MEF2C haploinsufficiency caused by either microdeletion of the 5q14.3 region or mutation is responsible for severe mental retardation with stereotypic movements, epilepsy and/or cerebral malformations

Titre	MEF2C haploinsufficiency caused by either microdeletion of the 5q14.3 region or mutation is responsible for severe mental retardation with stereotypic movements, epilepsy and/or cerebral malformations
Type de publication	Article de revue
Auteur	Le Meur, N. [1], Holder-Espinasse, M. [2], Jaillard, S. [3], Goldenberg, A. [4], Joriot, S. [5], Amati-Bonneau, Patrizia [6], Guichet, A. [7], Barth, Magalie [8], Charollais, A. [9], Journel, H. [10], Auvin, S. [11], Boucher, C. [12], Kerckaert, J. P [13], David, V. [14], Manouvrier-Hanu, S. [15], Saugier-Veber, P. [16], Frebourg, T. [17], Dubourg, C. [18], Andrieux, J. [19], Bonneau, Dominique [20]
Editeur	BMJ Publishing Group
Type	Article scientifique dans une revue à comité de lecture
Année	2010
Langue	Anglais
Date	2010
Numéro	1
Pagination	22 - 29
Volume	47
Titre de la revue	Journal of Medical Genetics
ISSN	1468-6244
Mots-clés	Cerebrum/abnormalities/metabolism [21], Child [22], Child, Preschool [23], Chromosome Deletion [24], Chromosomes, Human, Pair 5/genetics [25], Epilepsy/genetics [26], Haploidy [27], Humans [28], Infant [29], Intellectual Disability/genetics [30], MADS Domain Proteins/genetics [31], MEF2 Transcription Factors [32], Myogenic Regulatory Factors/genetics [33], Stereotypic Movement Disorder/genetics [34]

Résumé en anglais	BACKGROUND: Over the last few years, array-comparative genomic hybridisation (CGH) has considerably improved our ability to detect cryptic unbalanced rearrangements in patients with syndromic mental retardation.METHOD: Molecular karyotyping of six patients with syndromic mental retardation was carried out using whole-genome oligonucleotide array-CGH. RESULTS: 5q14.3 microdeletions ranging from 216 kb to 8.8 Mb were detected in five unrelated patients with the following phenotypic similarities: severe mental retardation with absent speech, hypotonia and stereotypic movements. Facial dysmorphic features, epilepsy and/or cerebral malformations were also present in most of these patients. The minimal common deleted region of these 5q14 microdeletions encompassed only MEF2C, the gene for a protein known to act in brain as a neurogenesis effector, which regulates excitatory synapse number. In a patient with a similar phenotype, an MEF2C nonsense mutation was subsequently identified. CONCLUSION: Taken together, these results strongly suggest that haploinsufficiency of MEF2C is responsible for severe mental retardation with stereotypic movements, seizures and/or cerebral malformations.
URL de la notice	http://okina.univ-angers.fr/publications/ua8312 [35]
DOI	10.1136/jmg.2009.069732 [36]
Lien vers le document	http://dx.doi.org/10.1136/jmg.2009.069732 [36]
Titre abrégé	J Med Genet

Liens

[1] http://okina.univ-angers.fr/publications?f[author]=14277
[2] http://okina.univ-angers.fr/publications?f[author]=14278
[3] http://okina.univ-angers.fr/publications?f[author]=14103
[4] http://okina.univ-angers.fr/publications?f[author]=14095
[5] http://okina.univ-angers.fr/publications?f[author]=14279
[6] http://okina.univ-angers.fr/patrizia.bonneau/publications
[7] http://okina.univ-angers.fr/publications?f[author]=13604
[8] http://okina.univ-angers.fr/publications?f[author]=25471
[9] http://okina.univ-angers.fr/publications?f[author]=14280
[10] http://okina.univ-angers.fr/publications?f[author]=14107
[11] http://okina.univ-angers.fr/publications?f[author]=14281
[12] http://okina.univ-angers.fr/publications?f[author]=14282
[13] http://okina.univ-angers.fr/publications?f[author]=14283
[14] http://okina.univ-angers.fr/publications?f[author]=14116
[15] http://okina.univ-angers.fr/publications?f[author]=14284
[16] http://okina.univ-angers.fr/publications?f[author]=14285
[17] http://okina.univ-angers.fr/publications?f[author]=14286
[18] http://okina.univ-angers.fr/publications?f[author]=14117
[19] http://okina.univ-angers.fr/publications?f[author]=14287
[20] http://okina.univ-angers.fr/d.bonneau/publications
[21] http://okina.univ-angers.fr/publications?f[keyword]=13549
[22] http://okina.univ-angers.fr/publications?f[keyword]=1216
[23] http://okina.univ-angers.fr/publications?f[keyword]=1534
[24] http://okina.univ-angers.fr/publications?f[keyword]=13548
[25] http://okina.univ-angers.fr/publications?f[keyword]=13550
[26] http://okina.univ-angers.fr/publications?f[keyword]=13551
[27] http://okina.univ-angers.fr/publications?f[keyword]=13552

[28] http://okina.univ-angers.fr/publications?f[keyword]=991
[29] http://okina.univ-angers.fr/publications?f[keyword]=3233
[30] http://okina.univ-angers.fr/publications?f[keyword]=12896
[31] http://okina.univ-angers.fr/publications?f[keyword]=13553
[32] http://okina.univ-angers.fr/publications?f[keyword]=13554
[33] http://okina.univ-angers.fr/publications?f[keyword]=13555
[34] http://okina.univ-angers.fr/publications?f[keyword]=13556
[35] http://okina.univ-angers.fr/publications/ua8312
[36] http://dx.doi.org/10.1136/jmg.2009.069732