



Identification of 28 novel mutations in the Bardet-Biedl syndrome genes: the burden of private mutations in an extensively heterogeneous disease

Submitted by Emmanuel Lemoine on Tue, 02/24/2015 - 15:41

Titre	Identification of 28 novel mutations in the Bardet-Biedl syndrome genes: the burden of private mutations in an extensively heterogeneous disease
Type de publication	Article de revue
Auteur	Muller, J. [1], Stoetzel, C. [2], Vincent, M. C [3], Leitch, C. C [4], Laurier, V. [5], Danse, J. M [6], Helle, S. [7], Marion, V. [8], Bennouna-Greene, V. [9], Vicaire, S. [10], Megarbane, A. [11], Kaplan, Josseline [12], Drouin-Garraud, V. [13], Hamdani, M. [14], Sigaudy, S. [15], Francannet, C. [16], Roume, J. [17], Bitoun, P. [18], Goldenberg, A. [19], Philip, N. [20], Odent, S. [21], Green, J. [22], Cossee, M. [23], Davis, E. E [24], Katsanis, N. [25], Bonneau, Dominique [26], Verloes, A. [27], Poch, O. [28], Mandel, J. L [29], Dollfus, H�el�ene [30]
Editeur	Springer Verlag
Type	Article scientifique dans une revue � comit� de lecture
Ann�e	2010
Langue	Anglais
Date	2010
Num�ro	5
Pagination	583 - 593
Volume	127
Titre de la revue	Human Genetics
ISSN	1432-1203
Mots-cl�s	Adult [31], Aged [32], Bardet-Biedl Syndrome/diagnosis/genetics [33], Chromatography, High Pressure Liquid [34], Chromosome Mapping [35], Decision Trees [36], Female [37], Gene Deletion [38], Gene Duplication [39], Gene Frequency [40], Genetic Testing [41], Homozygote [42], Humans [43], Male [44], Microsatellite Repeats [45], Middle Aged [46], Molecular Sequence Data [47], Mutation [48], Pedigree [49], Polymorphism, Single Nucleotide [50], Polymorphism, Single-Stranded Conformational [51], Sequence Analysis, DNA [52]

Résumé en
anglais

Bardet-Biedl syndrome (BBS), an emblematic disease in the rapidly evolving field of ciliopathies, is characterized by pleiotropic clinical features and extensive genetic heterogeneity. To date, 14 BBS genes have been identified, 3 of which have been found mutated only in a single BBS family each (BBS11/TRIM32, BBS13/MKS1 and BBS14/MKS4/NPHP6). Previous reports of systematic mutation detection in large cohorts of BBS families (n > 90) have dealt only with a single gene, or at most small subsets of the known BBS genes. Here we report extensive analysis of a cohort of 174 BBS families for 12/14 genes, leading to the identification of 28 novel mutations. Two pathogenic mutations in a single gene have been found in 117 families, and a single heterozygous mutation in 17 families (of which 8 involve the BBS1 recurrent mutation, M390R). We confirm that BBS1 and BBS10 are the most frequently mutated genes, followed by BBS12. No mutations have been found in BBS11/TRIM32, the identification of which as a BBS gene only relies on a single missense mutation in a single consanguineous family. While a third variant allele has been observed in a few families, they are in most cases missenses of uncertain pathogenicity, contrasting with the type of mutations observed as two alleles in a single gene. We discuss the various strategies for diagnostic mutation detection, including homozygosity mapping and targeted arrays for the detection of previously reported mutations.

URL de la notice <http://okina.univ-angers.fr/publications/ua8275> [53]
DOI [10.1007/s00439-010-0804-9](https://doi.org/10.1007/s00439-010-0804-9) [54]
Lien vers le document <http://dx.doi.org/10.1007/s00439-010-0804-9> [54]
Titre abrégé Hum Genet

Liens

- [1] [http://okina.univ-angers.fr/publications?f\[author\]=14078](http://okina.univ-angers.fr/publications?f[author]=14078)
- [2] [http://okina.univ-angers.fr/publications?f\[author\]=14079](http://okina.univ-angers.fr/publications?f[author]=14079)
- [3] [http://okina.univ-angers.fr/publications?f\[author\]=14080](http://okina.univ-angers.fr/publications?f[author]=14080)
- [4] [http://okina.univ-angers.fr/publications?f\[author\]=14081](http://okina.univ-angers.fr/publications?f[author]=14081)
- [5] [http://okina.univ-angers.fr/publications?f\[author\]=14082](http://okina.univ-angers.fr/publications?f[author]=14082)
- [6] [http://okina.univ-angers.fr/publications?f\[author\]=14083](http://okina.univ-angers.fr/publications?f[author]=14083)
- [7] [http://okina.univ-angers.fr/publications?f\[author\]=14084](http://okina.univ-angers.fr/publications?f[author]=14084)
- [8] [http://okina.univ-angers.fr/publications?f\[author\]=14085](http://okina.univ-angers.fr/publications?f[author]=14085)
- [9] [http://okina.univ-angers.fr/publications?f\[author\]=14086](http://okina.univ-angers.fr/publications?f[author]=14086)
- [10] [http://okina.univ-angers.fr/publications?f\[author\]=14087](http://okina.univ-angers.fr/publications?f[author]=14087)
- [11] [http://okina.univ-angers.fr/publications?f\[author\]=14088](http://okina.univ-angers.fr/publications?f[author]=14088)
- [12] [http://okina.univ-angers.fr/publications?f\[author\]=749](http://okina.univ-angers.fr/publications?f[author]=749)
- [13] [http://okina.univ-angers.fr/publications?f\[author\]=13549](http://okina.univ-angers.fr/publications?f[author]=13549)
- [14] [http://okina.univ-angers.fr/publications?f\[author\]=14090](http://okina.univ-angers.fr/publications?f[author]=14090)
- [15] [http://okina.univ-angers.fr/publications?f\[author\]=14091](http://okina.univ-angers.fr/publications?f[author]=14091)
- [16] [http://okina.univ-angers.fr/publications?f\[author\]=14092](http://okina.univ-angers.fr/publications?f[author]=14092)
- [17] [http://okina.univ-angers.fr/publications?f\[author\]=14093](http://okina.univ-angers.fr/publications?f[author]=14093)
- [18] [http://okina.univ-angers.fr/publications?f\[author\]=14094](http://okina.univ-angers.fr/publications?f[author]=14094)
- [19] [http://okina.univ-angers.fr/publications?f\[author\]=14095](http://okina.univ-angers.fr/publications?f[author]=14095)
- [20] [http://okina.univ-angers.fr/publications?f\[author\]=13567](http://okina.univ-angers.fr/publications?f[author]=13567)
- [21] [http://okina.univ-angers.fr/publications?f\[author\]=13606](http://okina.univ-angers.fr/publications?f[author]=13606)
- [22] [http://okina.univ-angers.fr/publications?f\[author\]=14096](http://okina.univ-angers.fr/publications?f[author]=14096)
- [23] [http://okina.univ-angers.fr/publications?f\[author\]=14097](http://okina.univ-angers.fr/publications?f[author]=14097)
- [24] [http://okina.univ-angers.fr/publications?f\[author\]=14098](http://okina.univ-angers.fr/publications?f[author]=14098)

- [25] [http://okina.univ-angers.fr/publications?f\[author\]=14099](http://okina.univ-angers.fr/publications?f[author]=14099)
- [26] <http://okina.univ-angers.fr/d.bonneau/publications>
- [27] [http://okina.univ-angers.fr/publications?f\[author\]=13537](http://okina.univ-angers.fr/publications?f[author]=13537)
- [28] [http://okina.univ-angers.fr/publications?f\[author\]=14100](http://okina.univ-angers.fr/publications?f[author]=14100)
- [29] [http://okina.univ-angers.fr/publications?f\[author\]=14101](http://okina.univ-angers.fr/publications?f[author]=14101)
- [30] [http://okina.univ-angers.fr/publications?f\[author\]=676](http://okina.univ-angers.fr/publications?f[author]=676)
- [31] [http://okina.univ-angers.fr/publications?f\[keyword\]=1002](http://okina.univ-angers.fr/publications?f[keyword]=1002)
- [32] [http://okina.univ-angers.fr/publications?f\[keyword\]=1072](http://okina.univ-angers.fr/publications?f[keyword]=1072)
- [33] [http://okina.univ-angers.fr/publications?f\[keyword\]=13389](http://okina.univ-angers.fr/publications?f[keyword]=13389)
- [34] [http://okina.univ-angers.fr/publications?f\[keyword\]=10850](http://okina.univ-angers.fr/publications?f[keyword]=10850)
- [35] [http://okina.univ-angers.fr/publications?f\[keyword\]=13390](http://okina.univ-angers.fr/publications?f[keyword]=13390)
- [36] [http://okina.univ-angers.fr/publications?f\[keyword\]=13391](http://okina.univ-angers.fr/publications?f[keyword]=13391)
- [37] [http://okina.univ-angers.fr/publications?f\[keyword\]=1075](http://okina.univ-angers.fr/publications?f[keyword]=1075)
- [38] [http://okina.univ-angers.fr/publications?f\[keyword\]=6400](http://okina.univ-angers.fr/publications?f[keyword]=6400)
- [39] [http://okina.univ-angers.fr/publications?f\[keyword\]=9189](http://okina.univ-angers.fr/publications?f[keyword]=9189)
- [40] [http://okina.univ-angers.fr/publications?f\[keyword\]=13392](http://okina.univ-angers.fr/publications?f[keyword]=13392)
- [41] [http://okina.univ-angers.fr/publications?f\[keyword\]=12989](http://okina.univ-angers.fr/publications?f[keyword]=12989)
- [42] [http://okina.univ-angers.fr/publications?f\[keyword\]=11681](http://okina.univ-angers.fr/publications?f[keyword]=11681)
- [43] [http://okina.univ-angers.fr/publications?f\[keyword\]=991](http://okina.univ-angers.fr/publications?f[keyword]=991)
- [44] [http://okina.univ-angers.fr/publications?f\[keyword\]=968](http://okina.univ-angers.fr/publications?f[keyword]=968)
- [45] [http://okina.univ-angers.fr/publications?f\[keyword\]=13393](http://okina.univ-angers.fr/publications?f[keyword]=13393)
- [46] [http://okina.univ-angers.fr/publications?f\[keyword\]=5941](http://okina.univ-angers.fr/publications?f[keyword]=5941)
- [47] [http://okina.univ-angers.fr/publications?f\[keyword\]=8594](http://okina.univ-angers.fr/publications?f[keyword]=8594)
- [48] [http://okina.univ-angers.fr/publications?f\[keyword\]=1133](http://okina.univ-angers.fr/publications?f[keyword]=1133)
- [49] [http://okina.univ-angers.fr/publications?f\[keyword\]=13394](http://okina.univ-angers.fr/publications?f[keyword]=13394)
- [50] [http://okina.univ-angers.fr/publications?f\[keyword\]=8596](http://okina.univ-angers.fr/publications?f[keyword]=8596)
- [51] [http://okina.univ-angers.fr/publications?f\[keyword\]=13395](http://okina.univ-angers.fr/publications?f[keyword]=13395)
- [52] [http://okina.univ-angers.fr/publications?f\[keyword\]=7686](http://okina.univ-angers.fr/publications?f[keyword]=7686)
- [53] <http://okina.univ-angers.fr/publications/ua8275>
- [54] <http://dx.doi.org/10.1007/s00439-010-0804-9>

Publié sur *Okina* (<http://okina.univ-angers.fr>)