

## Publication du laboratoire de Génétique Humaine

1. Charoute H, Bakhchane A, Benrahma H, Romdhane L, Gabi K, Rouba H, Fakiri M, Abdelhak S, Lenaers G, Barakat A. mediterranean fonder mutation database (MFMD): taking advantage from fonder mutations in genetics diagnosis, genetic diversity and migration history of the mediterranean Population. *Hum Mutat.* 2015 Jul.
2. Bakhchane A, Charif M, Salime S, Boulouiz R, Nahili H, Roky R, Lenaers G, Barakat A. Recessive TBC1D24 mutations are frequent in Moroccan non-syndromic hearing loss pedigrees. *PLoS One.* 2015 Sep 15;10(9)
3. Naasse Y, Charoute H, El Houate B, Elbekkay C, Razoki L, Malki A, Barakat A, Rouba H. Chromosomal abnormalities and Y chromosome microdeletions in infertile men from Morocco. *BMC Urol.* 2015 Sep 18;15(1):95
4. Lakbakbi El Yaagoubi F, Charoute H, Bakhchane A, Ajjemami M, Benrahma H, Errouagui A, Kandil M, Rouba H, Barakat A. Association analysis of APOA5 rs662799 and rs3135506 polymorphisms with obesity in Moroccan patients. *Pathol Biol.* . 2015 Oct 29
5. Ajjemami M, Ouatou S, Charoute H, Fakiri M, Rhaissi H, Benrahma H, Rouba H, Barakat A. Haplotype analysis of the Apolipoprotein A5 gene in Moroccan patients with the metabolic syndrome (2015). *J Diabetes Metab Disord.* Apr 16;14:29.
6. El Khachibi M, Diakite B, Hamzi K, Badou A, Senhaji MA, Bakhchane A, Jouhadi H, Barakat A, Benider A, Nadifi S. Screening of exon 11 of BRCA1 gene using the high resolution melting approach for diagnosis in Moroccan cancer patients (2015) . *BMC Cancer.* Feb 25;15:81.
7. Eloualid A, Rouba H, Rhaissi H, Barakat A, Louanjli N, Bashamboo A, McElreavey K. Prevalence of the aurora kinase C c.144delC mutation in infertile Moroccan men (2014). *Fertil Steril.* Apr;101(4):1086-90
8. Benrahma H, Charoute H, Lasram K, Boulouiz R, Atig RK, Fakiri M, Rouba H, Abdelhak S, Barakat A. Association analysis of IGF2BP2, KCNJ11 and CDKL1 polymorphisms with type 2 diabetes mellitus in moroccan population: a case-control study and meta-analysis (2014). *Biochem Genet.* Oct;52(9-10):430-42.
9. Sefri H, Benrahma H, Charoute H, Lakbakbi el Yaagoubi F, Rouba H, Lyoussi B, Nourlil J, Abidi O, Barakat A. TNF A-308G>A polymorphism in Moroccan patients with type 2 diabetes mellitus: a case-control study and meta-analysis (2014) . *Mol Biol Rep.* Sep;41(9):5805-11.
10. Lasram K, Ben Halim N, Hsouna S, Kefi R, Arfa I, Ghazouani W, Jamoussi H, Benrahma H, Kharrat N, Rebai A, Ben Ammar S, Bahri S, Barakat A, Abid A, Abdelhak S. Evidence for association of the E23K variant of KCNJ11 gene with type 2 diabetes in tunisian population: population-based study and meta-analysis (2014). *Biomed Res Int.* ;2014:265-274.
11. Hsouna S, Ben Halim N, Lasram K, Meiloud G, Arfa I, Kerkeni E, Romdhane L, Jamoussi H, Bahri S, Ben Ammar S, Abid A, Barakat A, Houmeida A, Abdelhak S, Kefi R. Study of the T16189C variant and mitochondrial lineages in tunisian and overall mediterranean region (2014). *Mitochondrial DNA.* 2014 Sep 10:1-6.
12. Hicham Charoute, Halima Nahili, Omar Abidi, Khalid Gabi, Hassan Rouba, Malika Fakiri and Abdelhamid Barakat,. The Moroccan Genetic Disease Database (MGDD): a database for DNA variations related to inherited disorders and disease susceptibility (2014). *European Journal of Human Genetics* 22, 322–326.
13. Ouatou S, Ajjemami M, Charoute H, Sefri H, Ghalim N, Rhaissi H, Benrahma H, Barakat A, Rouba H. Association of APOA5 rs662799 and rs3135506 polymorphisms with arteriel hypertension in Moroccan patients (2014). *Lipids Health Dis.* Apr 1;13:60.
14. Lasram K, Ben Halim N, Benrahma H, Mediene-Benchekor S, Arfa I, Hsouna S, Kefi R, Jamoussi H, Ben Ammar S, Bahri S, Abid A, Benhamamouch S, Barakat A, Abdelhak S. contribution of CDKL rs7756992 and IGF2BP2 re4402960 polymorphisms in type 2 diabetes

- complications, obesity risk and hypertension in the Tunisian population (2014). *J Diabetes*. Jan;7(1):102-13.
15. Jeddane L, Ailal F, Dubois-d'Enghien C, Abidi O, Benhsaien I, Kili A, Chaouki S, Kriouile Y, El Hafidi N, Fadil H, Abilkassem R, Rada N, Bousfiha AA, Barakat A, Stoppa-Lyonnet D, Bellaoui H (2013). Molecular defects in Moroccan patients with ataxia-telangiectasia (2013). *Neuromolecular Med*. Jun;15(2):288-94.
16. Charif M, Bakhechane A, Abidi O, Boulouiz R, Eloualid A, Roky R, Rouba H, Kandil M, Lenaers G, Barakat A. Analysis of CLDN14 gene in deaf Moroccan patients with non-syndromic hearing loss. *Gene* (2013). Jul 1;523(1):103-5.
17. Charif M, Boulouiz R, Bakhechane A, Benrahma H, Nahili H, Eloualid A, Rouba H, Kandil M, Abidi O, Lenaers G, Barakat A. Genetic and molecular analysis of the CLDN14 gene in Moroccan family with nonsyndromic hearing loss (2013). *Indian J Hum Genet*;19:331-6
18. Benrahma H, Abidi O, Melouk L, Ajjemami M, Rouba H, Chadli A, Oudghiri M, Farouqui A, Barakat A. Association of the C677T polymorphism in the human methylenetetrahydrofolate Reductase (MTHFR) gene with the genetic predisposition for type 2 diabetes mellitus in a Moroccan population (2012). *Genet Test Mol Biomarkers* 16:383-7
19. Charif M, Abidi O, Boulouiz R, Nahili H, Rouba H, Kandil M, Delprat B, Lenaers G, Barakat A. Molecular analysis of the TMPRSS3 gene in Moroccan families with non-syndromic hearing loss (2012). *Biochem Biophys Res Commun*. 419: 643-7.
20. Eloualid A, Abidi O, Charif M, El Houate B, Benrahma H, Louanjli N, Chadli E, Ajjemami M, Barakat A, Bashamboo A, McElreavey K, Rhaiissi H, Rouba H. Association of the MTHFR A1298C variant with unexplained severe male infertility (2012). *PLoS One*. 7:e34111.
21. Hilmani S, Abidi O, Benrahma H, Karkouri M, Sahraoui S, El Azhari A, Barakat A. Clinicopathological Features and Molecular Analysis of Primary Glioblastomas in Moroccan Patients (2012). *J Mol Neurosci*. Aug 4.
22. Eloualid A, Rhaiissi H, Reguig A, Bounaceur S, El Houate B, Abidi O, Charif M, Louanjli N, Chadli E, Barakat A, Bashamboo A, McElreavey K, Rouba H. Association of spermatogenic failure with the b2/b3 partial AZFc deletion (2012). *PLoS One* 7:e34902.
23. Majida Charif, Safaa Bounaceur, Omar Abidi , Halima Nahili, Hassan Rouba, Mostafa Kandil, Redouane Boulouiz, Abdelhamid Barakat. The c.242G>A mutation in LRTOMT gene is responsible for a high prevalence of deafness in the Moroccan population (2012). *Mol Biol Rep*. Dec;39 (12):11011-6
24. Von Ameln S, Wang G, Boulouiz R, Rutherford MA, Smith GM, Li Y, Pogoda HM, Nürnberg G, Stiller B, Volk AE, Borck G, Hong JS, Goodeyear RJ, Abidi O, Nürnberg P, Hofmann K, Richardson GP, Hammerschmidt M, Moser T, Wollnik B, Koehler CM, Teitel MA, Barakat A, Kubisch C. A Mutation in PNPT1, Encoding Mitochondrial-RNA-Import Protein PNPase, Causes Hereditary Hearing Loss. *Am.J.Hum.Genet* (2012). Nov2;91(5):919-17.
25. Senhaji MA, Abidi O, Nadifi S, Benchikhi H, Khadir K, Ben Rekaya M, Eloualid A, Messaoud O, Abdelhak S, Barakat A. c.1643\_1644delTG XPC mutation is more frequent in Moroccan patients with xeroderma pigmentosum (2012). *Arch Dermatol Res*. 2013 Jan;305(1):53-7.
26. Benrahma H, Arfa I, Charif M, Bounaceur S, Eloualid A, Boulouiz R, Nahili H, Abidi O, Rouba H, Chadli A, Oudghiri M, Farouqui A, Abdelhak S, Barakat A. Maternal effect and familial aggregation in a type 2 diabetic moroccan population (2011). *J Community Health* 36:943-8.
27. Abidi O, Knari S, Sefri H, Charif M, Senechal A, Hamel C, Rouba H, Zaghloul K, El Kettani A, Lenaers G, Barakat A. mutational analysis of the RB gene in Moroccan patients with retinoblastoma (2011). *Mol Vis* 17:3541-7.
28. Li Y, Pohl E, Boulouiz R, Schraders M, Nürnberg G, Charif M, Admiraal RJ, Von Ameln S, Baessmann I, Kandil M, Veltman JA, Nürnberg P, Kubish C, Barakat A, Kremer H, Wollnik B. Mutations in TPRN cause a progressive form of autosomal-recessive nonsyndromic hearing loss (2010). *Am J Hum Genet* Mar 86: 479-84.
29. Naamane H, El Maataoui O, Ailal F, Barakat A, Bennani S, Najib J, Hassar M, Saile R, Bousfiha AA. The

- 752delG26 mutation in the RFXANK gene associated with major histocompatibility complex class II deficiency: evidence for a fonder effect in the moroccan population (2010). Eur J Pediatr 169:1069-74.
30. Nahili H, Charif M, Boulouiz R, Bounaceur S, Benrahma H, Abidi O, Chafik A, Rouba H, Kandil M, Barakat A. Prevalence of the mitochondrial A1555G mutation in Moroccan patients with non-syndromic hearing loss (2010). Int J Pediatr Otorhinolaryngol 74 :1071-4.
31. Imken L, Rouba H, El Houate B, Louanjli N, Barakat A, Chafik A, McElreavey K. Mutations of the protamine locus: association with spermatogenic failure? (2009). Mol Hum Reprod 15:733-8.
32. Amor-Guéret M, Dubois-d'Enghien C, Laugé A, Oncercq-Delic R, Barakat A, Chadli E, Bousfiha AA, Benjelloun M, Flori E, Doray B, Laugel V, Lourenço MT, Gonçalves R, Sousa S, Couturier J, Stoppa-Lyonnet D. Three new BLM gene mutations associated with Bloom syndrome (2008). Genet Test 12: 257-61.
33. El Houate B, Rouba H, Imken L, Sibai H, Chafik A, Boulouiz R, Chadli E, Hassar M, McElreavey K, Barakat A. No association between T222P/LGR8 mutation and cryptorchidism in Moroccan population (2008). *Hormone research* 70:236-9
34. Nahili H, Boulouiz R, Abidi O , Imken L, hassan R, Chafik A, Hassar M, Barakat A .Absence of the GJB3 and GJB6 mutation in Moroccan patientswith autosomal recessivenon syndromic deafness (2008). *International Journal of Pediatric Otorhinolaryngology* 72:1633-6
35. Abidi O, Boulouiz R, Nahili H, Bakhouch K, Wakrim L, Rouba H, Chafik A, Hassar M, Barakat A. Carrier frequencies of mutations/polymorphisms in the Connexin 26 gene (GJB2) in the Moroccan population (2008) . *Genet Test* 12:569-574.
36. Boulouiz R, Li Y, Soualhine H, Abidi O, Chafik A, Nürnberg G, Becker C, Nürnberg P, Kubisch C, Wollnik B, Barakat A. A novel mutation in the espin gene causes autosomal recessive nonsyndromic hearing loss but no apparent vestibular dysfunction in a moroccan family (2008). *Am J Med Genet A* 146:3086-
37. Abidi O, Boulouiz R, Nahili H, Imken L, Rouba H, Chafik A, Barakat A. The analysis of three markers flanking GJB2 gene suggestes a single origin of the most common 35delG mutation in the Moroccan population (2008). *Biochem Biophys Res Commun* 377:971-4.
38. ABIDI O, Barakat A. La part de l'hérédité dans les déficits auditifs ou surdités. Les Technologies de Laboratoire (2008), N°12, 11-16
39. El Houate B, Rouba H, Sibai H, Barakat A, Chafik A, Chadly El B, Imken L, Bogatcheva NV, Feng S, AgoulniK Aiand McElreavey K. Novel mutations involving the INSL3 gene associated with cryptorchidism (2007). *J Urol* 177:1947-51
40. Abidi O, Boulouiz R, Nahili H, Ridal M, Alami M N, Tlili A, Rouba H, Masmoudi S, Chafik A, Hassar M, Barakat A. GJB2 (Connexin 26) gene mutations in Moroccan patients with Prelingual Nonsyndromic Hearing Impairment and carrier frequency of the common GJB2-35delG mutation (2007). *Inter J of Pediatric Otorhinolaryngology* 71:1239-4.
41. Imken L, El Houate B, Chafik A, Nahili H, Boulouiz R, Abidi R, Chadl E, Louanjli N, Elfath A, McElreavey K, Hassar M, Barakat A, Rouba H. AZF microdeletions and partial deletions of AZFc region on the Y chromosome in Moroccan men (2007). *Asian J Androl* 9:674-8.
42. Boulouiz R, Li Y, Abidi O, Bolz H, Chafik A, Kubisch C, Rouba H, Wollnik B, Barakat A. Analysis of MYO7A in a Moroccan family with Usher syndrome type 1B: novel loss-of-function mutation and nonpathogenicity of p.Y1719C (2007). *Mol Vis* 13:1862-5.