Curriculum Vitae

MARYAM RAFATI MD PhD

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2017-5

PERSONAL INFORMATION:

Date and Place of Birth	1 April 1978, Tehran, Iran
Nationality	Iranian
Marital Status	Married

EDUCATIONAL BACKGROUND:

2012	PhD Degree of Medical Genetics, Tehran University of Medical Sciences, Tehran, Iran Grade Point Average: **17.36 / 20** Quality of academic records is assessed as **"SUPERIOR"** by American Association of Collegiate Registrars and Admissions Officers (AACRAO), Office of International Education Services
2005	MD degree from Faculty of Medicine, Tehran University of Medical Sciences, Tehran, Iran Grade Point Average: **17.64 /20**
1996	High School Graduation Diploma, Khadijeh Kobra (Farzanegan) High School, Tehran, Iran Grade Point Average: **19.77 /20** Ranked **30th** among ~400000 students applying for medicine in National Admission Competition for Iranian Universities Entrance Exam.

THESIS:

PhD (2006-2012)	Design A Comprehensive Stepwise Diagnostic Approach to Mental Retardation by Determining the Molecular Profile of DNA Copy Number Abnormalities
MD (2002-2003)	Prevalence of Metabolic Syndrome in obese elementary students of 6th district of Tehran

ACADEMIC EXPERIENCE

2016-present	Founding Member, Hope Generation Fetal Health Research Institute, Tehran, Iran
2016-present	Member of Board of Directors, Hope Generation Foundation, Tehran, Iran
2012-Present	Assistant Professor, Department of Reproductive Genetics, Avicenna Biotechnology Research Institute, Tehran, Iran
2012-present	Scientific Board Member, National Committee for Genetic Counseling Network, Ministry of Health and Medical Education
2014-present	Scientific Board Member, Technical Committee of Reference Health Lab, Ministry of Health and Medical Education
2016-present	Associate Editor; Journal of Medical Biotechnology
2012-Present	Scientific Board Member, Seminars in Clinical Genetics
2013-Present	Peer Reviewer, Avicenna Journal of Medical Biotechnology
2012-Present	Peer Reviewer, Journal of Reproduction and Infertility
2012-present	Member of "Expert Committee on Maternal, Fetal and Neonatal Health", Avicenna Infertility Clinic, Avicenna Biotechnology Research Institute
2012-present	Member of "Expert Committee on Recurrent Miscarriage", Avicenna Infertility Clinic, Avicenna Biotechnology Research Institute
2006-present	Member of “Iranian Medical Genetics Society”

SUPERVISED THESES AND DISSERTATIONS

2014-Present, Co-supervisor
PhD Student of Medical Genetics

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

2015-Present, Co-supervisor
MSc Student of Medical Genetics

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

2015-2016, Co-supervisor
MSc Student of Medical Genetics

Determining the association of the variants linked to PLK4 gene in mother with fetal aneuploidy

2014-2016, Co-supervisor
MSc Student of Developmental Genetics

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

PROFESSIONAL EXPERIENCE

2015-present **Founding Member**	**Hope Generation Research Center,** Tehran, Iran
2016-present **Member of Board of Directors**	**Hope Generation Foundation,** Tehran, Iran
2014-present **Technical Director**	**Next Generation Sequencing Department** Hope Generation Foundation, Tehran, Iran
2012-present **Technical Director**	**Clinical Genetics Laboratories,** Avicenna Infertility Clinic, Avicenna Biotechnology Research Institute
2009-present **Technical Director**	**Molecular Genetics Laboratory** Hope Generation Foundation, Tehran, Iran
2012-present **Clinical Director**	**Genetic Counseling Clinic** Avicenna Infertility Clinic, Avicenna Biotechnology Research Institute
2012-present **Clinical Director**	**"Maternal, Fetal and Neonatal Health" Clinic** Avicenna Infertility Clinic, Avicenna Biotechnology Research Institute
2009-present **Genetic Counselor**	**Genetic Counseling Clinic** Gene Clinic, Tehran, Iran
2010-2016 **Technical Director**	**Molecular Genetic Laboratory** Gene Clinic, Tehran, Iran

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:

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

TEACHING EXPERIENCE

2012-Present
PhD students of Reproductive Health **Genetics in Infertility: Preimplantation Genetic Screening and Diagnosis**
(5 Semesters) Tehran University of Medical Sciences
Avicenna Research Institute

2013-Present
Infertility Fellowship
(3 Semesters) **Genetic Counselling In Infertility**
Genetic Tests in Infertility
Chromosomal Abnormalities
Single Gene Disorders in Infertility

2016 **Genetic Counseling: Cytogenetics**
General Practitioners (MD) State Welfare Organization of Iran

2016
PhD Students of Reproductive Biology **PGS-NGS: preimplantation genetic screening (PGS) of aneuploidies using next generation sequencing**
Tehran University of Medical Sciences

2016
PhD Students of Reproductive Biology **preimplantation genetic diagnosis (PGD) of single gene disorders using single cell whole genome amplification**
Tehran University of Medical Sciences

2015
PhD Students of Reproductive Biology **Molecular Cytogenetics: QF-PCR, Array CGH**
Tehran University of Medical Sciences

2015
PhD Students of Reproductive Biology **Molecular Cytogenetics: MLPA**
Tehran University of Medical Sciences

2015
PhD Students of Reproductive Biology **Chromosme abnormalities**
Tehran University of Medical Sciences

2015 **Genetic Counseling: Cytogenetics**
General Practitioners (MD) State Welfare Organization of Iran

	Avicenna Infertility Clinic, Avicenna Research Institute
2009-2011 PhD students of Medical Genetics (4 Semesters)	**Genetic Counselling, Practical Course** Department of Medical Genetics, Tehran University of Medical Sciences
2009-2011 Maternal Fetal Medicine Fellowship (3 Semesters)	**Medical Genetics Rotation** Department of Obstetrics and Gynecology, Tehran University of Medical Sciences
2011 Neonatalogy fellowship	**Medical Genetics Rotation** department of paediatrics and neonatalogy, Tehran University of Medical Sciences
2009 MSc Students	**Basics of Genetics** school of Nursing, Tehran University of Medical Sciences
1991 -2001	Teaching Genetics to High School Students, Farzanegan High School, Tehran.

MEMBERSHIP HISTORY:

2016-present	Founding Member, Hope Generation Research Center, Tehran, Iran
2016-present	Member of Board of Directors, Hope Generation Foundation, Tehran, Iran
2014-present	Member of "Pars Sina Yakhteh" Company (R&D Department)
2002-2003	Member of "Critical Thinking ", Students Research Center, Tehran University of Medical Sciences
1997-2001	Member of "Laser in Medicine" Group, Students Research Center, Tehran University of Medical Sciences (Project: Effect of low power He-Ne Laser in remission of bedsore)
1996-2000	Member of "Students Research Center", 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
Associate Principal Scientist

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

- Hope Generation Foundation

2013-2014
Associate 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
Associate Principal Scientist

Optimization and Clinical Application of Single Cell Whole Genome Amplification Technique

- Avicenna Infertility Clinic, Avicenna Research Institute

2010-2014
Associate 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
Associate 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
Associate 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
Associate 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
Associate Principal Scientist

Design and Establishment of "Comprehensive Genetic Center" and "clinical genetics laboratories"

- Imam Hospital Complex, Tehran University of Medical Sciences

RESEARCH EXPERIENCE:

Associate Principal Investigator:

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

Collaborating in the Research Project:

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

2002 **Children Obesity**, Endocrine and Metabolism Research Center, Tehran University of Medical Sciences

1997 **Photodynamic therapy**,
"Laser in medicine" group, Students Research Center, Tehran University of Medical Sciences

1996 **Effect of low power He-Ne Laser in Remission of Bedsore**,
"Laser in medicine" group, Students Research Center, Tehran University of Medical Sciences

CURRENT RESEARCH PROJECTS:

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

CERTIFICATES AND AWARDS:

PATENT **Patent No: 84457** **Date: August 2, 2014**	**Sexed Frozen Bovine Embryos** Avicenna Research Institute
AWARD **2015**	**Top Physician** Avicenna Research Institute
AWARD **2013**	**Top Lecturer** Annual Seminars and Conferences in Avicenna Research Institute
CERTIFICATES	
2007	Certificate of systematic search (ref viz) workshop held by Vice Chancellor for Research
2006	Certificate of Endnote workshop held by Vice Chancellor for Research
2005-December	Certificate of scientific writing workshop (elementary and intermediate level) held by Vice Chancellor for Research
2005-October	Certificate of "9th Hybrid course in cancer genetics"held by European School of Genetic Medicine in ESGM's main training center at Bertinoro di Romagna , Italy.
1998-May	Certificate of "Research methods "workshop held by Students Research Center of Tehran University of Medical Science
1993	Award of the First Exhibition of Innovations of Tehran High School Students For Designing The Benzene Model

PAPERS

Published Papers:

1- Primary ovarian insufficiency with t(5;13): a case report and literature review on disrupted genes.
Mohamadhashem F, **Rafati M**, Hoseininasab F, Rostami S, Tabatabaie R, Rezai S, Keramatipour M, Ghaffari SR.
Climacteric. 2017 Apr 28:1-5. doi: 10.1080/13697137.2017.1316255. [Epub ahead of print]

PMID: 28453298

2- A novel *ACVR1* mutation detected by whole exome sequencing in a family with an unusual skeletal dysplasia
Maryam Rafati, Faezeh Mohamadhashem, Azadeh Hoseini, Fatemeh Hoseininasab, Saeed RezaGhaffari
Eur J Med Genet. 2016 Jun;59(6-7):330-6. doi: 10.1016/j.ejmg.2016.05.007. Epub 2016 May 13.
PMID: 27182040

3- Prenatal Diagnosis of Tyrosinemia Type 1 Using Next Generation Sequencing.
Rafati M, Mohamadhashem F, Hoseini A, Ramandi SD, Ghaffari SR.
Fetal Pediatr Pathol. 2016 Apr 19:1-4. [Epub ahead of print]

PMID: 27093575

4- Increasing the yield in targeted next-generation sequencing by implicating CNV analysis, non-coding exons and the overall variant load: the example of retinal dystrophies.
Eisenberger T, Neuhaus C, Khan AO, Decker C, Preising MN, Friedburg C, Bieg A, Gliem M, Charbel Issa P, Holz FG, Baig SM, Hellenbroich Y, Galvez A, Platzer K, Wollnik B, Laddach N, Ghaffari SR, **Rafati M**, Botzenhart E, Tinschert S, Börger D, Bohring A, Schreml J, Körtge-Jung S, Schell-Apacik C, Bakur K, Al-Aama JY, Neuhann T, Herkenrath P, Nürnberg G, Nürnberg P, Davis JS, Gal A, Bergmann C, Lorenz B, Bolz HJ.

PLoS One. 2013 Nov 12;8(11):e78496. doi: 10.1371/journal.pone.0078496.

5- Familial intellectual disability in an Iranian family with a novel truncating mutation in CEP290.

Ghaffari SR, **Rafati M**, Ghaffari G, Morra M, Tekin M.

Clin Genet. 2014 Oct;86(4):387-90. doi: 10.1111/cge.12296. Epub 2013 Oct 31.

6- Heterozygosity assessment of five STR loci located at 5q13 region for preimplantation genetic diagnosis of spinal muscular atrophy.

Korzebor A, Derakhshandeh-Peykar P, Meshkani M, Hoseini A, **Rafati M**, Purhoseini M, Ghaffari SR.

Mol Biol Rep. 2013 Jan;40(1):67-72. doi: 10.1007/s11033-012-2011-3. Epub 2012 Nov 7.

PMID: 23132709 [PubMed - in process]

7- Familial Williams-Beuren syndrome ascertained by screening rather than targeted diagnosis.

Rafati M, Seyyedaboutorabi E, Brujerdi R, Moossavi S, Ghaffari SR.

Clin Dysmorphol**.** Jul;21(3):118-23. [Epub ahead of print]

PMID: 22473150 [PubMed - as supplied by publisher]

8- "Familial" versus "Sporadic" intellectual disability: contribution of common microdeletion and microduplication syndromes.

Rafati M, Seyyedaboutorabi E, Ghadirzadeh MR, Heshmati Y, Adibi H, Keihanidoust Z, Eshraghian MR, Javadi GR, Dastan J, Mosavi-Jarrahi A, Hoseini A, Purhoseini M, Ghaffari SR.

Mol Cytogenet. 2012 Jan 29;5(1):9.

PMID:22283845 [PubMed - in process]
Free PMC Article

9- "Familial" versus "sporadic" intellectual disability: contribution of subtelomeric rearrangements.

Rafati M, Ghadirzadeh MR, Heshmati Y, Adibi H, Keihanidoust Z, Eshraghian MR, Dastan J, Hoseini A, Purhoseini M, Ghaffari SR.

Mol Cytogenet. 2012 Jan 19;5(1):4.

PMID: 22260313 [PubMed - in process]
Free PMC Article

10- Identification of ten large deletions and one duplication in the F8 gene of eleven unrelated Iranian severe haemophilia A families using the multiplex ligation-dependent probe amplification technique.

Rafati M, Ravanbod S, Hoseini A, Rassoulzadegan M, Jazebi M, Enayat MS, Ala FA, Ghaffari SR.

Haemophilia. 2011 Jul;17(4):705-7. doi: 10.1111/j.1365-2516.2010.02476.x. Epub 2011 Mar 4. No abstract available.

PMID: 21371190 [PubMed - indexed for MEDLINE]

11- First Trimester Screening for Chromosomal Abnormalities by Integrated Application of Nuchal Translucency, Nasal Bone, Tricuspid Regurgitation, Ductus Venosus Flow Combined with Maternal Serum Free β-HCG and PAPP-A: A 5-Year Prospective Study.

Ghaffari SR, Tahmasebpour AR, Jamal A, Hantoushzadeh S, Eslamian L, Marsoosi V, Fattahi F, Rajaei M, Niroomanesh S, Borna S, Beigi A, Khazardoost S, Gargari SS, Sharbaf FR, Farrokhi B, Bayani N, Tehrani SE, Shahsavan K, Farzan S, Moossavi S, Ramezanzadeh F, Dastan J, **Rafati M**.

Ultrasound Obstet Gynecol. 2011 Jul 26:132. doi: 10.1002/uog.10051. [Epub ahead of print]

PMID: 21793085 [PubMed - as supplied by publisher]

12- Her2 amplification status in Iranian breast cancer patients: comparison of immunohistochemistry (IHC) and fluorescence in situ hybridisation (FISH).

Ghaffari SR, Sabokbar T, Dastan J, **Rafati M**, Moossavi S.

Asian Pac J Cancer Prev. 2011;12(4):1031-4.

PMID: 21790246 [PubMed - in process]

13- Tracing human papilloma virus in breast tumors of Iranian breast cancer patients.

Ghaffari SR, Sabokbar T, Meshkat Z, Fereidooni F, Dastan J, **Rafati M**, Zendehdel K.

Breast J. 2011 Mar-Apr;17(2):218-9. doi: 10.1111/j.1524-4741.2010.01053.x. Epub 2011 Jan 31. No abstract available.

PMID: 21276129 [PubMed - indexed for MEDLINE]

14- A novel truncating mutation in the E-cadherin gene in the first Iranian family with hereditary diffuse gastric cancer.

Ghaffari SR, **Rafati M**, Sabokbar T, Dastan J.

Eur J Surg Oncol. 2010 Jun;36(6):559-62. Epub 2010 May 14.

PMID: 20471195 [PubMed - indexed for MEDLINE]

15- Novel human pathological mutations. Gene symbol: CDH1. Disease: gastric cancer.

Ghaffari SR, Dastan J, **Rafati M**, Sabokbar T.

Hum Genet. 2009 Apr;125(3):337. No abstract available.

PMID: 19309801 [PubMed - indexed for MEDLINE]

16- Hematological Indices of Parents in Non-Immune Hydrops Fetalis Pregnancie

Saeed Reza Ghaffari, Farzaneh Larti, Tayebeh Sabokbar, **Maryam Rafati**, Jila Dastan, Laleh Eslamian, Fatemeh Rahimi, Sedighe Borna
Journal of Family and Reproductive Health, 2008 2(1), 33-36

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

BOOKS

1. **Genetics: Law, Ethics and Psychology, chapter 4: Human Enhancement Technology, Co-author**
   Avicenna Research Institute Publications. 2011 April

❖ **"Lecture Notes in Genetic Counseling" series**
"Ministry of Health and Medical Education", 2015
ISBN:978-600-5406-46-7
This series includes the below books:

2. **Lecture Notes in Genetic Counseling, Applied Principles, Co-author**
   Published Online: Official Website of "Ministry of Health and Medical Education", 2014

3. **Lecture Notes in Genetic Counseling, Ethical Issues, Co-author**
   "Ministry of Health and Medical Education", 2016
   ISBN: 978-600-5406-63-4

4. **Lecture Notes in Genetic Counseling, Legal Aspects, Co-author**
   "Ministry of Health and Medical Education", 201
   ISBN: 978-600-5406-53-5

5. **Lecture Notes in Genetic Counseling, Psychological Aspects, Co-author**
   "Ministry of Health and Medical Education", 2015
   ISBN:978-600-5406-57-3

6. **Lecture Notes in Genetic Counseling, Prenatal Screening of Common Chromosomal Aneuploidies, Co-author**
   "Ministry of Health and Medical Education", 2015
   ISBN: 978-600-5406-56-6

7. **Lecture Notes in Genetic Counseling, Intellectual Disability and Developmental Delay, Co-author**
   "Ministry of Health and Medical Education", 2015
   ISBN: 978-600-5406-59-7

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

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

10. **Lecture Notes in Genetic Counseling, Spinal Muscular Atrophy, Co-author**
    "Ministry of Health and Medical Education", 2015
    ISBN: 978-600-5406-58-0

11. **Lecture Notes in Genetic Counseling, Dystrophinopathies, Co-author**
    "Ministry of Health and Medical Education", 2015
    ISBN: 978-600-5406-54-2

12. **Lecture Notes in Genetic Counseling, Beta Thalassemia, Co-author**
    Published Online: Official Website of "Ministry of Health and Medical Education", 2014

13. **Lecture Notes in Genetic Counseling, Alpha Thalassemia, Co-author**
    Published Online: Official Website of "Ministry of Health and Medical Education", 2014

14. **Lecture Notes in Genetic Counseling, Hemophilia A, Co-author**
    Published Online: Official Website of "Ministry of Health and Medical Education", 2014

15. **Lecture Notes in Genetic Counseling, Phenylketonuria (PKU), Co-author**
    Published Online: Official Website of "Ministry of Health and Medical Education", 2014

16. **Lecture Notes in Genetic Counseling, Cystic Fibrosis, Co-author**
    Published Online: Official Website of "Ministry of Health and Medical Education", 2014

17. **Lecture Notes in Genetic Counseling, Autosomal Recessive Polycystic Kidney Disease (ARPKD), Co-author**
    Published Online: Official Website of "Ministry of Health and Medical Education", 2014

18. **Lecture Notes in Genetic Counseling, Autosomal Dominant Polycystic Kidney Disease (ADPKD), Co-author**
    Published Online: Official Website of "Ministry of Health and Medical Education", 2014

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

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

REGISTRATION of NEW MUTATIONS IN GenBank

Link to released novel mutations: http://www.ncbi.nlm.nih.gov/nuccore/?term=rafati+m

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	*

CITATIONS

Google Scholar Citations till June, 2016 (http://scholar.google.com)

Aggregate total of citations to my English papers: **146**

INVITED PRESENTATIONS

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- **Developments in Increasing the Pregnancy Rate of Assisted Reproductive Techniques using Preimplantation Genetic Screening using Next Generation Techniques (PGS-NGS): Iran Experience**
97th Seminar in Clinical Genetics, "a 1-year report on Iran Experience: Case Discussion", Tehran, Iran, 17 November, 2016

5- **Genetics of Endometriosis**

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

6- **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

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

8- **"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

9- **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"
Held by Ministry of Health and Medical Education, 2-3 March 2016

10- **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

11- 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

12- The Road from Next-generation Sequencing to Personalized Medicine: Technical Aspects

3rd Congress on Novel & Innovative Laboratory Technologies, Tehran, Iran, 6 October 2015

13- Genetic Counseling in Prenatal Screening of Aneuploidies

Conference on "Clinical and Laboratory Aspects of Prenatal Screening and Its Management"**,** Tehran, Iran, 26 November 2015

14- Expert panel on recurrent miscarriage: causes and recommended treatments

Conference on "recurrent miscarriage", Tehran, Iran, 12 November 2015

15- 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

16- **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

17- **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

18- **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

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

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

20- **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

21- **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

22- 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

23- Preimplantation Genetic Screening using Next Generation Techniques: PGS-NGS

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

24- Noninvasive Prenatal Testing (Cell-Free Fetal DNA)

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

25- Prenatal diagnosis of genetic disorders

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

26- Genetics in Congenital Disorders of Gastrointestinal and Urinary tract

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

27- 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

28- 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

29- Noninvasive Prenatal Testing (Cell-Free Fetal DNA)

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

30- ESHRE PGD Report

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

31- 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

32- Preimplantation genetic diagnosis, gender selection using FISH Technique

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

33- Preimplantation Genetic Diagnosis (PGD) of Single Gene Disorders

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

34- 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

35- 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

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

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

37- 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

38- 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

39- 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

40- 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

41- 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

42- First and Second Trimester Screening

Laboratory and Clinic Conference, "Prenatal Screening of Chromosome Abnormalities", Avicenna Biotechnology Research Institute, Tehran, Iran, 5 May 2013

43- 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

44- 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

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

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

46- Hereditary Thrombophilia in Recurrent Miscarriage

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

47- 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

48- Hereditary Breast and Ovarian Cancer

"Breast Cancer Seminar", Iranian Society of Pathology, 2010 May

49- Familial Mental Retardation Due to a Paternal Cryptic Subtelomeric Translocation: Partial Monosomy of 13qter and Partial Trisomy of 9pter, Detected by MLPA Technique

1st International and 5th Iranian Neurogenetics Congress, Lecturer, Tehran, Iran, 2011

50- Rapid prenatal diagnosis of common chromosomal abnormalities using MLPA technique

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

51- Assessment of common microdeletion and microduplication syndromes in Iranian patients with "Familial mental retardation".

1st International and 5th Iranian Neurogenetics Congress, Lecturer, Tehran, Iran, 2011

52- Genodermatoses

60th clinical genetic seminars, Imam Khomeini hospital, TUMS, 2009

POSTERS

1- Contribution of Subtelomeric Rearrangements in "Familial Mental Retardation": Investigation of 322 Patients from 102 Families

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

Invited to review the work of others

Board Member of Scientific Committees

1- Board Member of Scientific Committee of The 3rd National and the 1st International Congress on Endometriosis and Minimally Invasive Gynecology (EMIG), Tehran, Iran, 25-27 October 2016

2- Board Member of Scientific Committee of "The 2nd International Congress on Reproduction", ISERB, Tehran, Iran, May 2016

3- Board Member of Scientific Committee of "International Congress on Reproduction", ISERB, Tehran, Iran, May 2015

4- Board member of "Award of Excellence in the Field of Reproduction", Tehran, Iran, May 2015

5- Board Member of Scientific Committee of "2nd Iranian Medical Genetics Congress", Tehran, Iran, 2011

Review of Research Projects (2012-Present)

1- "Expression analysis of dopamine receptors (D2 family) in peripheral blood cells: comparing diabetic patients affected with diabetic foot with normal controls", Avicenna Biotechnology Research Institute, Tehran, Iran, 17 Nov 2015

2- "Genotyping of chlamydia trachomatis in semen samples", Avicenna Biotechnology Research Institute, Tehran, Iran, Mar 2012

3- "Investigation of AZFc microdeletion in patients with non-obstructive azoospermia", Avicenna Biotechnology Research Institute, Tehran, Iran, Mar 2012

4- "Assessment of the sensitivity and specificity of high resolution melting analysis technique in mutation analysis of RB1 gene", Avicenna Biotechnology Research Institute, Tehran, Iran, Aug 2013

5- "Association of EL polymorphisms with premature coronary artery disease", Academic Center for Education, Culture and Research (ACECR), Tehran, Iran, March 2014

6- "Expression analysis of AKAP3, PI2160 and PIWIL2 genes in breast cancer tissue in comparison with normal tissue", Academic Center for Education, Culture and Research (ACECR), Tehran, Iran, April 2014

7- "Targeted mutation analysis of BRCA1 in breast cancer patients followed by functional assay of detected mutations", Academic Center for Education, Culture and Research (ACECR), Tehran, Iran, Oct 2014

8- "Association of CYP2D6 polymorphisms with Tamoxifen resistance in breast cancer patients", Academic Center for Education, Culture and Research (ACECR), Tehran, Iran, Oct 2014

Review of Original Articles

1- Associate Editor, "Avicenna Journal of Medical Biotechnology"
2- Peer Reviewer, "Journal of Reproduction and Infertility"
3- Peer Reviewer, Journal of "Molecular Cytogenetics"
4- Peer Reviewer, "Journal of Pediatric Genetics"

WORKSHOPS

2015, 23-25 Nov
Scientific 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 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 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 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 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 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 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 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 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

MEDIA REPORTS

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

No.1: ISNA
http://www.isna.ir/news/95050109408/%D8%A8%D8%A7%D8%B1%D8%AF%D8%A7%D8%B1%DB%8C-%D8%AA%DA%A9-%D9%82%D9%84%D9%88%DB%8C%DB%8C-%D8%B2%D9%88%D8%AC-%D9%86%D8%A7%D9%82%D9%84-%D8%A8%DB%8C%D9%85%D8%A7%D8%B1%DB%8C-%DA%98%D9%86%D8%AA%DB%8C%DA%A9%DB%8C-%D8%AF%D8%B1-%D9%85%D8%B1%DA%A9%D8%B2-%D9%86%D8%A7%D8%A8%D8%A7%D8%B1%D9%88%D8%B1%DB%8C-%D8%A7%D8%A8%D9%86-%D8%B3%DB%8C%D9%86%D8%A7

No.2 Bashgah Khabarnegaran Javan
http://www.yjc.ir/fa/news/5723532/%D8%AA%D9%88%D9%84%D8%AF-%D8%A7%D9%88%D9%84%DB%8C%D9%86-%D9%86%D9%88%D8%B2%D8%A7%D8%AF-%D8%B3%D8%A7%D9%84%D9%85-%D8%AD%D8%A7%D8%B5%D9%84-%D8%A7%D8%B2-%D8%B1%D9%88%D8%B4-%D8%AA%D8%B4%D8%AE%DB%8C%D8%B5-%DA%98%D9%86%D8%AA%DB%8C%DA%A9%DB%8C-%D9%82%D8%A8%D9%84-%D8%A7%D8%B2-%D9%84%D8%A7%D9%86%D9%87%E2%80%8C%DA%AF%D8%B2%DB%8C%D9%86%DB%8C

No.3 Azad News Agency
http://www.ana.ir/news/129343

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

❖ 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

15 journal interviews

CLINICAL AND DIAGNOSTIC EXPERTISE

1- Genetic Counseling of More Than **8000** 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

- Next Generation Sequencing
    - Whole exome sequencing
    - Targeted capture sequencing (design gene sets)
- 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 Chromosomal Abnormalities
- Prenatal diagnosis of a wide range of single gene disorders
- MLPA
- STR analysis
- QF-PCR
- Real-time PCR
- Metaphase FISH
- Interphase FISH
- Basic Molecular Genetic Techniques (PCR, gel electrophoresis,…)
- Amplification of GC rich regions (triplet-primed PCR)
- Sanger Sequencing

COMPUTER SKILLS AND SOFTWARE

- Ion Reporter
- Torrent Suite, Proton Server
- Torrent Suite, PGM Server

- Mutation Surveyor
- Sequin
- Mutalyzer
- Chromas
- Progeny
- Coffalyser
- GeneMapper
- Genetic analyser data collection
- Microsoft office

RESEARCH INTERESTS

- Personalized Medicine
- Genomic Approach to Heterogeneous Disorders using Next Generation Sequencing
- Prenatal Diagnosis of genetic disorders
- Preimplantation Genetic Diagnosis
- Molecular Cytogenetics

REFERENCES

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