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### **Personal Data**

Born Stockholm Sweden  
Date of birth 23 Feb. 1964  
Marital Status Married

### **Education**

-Hadaf Elementary School Tehran Iran 1969-1970  
-E.B. Newton Elementary School Boston U.S.A. 1970-1972  
-Bahar Now Elementary School Tehran Iran 1972-1974  
-Iranzamin International School Tehran Iran 1974-1981  
-A.A. in Biomedical Technology Paramedical University Tehran Iran 1983-1986  
-Medical Student at Albert Szent-Gyorgyi Medical University Szeged, Hungary 1998-1999  
-M.D. Tehran Medical University, 1999-2006

### **Training Courses**

-Training in Chorionic Villi Sampling and Amniocentesis Genetics Center San Francisco U.S.A. 1985-1986  
-WHO sponsored course in traditional Chinese medicine Beijing China 1992  
-Attendance in 7<sup>th</sup> Course of European School of Medical Genetics Genova Italy 1994  
-Attendance in 8<sup>th</sup> Summer School of Myology organized by the Institute de Myologie, June 2005  
-Attendance in Hybrid Course in Genetic Counseling in Practice Nov. 2006  
-Attendance in Genetic Counseling Course in Behzisti 21/02/1391-02/04/1392, 10/05/2012-22/06/2012  
-Attendance in International Summer School "Rare Disease and orphan Drug Registries" 16-20 Sept. 2013, Rome, Italy  
-Attendance in "Advances in Fetal Medicine Course" 14-15 December 2013

### **Work Experience**

Cytogenetic Technician Massoud Laboratory Tehran Iran 1981-1985  
Supervisor of Cytogenetic Division of Prenatal diagnosis Pathology and Genetic Center Tehran Iran 1986-1998  
Supervisor of Clinical Genetics, Pathology and Genetic Center, Tehran Iran, 2006-present

### **Publications**

**Kariminejad** A, Schöls L, Schüle R, Tonekaboni SH, Abolhassani A, Fadaee M, Rosti RO, Gleeson JG. CYP2U1 mutations in two Iranian patients with activity induced dystonia, motor regression and spastic paraplegia. *Eur J Paediatr Neurol.* 2016 Jun 2.[Epub ahead of print]

**Kariminejad** A, Almadani N, Khoshaeen A, Olsson B, Moslemi AR, Tajsharghi H. Truncating CHRNG mutations associated with interfamilial variability of the severity of the Escobar variant of multiple pterygium syndrome. *BMC Genet.* 2016 May 31;17(1):71.

Fattahi Z, Kalhor Z, Fadaee M, Vazehan R, Parsimehr E, Abolhassani A, Beheshtian M, Zamani G, Nafissi S, Nilipour Y, Akbari MR, Kahrizi K, **Kariminejad** A, Najmabadi H. Improved diagnostic yield of neuromuscular disorders applying clinical exome sequencing in patients arising from a consanguineous population. *Clin Genet.* 2016 May 28 [Epub ahead of print]

Amos JS, Huang L, Thevenon J, **Kariminedjad A**, Beaulieu CL, Masurel-Paulet A, Najmabadi H, Fattahi Z, Beheshtian M, Tonekaboni SH, Tang S, Helbig KL, Alcaraz W, Rivière JB, Faivre L, Innes AM, Lebel RR, Boycott KM. Autosomal recessive mutations in THOC6 cause intellectual disability: Syndrome delineation requiring forward and reverse phenotyping. *Clin Genet*. 2016 Apr 22 [Epub ahead of print].

Fadaee M, **Kariminejad A**, Fattahi Z, Nafissi S, Godarzi HR, Beheshtian M, Vazehan R, Akbari MR, Kahrizi K, Najmabadi H. Report of limb girdle muscular dystrophy type 2a in 6 Iranian patients, one with a novel deletion in CAPN3 gene. *Neuromuscul Disord*. 2016;26(4-5):277-82.

Faraji Zonooz M, Sabbagh-Kermani F, Fattahi Z, Fadaee M, Akbari MR, Amiri R, Vahidnezhad H, Uitto J, Najmabadi H, **Kariminejad A**. Whole Genome Linkage Analysis Followed by Whole Exome Sequencing Identifies Nicastrin (NCSTN) as a Causative Gene in a Multiplex Family with  $\gamma$ -Secretase Spectrum of Autoinflammatory Skin Phenotypes. *J Invest Dermatol*. 2016 ;136(6):1283-6.

Bonafé L, **Kariminejad A**, Li J, Royer-Bertrand B, Garcia V, Mahdavi S, Bozorgmehr B, Lachman RL, Mittaz-Crettol L, Campos-Xavier B, Nampoothiri S, Unger S, Rivolta C, Levade T, Superti-Furga A. Peripheral osteolysis in adults linked to ASAHI (acid ceramidase) mutations: A new presentation of Farber disease. *Arthritis Rheumatol*. 2016; Mar 4 [Epub ahead of print]

**Kariminejad A**, Ghaderi-Sohi S, Hossein-Nejad Nedai H, Varasteh V, Moslemi AR, Tajsharghi H. Lethal multiple pterygium syndrome, the extreme end of the RYR1 spectrum. *BMC Musculoskelet Disord*. 2016 ;17:109.

Tafakhori A, Yu Jin Ng A, Tohari S, Venkatesh B, Lee H, Eskin A, Nelson SF, Bonnard C, Reversade B, **Kariminejad A**. Mutation in TWINKLE in a Large Iranian Family with Progressive External Ophthalmoplegia, Myopathy, Dysphagia and Dysphonia, and Behavior Change. *Arch Iran Med*. 2016;19(2):87-91.

Todd EJ, Yau KS, Ong R, Slee J, McGillivray G, Barnett CP, Haliloglu G, Talim B, Akcoren Z, **Kariminejad A**, Cairns A, Clarke NF, Freckmann ML, Romero NB, Williams D, Sewry CA, Colley A, Ryan MM, Kiraly-Borri C, Sivadorai P, Allcock RJ, Beeson D, Maxwell S, Davis MR, Laing NG, Ravenscroft G. Next generation sequencing in a large cohort of patients presenting with neuromuscular disease before or at birth. *Orphanet J Rare Dis*. 2015;17:10:148.

Barzegar M, Asadi-Kani Z, Mozafari N, Vahidnezhad H, **Kariminejad A**, Toossi P. Using immunofluorescence (antigen) mapping in the diagnosis and classification of epidermolysis bullosa: a first report from Iran. *Int J Dermatol*. 2015;54:10:e416-23.



**Kariminejad A**, Nafissi S, Nilipoor Y, Tavasoli A, Van Veldhoven PP, Bonnard C, Ng YT, Majoie CB, Reversade B, Hennekam RC. Intellectual disability, muscle weakness and characteristic face in three siblings: A newly described recessive syndrome mapping to 3p24.3-p25.3. *Am J Med Genet A*. 2015;167A:11:2508-15.

Deml B, Reis LM, Lemyre E, Clark RD, **Kariminejad A**, Semina EV. Novel mutations in PAX6, OTX2 and NDP in anophthalmia, microphthalmia and coloboma. *Eur J Hum Genet*. 2016;24:4:535-41

Marchegiani S, Davis T, Tessadori F, van Haaften G, Brancati F, Hoischen A, Huang H, Valkanas E, Pusey B, Schanze D, Venselaar H, Vulto-van Silfhout AT, Wolfe LA, Tiftt CJ, Zervas PM, Zambruno G, **Kariminejad A**, Sabbagh-Kermani F, Lee J, Tsokos MG, Lee CC, Ferraz V, da Silva EM, Stevens CA, Roche N, Bartsch O, Farndon P, Bermejo-Sanchez E, Brooks BP, Maduro V, Dallapiccola B, Ramos FJ, Chung HY, Le Caignec C, Martins F, Jacyk WK, Mazzanti L, Brunner HG, Bakkers J, Lin S, Malicdan MC, Boerkoel CF, Gahl WA, de Vries BB, van Haelst MM, Zenker M, Markello TC. Recurrent Mutations in the

Basic Domain of TWIST2 Cause Ablepharon Macrostomia and Barber-Say Syndromes. *Am J Hum Genet.* 2015 Jul 2;97(1):99-110.

Norgett EE, Yii A, Blake-Palmer KG, Sharifian M, Allen LE, Najafi A, **Kariminejad A**, Karet Frankl FE. A role for VAX2 in correct retinal function revealed by a novel genomic deletion at 2p13.3 causing distal Renal Tubular Acidosis: case report. *BMC Med Genet.* 2015 ;13;16:38

Rostami P, Valizadegan S, Ghalandary M, Mehrjouy MM, Esmail-Nia G, Khalili S, Shahmoradi SS, Imanian H, Hadavi V, Ghaderi-Sohi S, Almadani N, Afroozan F, **Kariminejad A**, Kariminejad R, Najmabadi H. Prenatal Screening for Aneuploidies Using QF-PCR and Karyotyping: A Comprehensive Study in Iranian Population. *Arch Iran Med.* 2015;18(5):296-303.

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Deml B, **Kariminejad A**, Borujerdi RH, Muheisen S, Reis LM, Semina EV. Mutations in MAB21L2 result in ocular Coloboma, microcornea and cataracts. *PLoS Genet.* 2015 ;11(2)e1005002

Fattahi Z, Kahrizi K, Nafissi S, Fadaee M, Abedini SS, **Kariminejad A**, Akbari MR, Najmabadi H. Report of a patient with limb-girdle muscular dystrophy, ptosis and ophthalmoparesis caused by plectinopathy. *Arch Iran Med.* 2015;18:1:60-4.

**Kariminejad A**, Rajaei A, Ashrafi MR, Alizadeh H, Tonekaboni SH, Malamiri RA, Ghofrani M, Karimzadeh P, Mohammadi MM, Baghalshoostari A, Bozorgmehr B, Kariminejad MH, Postma N, Abbink TE, van der Knaap MS. Eight novel mutations in MLC1 from 18 Iranian patients with megalencephalic leukoencephalopathy with subcortical cysts. *Eur J Med Genet.* 2015;58:2:71-4

Martin CA, Ahmad I, Klingseisen A, Hussain MS, Bicknell LS, Leitch A, Nürnberg G, Toliat MR, Murray JE, Hunt D, Khan F, Ali Z, Tinschert S, Ding J, Keith C, Harley ME, Heyn P, Müller R, Hoffmann I, Daire VC, Dollfus H, Dupuis L, Bashamboo A, McElreavey K, **Kariminejad A**, Mendoza-Londono R, Moore AT, Saggari A, Schlechter C, Weleber R, Thiele H, Altmüller J, Höhne W, Hurler ME, Noegel AA, Baig SM, Nürnberg P, Jackson AP. Mutations in PLK4, encoding a master regulator of centriole biogenesis, cause microcephaly, growth failure and retinopathy. *Nat Genet.* 2014;46:12:1283-92.

Campeau PM, Hennekam. DOORS syndrome: phenotype, genotype and comparison with Coffin-Siris syndrome. RC; DOORS syndrome collaborative group. *Am J Med Genet C Semin Med Genet.* 2014;166C:3:327-32

Acuna-Hidalgo R, Schanze D, **Kariminejad A**, Nordgren A, Kariminejad MH, Conner P, Grigelioniene G, Nilsson D, Nordenskjöld M, Wedell A, Freyer C, Wredenberg A, Wiczorek D, Gillessen-Kaesbach G, Kayserili H, Elcioglu N, Ghaderi-Sohi S, Goodarzi P, Setayesh H, van de Vorst M, Steehouwer M, Pfundt R, Krabichler B, Curry C, MacKenzie MG, Boycott KM, Gilissen C, Janecke AR, Hoischen A, Zenker M. Neulaxova syndrome is a heterogeneous metabolic disorder caused by defects in enzymes of the L-serine biosynthesis pathway. *Am J Hum Genet.* 2014;95:3:285-93.

Loghmani Khouzani H, **Kariminejad A**, Zamani G, Ghalandary M, Bozorgmehr B, Amirsalari S, Mojahedi F, Tonekaboni SH, Kariminejad R, Najmabadi H. Investigation of Microdeletions in Syndromic Intellectual Disability by MLPA in Iranian Population. *Arch Iran Med.* 2014; 17:7:471-4.

Fattahi Z, Rostami P, Najmabadi A, Mohseni M, Kahrizi K, Akbari MR, **Kariminejad A**, Najmabadi H. Mutation profile of BBS genes in Iranian patients with Bardet-Biedl syndrome: genetic characterization and report of nine novel mutations in five BBS genes. *J Hum Genet.* 2014;59:7:368-75.

**Kariminejad A**, Bozorgmehr B, Najafi A, Khoshaeen A, Ghalandari M, Najmabadi H, Kariminejad MH, Vanakker OM, Hosen, Fransiska Malfait MJ, Quaglino D, Florijn RJ, Bergen AAB, Hennekam RC. Retinitis Pigmentosa, Cutis Laxa and Pseudoxanthoma Elasticum-Like Skin Manifestations Associated with GGCX Mutations. *J Invest Dermatol.* 2014;134:9:2331-8.

**Kariminejad A**, Barzegar M, Abdollahimajd F, Pramanik R, McGrath J.A. Olmsted syndrome in an Iranian boy with a new de novo mutation in TRPV3. *Clin Exp Dermatol*. 2014; 39(4):492-5

**Kariminejad A**, Bozorgmehr B, Alizadeh H, Ghaderi-Sohi S, Toksoy G, Oya Uyguner Z, Kayserili H. Skull defects, alopecia, hypertelorism, and notched alae nasi caused by homozygous ALX4 gene mutation. *Am J Med Genet A*. 2014 May; 164A:5:1322-7.

Novarino G, Fenstermaker AG, Zaki MS, Hofree M, Silhavy JL, Heiberg AD, Abdellateef M, Rosti B, Scott E, Mansour L, Masri A, Kayserili H, Al-Aama JY, Abdel-Salam GM, **Kariminejad A**, Kara M, Kara B, Bozorgmehr B, Ben-Omran T, Mojahedi F, Mahmoud IG, Bouslam N, Bouhouche A, Benomar A, Hanein S, Raymond L, Forlani S, Mascaro M, Selim L, Shehata N, Al-Allawi N, Bindu PS, Azam M, Gunel M, Caglayan A, Bilguvar K, Tolun A, Issa MY, Schroth J, Spencer EG, Rosti RO, Akizu N, Vaux KK, Johansen A, Koh AA, Megahed H, Durr A, Brice A, Stevanin G, Gabriel SB, Ideker T, Gleeson JG. Exome sequencing links corticospinal motor neuron disease to common neurodegenerative disorders. *Science*. 2014 Jan; 343(6170):506-11.

Hafezi-Nejad, Mohsen Khosravi N, Bayat N, **Kariminejad A**, Hadavi V, Oberkanins C, Azarkeivan A, Najmabadi H. Characterizing a Cohort of  $\alpha$ -Thalassemia Couples Collected During Screening for Hemoglobinopathies: 14 Years of an Iranian Experience. *Hemoglobin*. 2014; 38:3:153-7.

Campeau PM, Kasperaviciute D, Lu JT, Burrage LC, Kim C, Hori M, Powell BR, Stewart F, Félix TM, van den Ende J, Wisniewska M, Kayserili H, Rump P, Nampoothiri S, Aftimos S, Mey A, Nair LD, Begleiter ML, De Bie I, Meenakshi G, Murray ML, Repetto GM, Golabi M, Blair E, Male A, Giuliano F, **Kariminejad A**, Newman WG, Bhaskar SS, Dickerson JE, Kerr B, Banka S, Giltay JC, Wieczorek D, Tostevin A, Wisniewska J, Cheung SW, Hennekam RC, Gibbs RA, Lee BH, Sisodiya SM. The genetic basis of DOORS syndrome: an exome-sequencing study. *Lancet Neurol*. 2014; 13:1:44-58.

Gardeitchik T, Mohamed M, Fischer B, Lammens M, Lefeber D, Lace B, Parker M, Kim KJ, Lim BC, Häberle J, Garavelli L, Jagadeesh S, **Kariminejad A**, Guerra D, Leão M, Keski-Filppula R, Brunner H, Nijtmans L, van den Heuvel B, Wevers R, Kornak U, Morava E. Clinical and biochemical features guiding the diagnostics in neurometabolic cutis laxa. *Eur J Hum Genet*. 2014;22:7:888-95.

Barzegar M, Mozafari N, **Kariminejad A**, Asadikani Z, Ozoemena L, McGrath JA. A new homozygous nonsense mutation in LAMA3A underlying laryngo-onycho-cutaneous syndrome. *Br J Dermatol*. 2013; 169:6:1353-6.

Bozorgmehr B, **Kariminejad A**, Nafissi S, Jebelli B, Andoni U, Gartiaux C, Ledeuil C, Allamand V, Richard P, Kariminejad MH. Ullrich Congenital Muscular Dystrophy (UCMD): Clinical and Genetic Correlations. *Iran J Child Neurol*. 2013; 7(3):15-22.

Malfait F, **Kariminejad A**, Van Damme T, Gauche C, Syx D, Merhi-Soussi F, Gulberti S, Symoens S, Vanhauwaert S, Willaert A, Bozorgmehr B, Kariminejad MH, Ebrahimiadib N, Hausser I, Huisseune A, Fournel-Gigleux S, De Paepe A. Defective initiation of glycosaminoglycan synthesis due to B3GALT6 mutations causes a pleiotropic Ehlers-Danlos-syndrome-like connective tissue disorder. *Am J Hum Genet*. 2013; 92:6:935-45.

**Kariminejad A**, Stollfuß B, Li Y, Bögershausen N, Boss K, Hennekam RC, Wollnik B. Severe Cenani-Lenz syndrome caused by loss of LRP4 function. *Am J Med Genet A*. 2013; 161A:6:1475-9.

**Kariminejad A**, Hennekam RC. Aponia, microstomia, deafness, retinal dystrophy, duplicated halluces and intellectual disability. *Am J Med Genet A*. 2012; 158A:11:2756-62.

**Kariminejad A**, Radmanesh F, Rezayi AR, Tonekaboni SH, Gleeson JG. Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome: a case report. *J Child Neurol*. 2013; 28:5:651-7.

Chassaing N, Ragge N, **Kariminejad A**, Buffet A, Ghaderi-Sohi S, Martinovic J, Calvas P. Mutation analysis of the STRA6 gene in isolated and non-isolated anophthalmia/microphthalmia. *Clin Genet*. 2013; 83:3:244-50.

- Sansbury FH, Flanagan SE, Houghton JA, Shuixian Shen FL, Al-Senani AM, Habeb AM, Abdullah M, **Kariminejad A**, Ellard S, Hattersley AT. SLC2A2 mutations can cause neonatal diabetes, suggesting GLUT2 may have a role in human insulin secretion. *Diabetologia*. 2012; 559:2381-5.
- Van Dijk FS, Cobben JM, **Kariminejad A**, Maugeri A, Nikkels PG, van Rijn RR, Pals G. Osteogenesis Imperfecta: A Review with Clinical Examples. *Mol Syndromol*. 2011; 21:1-20.
- Vogel MJ, van Zon P, Brueton L, Gijzen M, van Tuil MC, Cox P, Schanze D, **Kariminejad A**, Ghaderi-Sohi S, Blair E, Zenker M, Scambler PJ, Ploos van Amstel HK, van Haelst MM. Mutations in GRIP1 cause Fraser syndrome. *J Med Genet*. 2012; 49:5:303-6.
- Kariminejad A**, Ghaderi-Sohi S, Kariminejad MH, Lachman R. An/micr-ophthalmia, cleft lip/palate, and short limbs: a new syndrome simulating a short rib syndrome. *Fetal Pediatr Pathol*. 2012; 31:5:295-9.
- Guergueltcheva V, Müller JS, Dusl M, Senderek J, Oldfors A, Lindbergh C, Maxwell S, Colomer J, Mallebrera CJ, Nascimento A, Vilchez JJ, Muelas N, Kirschner J, Nafissi S, **Kariminejad A**, Nilipour Y, Bozorgmehr B, Najmabadi H, Rodolico C, Sieb JP, Schlotter B, Schoser B, Herrmann R, Voit T, Steinlein OK, Najafi A, Urtizbera A, Soler DM, Muntoni F, Hanna MG, Chaouch A, Straub V, Bushby K, Palace J, Beeson D, Abicht A, Lochmüller H. Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. *J Neurol*. 2012;259:5:838-50
- Rohrbach M, Vandersteen A, Yiş U, Serdaroglu G, Ataman E, Chopra M, Garcia S, Jones K, **Kariminejad A**, Kraenzlin M, Marcelis C, Baumgartner M, Giunta C. Phenotypic variability of the kyphoscoliotic type of Ehlers-Danlos syndrome (EDS VIA): clinical, molecular and biochemical delineation. *Orphanet J Rare Dis*. 2011;23; 6:46.
- Kretz R, Bozorgmehr B, Kariminejad MH, Rohrbach M, Hausser I, Baumer A, Baumgartner M, Giunta C, **Kariminejad A**, Häberle J. Defect in proline synthesis: pyrroline-5-carboxylate reductase 1 deficiency leads to a complex clinical phenotype with collagen and elastin abnormalities. *J Inher Metab Dis*. 2011;34:3:731-9
- Kariminejad A**, Kariminejad R, Moshtagh A, Zanganeh M, Kariminejad MH, Neuenschwander S, Okoniewski M, Wey E, Schinzel A, Baumer A. Pericentric inversion of chromosome 18 in parents leading to a phenotypically normal child with segmental uniparental disomy 18. *Eur J Hum Genet*. 2011; 19(5):555-60.
- Senderek J, Müller JS, Dusl M, Strom TM, Guergueltcheva V, Diepolder I, Laval SH, Maxwell S, Cossins J, Krause S, Muelas N, Vilchez JJ, Colomer J, Mallebrera CJ, Nascimento A, Nafissi S, **Kariminejad A**, Nilipour Y, Bozorgmehr B, Najmabadi H, Rodolico C, Sieb JP, Steinlein OK, Schlotter B, Schoser B, Kirschner J, Herrmann R, Voit T, Oldfors A, Lindbergh C, Urtizbera A, von der Hagen M, Hübner A, Palace J, Bushby K, Straub V, Beeson D, Abicht A, Lochmüller H. Hexosamine biosynthetic pathway mutations cause neuromuscular transmission defect. *Am J Hum Genet*. 2011;11: 88(2):162-72.
- Van Dijk FS, Huizer M, **Kariminejad A**, Marcelis CL, Plomp AS, Terhal PA, Meijers-Heijboer H, Weiss MM, van Rijn RR, Cobben JM, Pals G. Complete COL1A1 allele deletions in osteogenesis imperfecta. *Genet Med*. 2010; 12:11:736-41.
- Kariminejad A**, Kariminejad R, Tzschach A, Najafi H, Ahmed A, Ullmann R, Ropers HH, Kariminejad MH. 11q14.1-11q22.1 deletion in a 1-year-old male with minor dysmorphic features. *Am J Med Genet A*. 2010; 152A:10:2651-5.
- Sharifian M, Esfandiar N, Mazaheri S, **Kariminejad A**, Mohkam M, Dalirani R, Esmaili R, Ahmadi M, Hassas-Yeganeh M. Distal Renal Tubular Acidosis and Its Relationship with Hearing Loss in Children. *Iran J Kidney Dis*. 2010; 4:202-206.
- Matejas V, Hinkes B, Alkandari F, Al-Gazali L, Annexstad E, Aytac MB, Barrow M, Bláhová K, Bockenbauer D, Cheong HI, Maruniak-Chudek I, Cochat P, Dötsch J, Gajjar P, Hennekam RC, Janssen F, Kagan M, **Kariminejad A**, Kemper MJ, Koenig J, Kogan J, Kroes HY, Kuwertz-Bröking E, Lewanda AF, Medeira A, Muscheites J, Niaudet P, Pierson M, Saggat A, Seaver L, Suri M, Tsygin A, Wühl E, Zurowska A, Uebe S, Hildebrandt F, Antignac C, Zenker M. Mutations in the Human Laminin beta2 (LAMB2) gene and the Associated Phenotypic Spectrum. *Hum Mutat*. 2010; 9:992-1002.

**Kariminejad A**, Bozorgmehr B, Khatami A, Kariminejad MH, Giunta C, Steinmann B. Ehlers-Danlos syndrome (EDS VI), Kyphoscoliotic type; a differential diagnosis to be considered in hypotonic newborns, Iranian J Pediatr. 2010; 20:261-268.

Karimi A, Peiravian F, Amirghofran AA, **Kariminejad A**. Absent pulmonary valve, intact interventricular septum, rudimentary aortic non-coronary cusp and ascending aortic aneurysm in a single patient. Interact Cardiovasc Thorac Surg. 2010; 4:636-8.

**Kariminejad A**, Goodarzi P, Asghari Roodsari A, Kariminejad MH. Amelia. Cleft Lip and Holoprosencephaly: A Distinct Entity. Am J Med Genet. 2010;149A:2828-2831.

**Kariminejad A**, Kariminejad R, Tzschach A, Ullmann R, Ahmed A, Asghari Roodsari A, Salehpour S, Afroozan F, Ropers H, Kariminejad MH. Craniosynostosis in a patient with 2q37.3 Deletion 5q34 Duplication. Association of Extra Copy of MSX2 with Craniosynostosis. American Journal of Medical Genetics Part A. 2009; 149:1544-1549.

Ashrafi MH, **Kariminejad A**, Alizadeh H, Bozorgmehr B, Amoeian S, Kariminejad MH. A case of Megalencephalic Leukoencephalopathy with Subcortical Cysts in an Iranian consanguineous Family: Molecular Genetic Study. Iran J Pediatr. 2009; 19:425-429.

**Kariminejad A**, Goodarzi P, Thanh Huong LT, Wehnert MS. Restrictive dermopathy: Molecular diagnosis of restrictive dermopathy in a stillborn fetus from a consanguineous Iranian family. Saudi Medical Journal. 2009; 30:150-153.

Elting M, **Kariminejad A**, Sonnaville ML, Ottenkamp J, Bauhuber S, Bozorgmehr B, Zenker M, Cobben JM. Johanson-Blizzard Syndrome caused by Identical UBR1 Mutations in Two Unrelated Girls, One with a Cardiomyopathy. Am J Med Genet. 2008; 146A: 3058-3061.

**Kariminejad A**, Bozorgmehr B, Ashrafi MR, Kariminejad MH. Skull defects, alopecia and distinctive facies: a new syndrome? Clinical Dysmorphology. 2008; 17: 203-205.

**Kariminejad A**, Bozorgmehr B, Sedighi Gilani MA, Almadani N, Kariminejad MH. Clinical Variability in Acro-Cardio-Facial-Syndrome. Am J Med Genet. 2008; 146A: 1977-1979.

**Kariminejad A**, Kariminejad MH, Lashgarian N, Results of cytogenetics analysis of 521 amniotic fluid cell cultures (amniocenteses) performed in Iran. Medical Journal of Islamic Republic of Iran. 1999; 13: 3; 161-166.

**Kariminejad A**, Shafeghati Y, Kariminejad R, Supernumerary Nipples in a Bartsocas-Papas Patient in a consanguineous Iranian family. Clinical Dysmorphology. 1998; 8: 155-156:.

**Kariminejad A**, Shafeghati Y, Kariminejad R, Nabavi-Nia N, Kodoma K, Kariminejad MH. New Findings in a Patient with distal 13q-. Clinical Dysmorphology. 1997; 7: 153-154:.

**Kariminejad A**, Kariminejad R, Najafi H, Kariminejad MH. Blepharophimosis Syndrome (BPES) and additional abnormalities in a female with a balanced X:3 translocation, Clinical Dysmorphology. 1996; 5: 259-261.

**Kariminejad A**, Nilforoushan MA, Shafeghati Y, Kariminejad MH. Baller-Gerold Syndrome. A case of craniosynostosis and unilateral radial aclasis, and review of literature. Iranian Journal of Medical Sciences. 1994; 19: 1; 64-68.