	1118TC	Next Generation Sequencing Revealed a Novel Mutation in NR2E3 in an Iranian Patient with Retinitis Pigmentosa	2 April 2015	*
13	KR052805.1	TOPORS	2047CT	Next Generation Sequencing Revealed a Novel Mutation in TOPORS Gene (E3 ubiquitin-protein ligase Topors isoform 1) in an Iranian Patient with Retinitis Pigmentosa	2 April 2015	*
14	KP869066	FANCG	260delG	Identification of a Truncating Mutation in FANCG Gene in an Iranian Patient with Fanconi Anemia using Targeted Next Generation Sequencing Technique	3 March 2015	*
15	KP869067	BCKDHB	988GA	Targeted Next Generation Sequencing Revealed a Novel Mutation in BCKDHB gene in an Iranian Patient with Maple Syrup Urine Disease	3 March 2015	*
16	KP869068	KCNT1	1421GT	Whole Exome Sequencing Revealed a Novel Heterozygous Mutation in KCNT1 Gene in an Iranian Patient with Intellectual Disability	3 March 2015	*
17	KP836347	SLC26A4	164delG	Next Generation Sequencing Revealed a Novel Mutation in SLC26A4 Gene in an Iranian Patient with Autosomal Recessive Hearing Impairment	24 Feb 2015	*
18	KP836348	TULP1	1111AC	Next Generation Sequencing Revealed a Novel Mutation in TULP1 Gene in an Iranian Patient with Autosomal Recessive Retinitis Pigmentosa	24 Feb 2015	*
19	KP836349	USH2A	13234CA	Next Generation Sequencing Revealed a Novel Variant in USH2A Gene in an Iranian Family with Inherited Retinal Disorder	24 Feb 2015	*
20	KP718608	AGL	1592CG	Identification of a novel mutation in AGL gene in anIranian patient with glycogen storage disease using Targeted **Next Generation Sequencing Technique**	30 Jan 2015	*
21	KP718609	PYGM	2398CT	Identification of a Novel Mutation in PYGM Gene in an Iranian Patient with Glycogen Storage Disease using Targeted **Next Generation**	30 Jan 2015	*

				Sequencing Technique		
22	KP718610	RHO	930CG	Identification of a Novel Mutation in RHO Gene in an Iranian Patient with Autosomal Dominant Retinitis Pigmentosa	30 Jan 2015	*
23	KP718611	ALDOA	289GC	Identification of a Novel Mutation in ALDOA Gene in an Iranian Patient with Glycogen Storage Disease using Targeted **Next Generation Sequencing Technique**	30 Jan 2015	*
24	KM873048.1	ALSM1	5846delTC	A Novel Mutation in ALSM1 Gene, Detected in a Patient with Familial Autosomal Recessive Cone Rod Dystrophy using **whole Exome Sequencing**	7 Oct 2014	*
25	KM873049.1	HSD3B2	690GA	A Novel Mutation in HSD3B2 Gene, Detected in a Patient with Congenital Adrenal Hyperplasia	7 Oct 2014	*
26	KM873050.1	AVCR1	737TA	A Novel Mutation in ACVR1 Gene, Detected in a Patient with Fibrodysplasia Ossificans Progressive using **whole Exome Sequencing**	7 Oct 2014	*
27	KJ849294.1	RHO	R177K	A Novel Mutation in RHO Gene Detected In an Iranian Family with Autosomal Dominant Retinitis Pigmentosa	25 May 2014	*
28	EU709494.1	CDH1	G759X	A novel mutation in the E-cadherin gene in the first family with hereditary diffuse gastric cancer reported in Iran	2008	*

HOLDING SEMINARS AND CONGRESS

❖ Head

2nd Iranian Medical Genetics Congress, Tehran, Iran, 2011

❖ Scientific Secretariat

100 Clinical Genetics Seminars since 2003
Details of the selected seminars in recent 5-year period are provided below:

1- **"Symposium of Genetic Counseling, Comprehensive Community Genetics Program"**
100th Seminar in Clinical Genetics, Held by Ministry of Health and Medical Education, 2-3 March 2017

2- **Clinical Application of Next Generation Sequencing, approaching to Inborn Errors of Metabolism: Report on a 2-year Iran Experience and Case Discussion**
99th Seminar in Clinical Genetics, "Report on a 2-year Experience of Next Generation Sequencing in the "First Iranian NGS Core Facility": Discussion on Positive Findings in Patients with Inborn Errors of Metabolism, Tehran, Iran, 26 January 2017

3- **Clinical Application of Next Generation Sequencing, approaching to Intellectual Disability and Developmental Delay: Report on a 2-year Iran Experience and Case Discussion**
98th Seminar in Clinical Genetics, "Report on a 2-year Experience of Next Generation Sequencing in the "First Iranian NGS Core Facility": Discussion on Positive Findings in Patients with Intellectual Disability/Developmental Delay, Tehran, Iran, 5 January 2017

4- **Preimplantation Genetic Screening and Diagnosis using Next Generation Sequencing (PGS-NGS): Iran Experience**
97th Seminar in Clinical Genetics, Tehran, Iran, November 2016

5- **Prenatal diagnosis, Rights of the Fetus and the Family, Rights and Responsibilities of the Physician and the Lab: Technical Aspects and Case Presentation**
96th Seminar in Clinical Genetics, "Prenatal diagnosis: Rights of the Fetus and the Family, Rights and Responsibilities of the Physician and the Lab", Tehran, Iran, 20 August 2016

6- **"Personalized Medicine in Diagnosis and Treatment of Cancer: Application of Next Generation Sequencing"**
95th Seminar in Clinical Genetics, Tehran, Iran, 21 July, 2016

7- **A) Algorithmic Approach in Genetic Counseling**
B) Pre-marriage Genetic Counseling
C) Genetic Counseling in Hereditary Hearing Loss
D) Genetic Counseling in Hereditary Visual Loss
E) Prenatal Screening of Chromosomal Abnormalities
"Symposium of Genetic Counseling, Comprehensive Community Genetics Program"
94th Seminar in Clinical Genetics, jointly Held by Ministry of Health and Medical Education, 2-3 March 2016

8- **Session1: "NGS technique: from library preparation to high throughput sequencing"**
Session 2: "NGS: Basic and advanced data analysis"
Session 3: "Standard clinical report of exome/targeted sequencing based on ACMG guideline"
Session 4: "Experts Panel"
93th Seminar in Clinical Genetics, "Report on the 1-year Experience of Next Generation Sequencing in the "First Iranian NGS Core Facility": Technical, Clinical and Research Aspects", Tehran, Iran, 7 January 2016

9- **Prenatal diagnosis, Rights of the Fetus and the Family, Rights and Responsibilities of the Physician and the Lab: Technical Aspects and Case Presentation**

92th Seminar in Clinical Genetics, "Prenatal diagnosis: Rights of the Fetus and the Family, Rights and Responsibilities of the Physician and the Lab", Tehran, Iran, 17 December 2015

10- **Expert panel on Genetic Counseling in Reproductive Genetics and Prenatal Diagnosis: Genetic, Ethical and Legal Issues**

91th Seminar in Clinical Genetics, "Genetic Counseling", Tehran, Iran, 27 August 2015

11- **Screening of Common Aneuploidies: NIPT versus Conventional Genetic Testing**

90th Seminar in Clinical Genetics, "Prenatal Screening ", Tehran, Iran, 30 July 2015

12- **Preimplantation Genetic Screening using Next Generation Techniques: PGS-NGS**

89th Seminar in Clinical Genetics, "Developments in Increasing the Pregnancy Rate Of Assisted Reproductive Techniques, 28 May, 2015

13- **Technical Aspects of Gender Selection Using Preimplantation Genetic Diagnosis (PGD)**

88th Seminar in Clinical Genetics, "Gender selection: fact or fiction", Tehran, Iran, 30 April 2015

14- **Prenatal diagnosis of genetic disorders**

87th Seminar in Clinical Genetics, "Maternal, Fetal and Neonatal Health", Tehran, Iran, 27 Nov 2014

15- Clinical Application of PGD for a Wide Range of Single Gene Disorders using Single Cell Whole Genome Amplification technology

86th Seminar in Clinical Genetics, "PGD of Single Gene Disorders", Tehran, Iran, 15 Oct 2014

16- Increasing the implantation rate: preimplantation genetic screening using next generation sequencing (PGS-NGS)

85th Seminar in Clinical Genetics, "Repeated Implantation Failure: Causes and Management",Tehran, Iran, 25 Sep 2014

17- ESHRE PGD Report

84th Seminar in Clinical Genetics, "Gender selection: fact or fiction?", Tehran, Iran, 14 Aug 2014

18- A Comprehensive Approach to Genetic Investigation of Products of Conception

83th Seminar in Clinical Genetics, "genetic and pathologic investigation of products of conception", Tehran, Iran, 10July 2014

19- Preimplantation genetic diagnosis, gender selection using FISH Technique

82th Seminar in Clinical Genetics, "Gender selection: fact or fiction", Tehran, Iran, 19 June 2014

20- Technical Aspects of Prenatal Aneuploidy Detection using Cell-free fetal DNA

81th Seminar in Clinical Genetics, "Noninvasive Prenatal Testing of Common Chromosomal Abnormalities", Tehran, Iran, 8 May 2014

21- Noninvasive Prenatal Testing of Single Gene Disorders using Cell-Free Fetal DNA

80th Seminar in Clinical Genetics, "Noninvasive Prenatal Testing", Tehran, Iran, 27 Feb 2014

22- Application of Next Generation Sequencing in Mutation Analysis of Genodermatoses

78th Seminar in Clinical Genetics "Genomic Approach to Genodermatoses", Tehran, Iran, 26 Dec 2013

23- Application of Next Generation Sequencing in Mutation Analysis of Inborn Errors of Metabolism

77th Seminar in Clinical Genetics, "Genomic Approach to Inborn Errors of Metabolism", Tehran, Iran, 28 Nov 2013

24- Application of Next Generation Sequencing in Mutation Analysis of Retinitis Pigmentosa

76th Seminar in Clinical Genetics, "Genomic Approach to Inherited Visual Impairment", Tehran, Iran, 14 Nov 2013

25- Application of Next Generation Sequencing in Mutation Analysis of Retinitis Pigmentosa

75th Seminar in Clinical Genetics, "Genomic Approach to Inherited Visual Impairment", Tehran, Iran,31 Oct 2013

26- Application of Next Generation Sequencing in Mutation Analysis of Hereditary Ataxias

74th Seminar in Clinical Genetics, "Genomic Approach to Hereditary Ataxias", Tehran, Iran, Sep 2013

27- Next Generation Sequencing in Hereditary Cancers

73th Seminar in Clinical Genetics , "A Comprehensive Approach to Diagnosis and Prevention of Hereditary Cancers", Tehran, Iran, 4 July 2013

28- Stepwise Diagnostic Approach to Intellectual Disability: Conventional as Well as Next Generation Sequencing Techniques

72th, "Genomic Approach to Intellectual Disability", Tehran, Iran, 30 May 2013

29- Basic Principles of Next Generation Sequencing Technology: Comparing Different Platforms

71th Seminar in Clinical Genetics, "Application of New Genomic Approach to Solve Old Problems of Medical Genetics: Advantages, Challenges and Future Prospects", Tehran, Iran, 2 May 2013

30- PGD using PCR-Based Techniques: Advantages and Limitations

70th Seminar in Clinical Genetics, "Preimplantation Geneic Diagnosis of Single Gene Disorders" , Tehran, Iran, 28 Feb 2013

31- PGD using Hybridization-Based Techniques: Advantages and Limitations

69th Seminar in Clinical Genetics, "Preimplantation Geneic Diagnosis of Chromosomal Abnormalities" ,Tehran, Iran, 7 Feb 2013

32- Hereditary Thrombophilia in Recurrent Miscarriage

68th Seminar in Clinical Genetics, "Thrombophilia and Recurrent Miscarriage", Tehran, Iran, 10 Jan 2013

33- Genetic investigation of products of conception using molecular cytogenetic techniques

67th Seminar in Clinical Genetics, "Chromosomal Abnormalities in Recurrent Miscarriages", Tehran, Iran, 13 December 2012

- Seminars 1-66 were held at Imam Khomeini hospital, Tehran University of Medical Sciences, 2003-2009

INVITED PRESENTATIONS (SELECTED)

1- **Head of Panel Discussion on "Personalized Medicine in Clinical Practice"**
1st Congress on Personalized Medicine, Tehran, Iran, 25 February 2017

2- **Genetics of Endometriosis**
The 3rd National and the 1st International Congress on Endometriosis and Minimally Invasive Gynecology (EMIG), Tehran, Iran, 25-27 October 2016

3- **"A Comprehensive Genetic Approach to Premature Ovarian Insufficiency: New Opportunities with Next Generation Sequencing"**
Laboratory and Clinic Conference, Avicenna Biotechnology Research Institute , Tehran, Iran, 19 June 2016

4- **The Road from Next-generation Sequencing to Personalized Medicine: Technical Aspects**
3rd Congress on Novel & Innovative Laboratory Technologies, Tehran, Iran, 6 October 2015

5- **Genetic Counseling in Prenatal Screening of Aneuploidies**
Conference on "Clinical and Laboratory Aspects of Prenatal Screening and Its Management"**,** Tehran, Iran, 26 November 2015

6- **Expert panel on recurrent miscarriage: causes and recommended treatments**
Conference on "recurrent miscarriage", Tehran, Iran, 12 November 2015

7- **Clinical outcome of Increased Nuchal Translucency Detected In Prenatal Screening of Aneuploidies: Data Presentation of more than 45000 Screened Pregnancies**

3rd Congress on "Diagnosis and Treatment Of Fetal Anomalies", Tehran, Iran, 1 October 2015

8- **Advanced Genetic Tools in Breast Cancer: Targeted Treatment of Breast Cancer using Next Generation Sequencing**

Conference on "Approach to Breast Cancer", Tehran, Iran, 1 October 2015

9- **Clinical Application of Next Generation Sequencing in Prenatal Diagnosis of Inborn Errors of Metabolism**

Conference on "Mother and Newborn Health", Tehran, Iran, 17 September 2015

10- **Preimplantation Genetic Diagnosis (PGD) using Single-Cell Whole Genome Amplification: Report on 3 Year Experience in A Clinical Setting for a Wide Range of Genetic Disorders**

International Congress on Reproduction, ISERB, Board Member of Panel: "Genetics and Epigenetics in reproduction", 25 May 2015

11- **Preimplantation Genetic Screening using Next Generation Techniques: PGS-NGS**

Conference on "Recurrent Miscarriage: Causes and Management", Tehran, Iran, 5 March 2014

12- **Noninvasive Prenatal Testing (Cell-Free Fetal DNA)**

1st International Congress of Minimally Invasive Gynecology and Obstetrics, Tehran, Iran, 9 Jan 2015

13- **Noninvasive Prenatal Testing (Cell-Free Fetal DNA)**

Conference on "Different Aspects of Fetal Health", Tehran, Iran, 21 Aug 2014

14- **Genetics in Congenital Disorders of Gastrointestinal and Urinary tract**

26th International Congress of Pediatrics**,** Tehran, Iran, 18 Oct 2014

15- **Preimplantation Genetic Diagnosis (PGD) of Single Gene Disorders**

Laboratory and Clinic Conference, Avicenna Biotechnology Research Institute , Tehran, Iran, 18 June 2014

16- **Genetics in Early Diagnosis, Treatment and Prevention of Breast Cancer**
10th International Congress of Obstetrics and Gynecology, Board Member of Panel: "Prophylactic Bilateral Mastectomy", Tehran, Iran, 26 Sep 2013

17- **First and Second Trimester Screening**
Laboratory and Clinic Conference, "Prenatal Screening of Chromosome Abnormalities", Avicenna Biotechnology Research Institute, Tehran, Iran, 5 May 2013

18- **Hereditary Breast and Ovarian Cancer**"Breast Cancer Seminar", Iranian Society of Pathology, 2010 May

19- **Genetic Education Series**, ministry of health, 2006, Tehran, Iran

20- **Genetic Education Series**, rehabilitation and welfare organization, 2007, Tehran, Iran

21- **Integrating genetic counseling into Iranian PHC program**, *WHO regional meeting, Tehran, 2005*

22- **Toward a genomic approach for prevention of diseases**, *WHO, EMRO meeting, Tehran 2004*

23- **Applications of Genetics in Cancer epidemiology**, Tehran, 2005, Cancer epidemiology workshop

24- **Genetics for the pediatricians**, *Tehran 2005, International pediatrics congress*

25- **Genetics for the obstetricians**, Tehran, *Nov, 2005, Ob&Gyn CME*

26- **Preconception genetic counseling**, *Tehran 2005, Ob&Gyn CME*

27- Genetic counseling for the families affected with familial ovarian and breast cancer, clinical genetics conference, *Tehran 2005*

28- Applications of Molecular cytogentics in clinical practice, *International congress in Genetics held by National Center of Genetics Engineering and biotechnology. Tehran 2004*

29- Advances in Genetics, an update, *Iranian national conference in laboratories quality improvement, Tehran 2005*

***30*- Applications of DNA micro array technology in the diagnosis of cancer**, *Annual conference of the Iranian Cancer Institute, December 2002, Tehran, Iran*

31- New molecular approaches in the presymptomatic and early detection of breast cancer, *Annual conference of the Iranian Surgical Society, May 2003, Tehran, Iran*

32- New approaches to preimplantation genetic diagnosis. Second regional workshop on the application of advanced molecular methods for the diagnosis of human genetic disease. 2-6 May 2002. Tehran, Iran.

33- Preimplantation genetic diagnosis: current status and future developments. Human genome diversity (HGD) conference. 7-8 May 2002. Tehran, Iran.

34- Principles of genetic counselling. Feb 2002, Tehran, Iran.

35- Applications of computer and information technology in new medicine, Faculty of Medicine, December 2001, Tehran Iran

36- New developments in cytogenetics, 1st congress on Medical Genetics and Molecular Medicine, December 2001, Tehran, Iran

37- Genetics in modern medicine, seminars in pathology, October 2001, Tehran, Iran

38- Genetics of addiction, Iranian conference on preventive measures against drug abuse, 2000, Zahedan, Iran

39- Genetics in modern medical practice, First Iranian congress on Human Genetics, 2000, Shahr-e-Kurd, Iran

40- Applications of molecular cytogenetics in modern medicine, May 2000, School of Public Health, Tehran University, Tehran Iran

41- Cytogenetics, past, present and future Cytogenetic Symposium, Glasgow 23rd May 1997, Glasgow, UK

42- Comparartive Genomic Hybridization in clinical genetics, University of Glasgow seminar in Medical Genetics, 11th February 1998, Glasgow, UK

43- Copmarative Genomic Hybridization to DNA microarrays, British Human Genetics Conference 28th September 1998, York, UK

44- Human Genome Project: clinical implications, Tehran Medical School, Feb 2001, Tehran, Iran

45- Approaches to rapid cytogenetic analysis in newborns, Department of Medical Genetics seminar, 25Th October 1995, University of Glasgow, Glasgow, UK

46- A novel strategy for cryptic telomeric translocation screening in patients with idiopathic mental retardation, British Human Genetics Conference, 17th September 1997, University of York, York, UK

ABSTRACTS

1- 2nd International Congress on Reproduction (ISERB 2016), Tehran, Iran, 18-20 May 2016

Journal of Reproduction and Infertility, Volume 17, Issue 2, May 2016 (Supplement)
I-35: Association of fetal aneuploidy with mother's genotype
Maryam Rafati, Hosna Amiri, Faezeh Mohamadhashem, Asieh Darunkolaee, Zeinab Barati, Saeed Reza Ghaffari

2- The 9th International & 14th National Congress on Quality Improvement in Clinical Laboratories, Tehran, Iran, 19-22 April 2016
Clinical Application of Next Generation Sequencing in Management and Treatment of Recurrent Miscarriage
Saeed Reza Ghaffari, Maryam Rafati

3- 3rd Congress on Novel and Innovative Laboratory Technologies, Tehran, Iran, 4-6 October, 2015
Personalized Medicine: Present Opportunities, Future Prospects
Saeed Reza Ghaffari, Maryam Rafati

4- 3rd Congress on Novel and Innovative Laboratory Technologies, Tehran, Iran, 4-6 October, 2015
Clinical Application of Next Generation Sequencing (NGS) in Personalized Medicine
Saeed Reza Ghaffari, Maryam Rafati

5- International Congress on Reproduction (ISERB 2015)), Tehran, Iran, 23-25 May 2015
O-37: Preimplantation Genetic Diagnosis: Iran and World Experience
Saeed Reza Ghaffari, Maryam Rafati, Mohammad Reza Sadeghi, Mohammad Mahdi Akhondi

6- International Congress on Reproduction (ISERB 2015)), Tehran, Iran, 23-25 May 2015
O-65: New Opportunities for Improvement of Implantation Rate using Preimplantation Genetic Screening (PGS)
Saeed Reza Ghaffari, Maryam Rafati, Mohammad Reza Sadeghi, Mohammad Mahdi Akhondi

POSTER

- Comparative Genomic Hybridisation in the investigation of haematological malignancies, British Human Genetics Conference, 15th-17th September 1997, University of York, York, UK

- Comparartive Genomic Hybridisation reveals the origin of a mosaic, supernumerary ring chromosome in a 70 year old lady with mental retardation, British Human Genetics Conference, 15th-17th September 1997, University of York, York, UK

- Overnight Comparative Genomic Hybridisation, a practical approach to rapid chromosome analysis. British Human Genetics Conference, 28th-30th September 1998, University of York, York, UK

MEDIA REPORTS

A) **The first Iranian Child Born after Preimplantation Genetic Diagnosis (PGD) of Cystic Fibrosis**

IRNA
http://www8.irna.ir/fa/News/81412541/

B) **Opening Ceremony of Launching the First National Next Generation Sequencing Facility in Iran**

http://www.behdasht.gov.ir/news/%DA%AF%D8%B2%D8%A7%D8%B1%D8%B4+%D8%AA%D8%B5%D9%88%DB%8C%D8%B1%DB%8C/119765/%D8%A7%D9%81%D8%AA%D8%AA%D8%A7%D8%AD+%D8%AF%D9%BE%D8%A7%D8%B1%D8%AA%D9%85%D8%A7%D9%86+%D9%86%D8%B3%D9%84+%D8%AC%D8%AF%DB%8C%D8%AF+%D8%AA%D8%B9%DB%8C%DB%8C%D9%86+%D8%AA%D9%88%D8%A7%D9%84%DB%8C+%D9%87%D8%A7%DB%8C+%DA%98%D9%86%D8%AA%DB%8C%DA%A9%DB%8C-+%D9%85%D9%88%D8%B3%D8%B3%D9%87+%D9%BE%D8%B2%D8%B4%DA%A9%DB%8C+%D9%86%D8%B3%D9%84+%D8%A7%D9%85%DB%8C%D8%AF

http://khabarfarsi.com/n/11687415

http://tnews.ir/news/A7DC36093827.html

❖ TV INTERVIEWS

No.1
Date Published: 18 Jan 2015
Author: Shahsavar Hoseini (reporter of "IRIB News Agency")
Name of Journal/Magazine/Newspaper/Website: TV Channel 1, News

No.2
Date Published: 29 Jan 2015
Author: Sharif (reporter of "Salamat News Agency")
Name of Journal/Magazine/Newspaper/Website: Salamat TV Channel

❖ OTHERS

No.3
Date Published: 18 Jan 2015

Author: Shahsavar Hoseini (reporter of "IRIB News Agency")
Name of Journal/Magazine/Newspaper/Website: Official website of "IRIB News Agency" (http://www.iribnews.ir/NewsText.aspx?ID=365540)

No.4
Date Published: 18 Jan 2015
Author: Reporter of "Ministry of Health and Medical Education "
Name of Journal/Magazine/Newspaper/Website: Official website of "Ministry of Health and Medical Education " (http://www.behdasht.gov.ir/news)

No.5
Khabar Farsi (http://khabarfarsi.com/ext/11687415)

No.6
TNews (http://tnews.ir/news/A7DC36093827.html)

No.7
Parseek Khabar (http://news.parseek.com/Url/?id=10225251)

No.8
Khabargo

C) Genetic Investigation of Products of Conception

❖ **TV INTERVIEWS**

Genetic investigation of products of conception

Jam-e-Jam TV, 2014

❖ **OTHERS:**

No.1
Mehrkhaneh (http://mehrkhane.com/fa/news)

No.2
Fars News (http://www.farsnews.com/newstext.php?nn=13930920000238)

D) Three-parent babies

Interviews:
No.1
Nameh News (http://namehnews.ir/fa/news/)

No.2

Khabar Farsi (http://khabarfarsi.com/n/7047338)

E) Opening the "Comprehensive Genetic Center", Hope Generation Foundation

No.1
Fars News (http://www.farsnews.com/plarg.php?nn=168184&st=388481)
(http://www.farsnews.com/plarg.php?nn=168188&st=388481)

13 TV Interviews

1- **Polycystic Ovarian Syndrome (PCOS)**
Genetic Aspects of PCOS, "End-of-Infertility" Series, channel 4, 24 Dec 2015

2- **Premature Ovarian Insufficiency (POI)**
Genetics in POI, "End-of-Infertility" Series, channel 4, 5 Nov 2015

3- **Polycystic Ovarian Syndrome (PCOS)**
Genetic Aspects of PCOS, "End-of-Infertility" Series, channel 4, 15 Aug 2015

4- **Premature Ovarian Insufficiency (POI)**
Genetics in POI, "End-of-Infertility" Series, channel 4, 7 March 2015

5- **Common Misconceptions in Genetic Counseling**
"End-of-Infertility" Series, Channel 4, 28 Feb 2015

6- **Next Generation Sequencing, Opening of the First Next Generation Sequencing Facility in Iran**
Channel Salamat, 29 Jan 2015

7- **Opening Ceremony for the Launch of the First National Next Generation Sequencing Facility in Iran**
IRIB News, Channel 1, 18 Jan 2015

8- **Genetic disorders causing infertility and/or recurrent miscarriage**
"End-of-Infertility" Series, Channel 4, 6 Dec 2014

9- **Prevention of genetic disorders using preimplantation genetic diagnosis (PGD)**

"End-of-Infertility" Series, Channel 4, 29 Nov 2014

10- Genetic tests
"End-of-Infertility" Series, Channel 4, 22 Nov 2014

11- Genetics in recurrent miscarriage
"End-of-Infertility" Series, Channel 4, 21 June 2014

12- Genetic investigation of products of conception
Jam-e-Jam TV, 2014

13- Genetic counseling
"Come Back to Home" series, Channel 5, 2013

CLINICAL AND DIAGNOSTIC EXPERTISE

1- Genetic Counseling of more than **50000** Families

2- Next Generation Sequencing (Ion Torrent Platform)

- Whole Exome Sequencing (Ion Ampliseq Exome RDY)
- Targeted Capture Sequencing (Ion Ampliseq)
- Low Pass Aneuploidy Detection
- High resolution HLA typing using NGS
- NGS analysis on DNA obtained from single cell whole genome amplification (PGS-NGS)
- NGS study on products of conception

3- Preimplantation Genetic Screening (PGS) of Aneuploidies using Next Generation Sequencing (NGS)

4- Preimplantation Genetic Diagnosis (PGD) of Single Gene Disorders including:

- Beta thalassemia
- Hearing loss
- Cystic fibrosis
- Spinal Muscular Atrophy
- Duchenne Muscular Dystrophy
- Achondroplasia
- Arylsulphatase Deficiency (Metachromatic Leukodystrophy)
- Fibrodysplasia Ossificans Progressiva

5- Preimplantation Genetic Diagnosis (PGD) of Chromosomal Abnormalities and Gender Selection using FISH Technique (More Than 2000 Embryos)

6- Prenatal Diagnosis of:

- Beta thalassemia
- Alpha thalassemia
- Tyrosinemia
- Crigler-Najar syndrome
- Cockayne syndrome

- Laron Syndrome
- ARC Syndrome
- Hypohidrotic Ectodermal Dysplasia
- Congenital Adrenal Hyperplasia (HSD3B2 mutation)
- Hearing Loss (GJB2 mutation)
- Wolfram syndrome
- Alstrom syndrome
- Hereditary Diffuse Gastric Cancer (CDH1 mutation)
- Maple syrup urine disease
- Williams Syndrome
- DiGeorge Syndrome
- Spinal Muscular Atrophy
- Duchenne Muscular Dystrophy

7- Rapid Prenatal Aneuploidy Detection (~1200 Amniotic Fluid or CVS samples)

8- Postnatal Diagnostic Tests

- Wilson Disease
- Tuberous Sclerosis
- Glutaric acidemia
- Neurofibromatosis
- Glycogen storage disease
- GM1 Gangliosidosis
- Methyl Malonic Acidemia (MMA)
- Niemann Pick Disease
- Microcephaly
- gastrointestinal defectss and immunodeficiency syndrome (TTC7A mutation)
- Malignant migrating partial seizures (KCNT1 mutation)
- Hearing loss
- Retinitis pigmentosa
    - Autosomal recessive
    - Autosomal dominant
    - X-linked
- Fragile-X syndrome
- Subtelomeric rearrangements
- Williams Syndrome

- DiGeorge Syndrome
- Prader-Willi Syndrome
- Microdeletion/duplication syndromes
- Spinal Muscular Atrophy
- Duchenne/Becker Muscular Dystrophy
- Charcot Marie Tooth
- Hemophilia A

9- Genetic Investigation of Products of Conception (520 patients)

TECHNICAL EXPERTISE

- **Genomics**

Array based technology, suspension hybridization (Luminex), Array CGH,

- Next Generation Sequencing
  - Whole exome sequencing
  - Targeted capture sequencing (design gene sets)

- **Molecular Genetics**:

DNA and RNA extraction (blood, fixed cells, solid tumour, bone marrow, …), Restriction fragments length polymorphism, polymerase chain reaction (standard PCR, DOP-PCR) and RT PCR, Design of oligonucleotide primers, Protein truncation test (PTT), Direct PCR product sequencing, Magnetic separation of cells and DNA molecules, Southern Blotting, ARMS, ASO,.Design and application of DNA chips technology (microarrays)

- Single-Cell Whole Genome Amplification
- Preimplantation Genetic Diagnosis (PGD) of Chromosomal Abnormalities (single cell FISH)
- Preimplantation Genetic Diagnosis (PGD) of Single Gene Disorders
- Prenatal diagnosis of a wide range of single gene disorders

- **Molecular Cytogenetics:**

Prenatal Diagnosis of Chromosomal Abnormalities
YAC and cosmid cloning, Fluorescence in situ hybridization (Multi-colour FISH, Direct and indirect labelling method.…), Single cell FISH: preimplantation genetic diagnosis (PGD), Comparative Genomic Hybridization (Global analysis, profile analysis), Single cell CGH, Primed in situ

hybridization (PRINS), Microarray Chips, Microarray CGH, Fiber FISH, Spectral Karyotyping (SKY) and M-FISH , CCK, QF-PCR, MLPA

- **Conventional Cytogenetics**:

Tissue culture (blood, bone marrow, solid tumor, AF), chromosome preparation, banding techniques, specialized techniques (High resolution banding, fragile sites, sister chromatid, …), Standard chromosome analysis and Karyotyping,

COMPUTER SKILLS AND SOFTWARE

Medical Genetic-related software

- Ion Reporter
- Torrent Suite, Proton Server
- Torrent Suite, PGM Server
- Mutation Surveyor
- Sequin
- Mutalyzer
- Chromas
- Progeny
- Coffalyser
- GeneMapper
- Genetic analyser data collection

General computer skills

- Dynamic Websites (see http://medicine.tums.ac.ir)

- Windows:

Advanced level: Windows 9x, 2003 and XP. Vista Microsoft office 2003 (Word, PowerPoint, Excel, Access, Frontpage), FoxPro , Video capture and editing, Minitab , Photoshop , Macromedia Flash , Macromedia Authoware and..

Basic level: Visual studio 2001 (Visual basic 6, Visual C++, Visual J++)

- Macintosh:

Advanced level: Mac OS , Mac Draw Pro, Claris Works, IP lab image analysis software, Smart Capture image analysis system, VYSIS CGH, FISH and automatic karyotyping station V.3. Video capture and editing

- Internet:

- HTML language and Java for web authoring. Front page 2007 and 2007 (XP). FTP, Design of a small network (LAN), .NET, dynamic web site)

RESEARCH INTERESTS

- Genome based technologies including next generation sequencing and exome sequencing
- Application of molecular and molecular cytogenetics techniques, including FISH, PRINS, CGH, SKY, CCK and DNA micoarray technology for the identification of new disease genes using the recently published Human Genome Project data.
- Improvement of current molecular cytogenetics techniques for detection of cryptic abnormalities in patients with cancer and constitutional chromosome abnormalities.
- Development and application of FISH and PCR techniques for the preimplantation diagnosis of molecular and chromosomal abnormalities.
- Development and application of single cell CGH and SKY as a new approach for genome wide screening preimplantation genetic diagnosis
- Primary health care approaches for prevention and control of congenital and genetic disorders
- Fetal cells in maternal blood, Fetal DNA in maternal serum, maternal serum biochemical markers in 1st and second trimester screening
    - Suspension hybridization techniquesPersonalized Medicine
    - Genomic Approach to Heterogeneous Disorders using Next Generation Sequencing
    - Prenatal Diagnosis of genetic disorders
    - Preimplantation Genetic Diagnosis

REFERENCES

❖ **Professor J Michael Connor**

Institute of Medical Genetics

Yorkhill Hospitals Campus

Glasgow G3 8SJ

UK

- **Dr Douglas E Wilcox**

Institute of Medical Genetics,

Yorkhill Hospitals Campus

Glasgow G3 8SJ

UK

- **Dr Elizabeth Boyd**

Institute of Medical Genetics

Yorkhill Hospitals Campus

Glasgow G3 8SJ

UK