掲載した論文（発表題目）	発表者氏名	発表した場所（学会誌・雑誌等名）	発表した時期	国内・外の別
iPS細胞（幹細胞）を用いる医療の近未来	山本俊至, 下島圭子	遺伝子医学MOOK別冊「今さら聞けない『遺伝医学』」. pp173-181, メディカルドゥ, 大阪, 2014.	2014	国内
1p36欠失症候群	山本俊至	今日の小児治療指針, 水口雅, ら編. 医学書院, 東京 [in press]	2014	国内
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マイクロアレイ染色体検査 概論	山本俊至	小児内科47巻増刊号 病態生理2 [in press]	2014	国内
アレイCGH法によるてんかんの分子診断	山本俊至	医学のあゆみ [in press]	2014	国内
Morphological characterization of mammalian Timeless in the mouse brain development	Inaguma Y, Ito H, Hara A, Iwamoto I, Matsumoto A, Yamagata T, Tabata H, Nagata KI	Neurosci Res. 2014;92:21-28.	2014	国外
Acute neuropharmacological effects of atomoxetine on inhibitory control in ADHD children: a fNIRS study	Nagashima M, Monden Y, Dan I, Dan H, Tsuzuki D, Mizutani T, Kyutoku Y, Gunji Y, Hirano D, Taniguchi T, Shimoizumi H, Momoi MY, Watanabe E, Yamagata T	Neuroimage Clin. 2014;6:192-201.	2014	国外

掲載した論文（発表題目）	発表者氏名	発表した場所（学会誌・雑誌等名）	発表した時期	国内・外の別
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LIN7A depletion disrupts cerebral cortex development contributing to intellectual disability in 12q21-deletion syndrome	Matsumoto A, Mizuno M, Hamada N, Nozaki Y, Jimbo EF, Momoi MY, Nagata K, Yamagata T	PLoS One 2014;9:e92695.	2014	国外
New MT-ND6 and NDUFA1 mutations in mitochondrial respiratory chain disorders	Uehara N, Mori M, Tokuzawa Y, Mizuno Y, Tamaru S, Kohda M, Moriyama Y, Nakachi Y, Matoba N, Sakai T, Yamazaki T, Harashima H, Murayama K, Hattori K, Hayashi J, Yamagata T, Fujita Y, Ito M, Tanaka M, Nibu K, Ohtake A, Okazaki Y	Ann Clin Transl Neurol. 2014;1:361-9.	2014	国外
Genotype-phenotype correlation of Coffin-Siris syndrome caused by mutations in SMARCB1, SMARCA4, SMARCE1, and ARID1A	Kosho T, Okamoto N, Yamagata T, Coffin-Siris Syndrome International Collaborators	Am J Med Genet C 2014;166:262-275.	2014	国外
Persistent presence of the anti-myelin oligodendrocyte glycoprotein autoantibody in a pediatric case of acute disseminated encephalomyelitis followed by optic neuritis	Miyauchi A, Monden Y, Watanabe M, Sugie H, Morita M, Kezuka T, Momoi M, Yamagata T	Neuropediatrics 2014;45: 196-9.	2014	国外

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Nasal oxytocin administration reduces food intake without affecting locomotor activity and glycemia with c-Fos induction in limited brain areas	Maejima Y, Rita RS, Santoso P, Aoyama M, Hiraoka Y, Nishimori K, Gantulga D, Shimomura K, Yada T	Neuroendocrinology, 2015, [in press]	2015	国外
Peripheral oxytocin activates vagal afferent neurons to suppress feeding in normal and leptin-resistant mice: A route for ameliorating hyperphagia and obesity	Iwasaki Y, Maejima Y, Suyama S, Yoshida M, Arai T, Katsurada K, Kumari P, Nakabayashi H, Kakei M, Yada T	Am J Physiol Regul Integr Comp Physiol, 308:R360-R369, 2015.	2015	国外
Paraventricular NUCB2/nesfatin-1 is directly targeted by leptin and mediates its anorexigenic effect	Darambazar G, Nakata M, Okada T, Wang L, Li E, Shinozaki A, Motoshima M, Mori M, Yada T	Biochem Biophys Res Commun, 456(4):913-918, 2015.	2015	国外
Glucagon directly interacts with vagal afferent nodose ganglion neurons to induce Ca2+ signaling via glucagon receptors	Ayush EA, Iwasaki Y, Iwamoto S, Nakabayashi H, Kakei M, Yada T	Biochem Biophys Res Commun. 456(3):727-732, 2015.	2015	国外
Partial blockade of Kv2.1 channel potentiates GLP-1 ' s insulinotropic effects in islets and reduces its dose required for improving glucose tolerance in type 2 diabetic male mice	Rita R, Dezaki K, Kurashina T, Kakei M, Yada T	Endocrinology 156(1):114-123, 2015	2015	国外
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Early manifestations of BPAN in a pediatric patient.	Okamoto N, Ikeda T, Hasegawa T, Yamamoto Y, Kawato K, Komoto T, Imoto I.	Am J Med Genet A 164A; 3095-3099, 2014	2014	国外
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Autosomal recessive cystinuria caused by genome-wide paternal uniparental isodisomy in a patient with Beckwith-Wiedemann syndrome	Ohtsuka Y, Higashimoto K, Sasaki K, Jozaki K, Yoshinaga H, Okamoto N, Takama Y, Kubota A, Nakayama M, Yatsuki H, Nishioka K, Joh K, Mukai T, Yoshiura KI, Soejima H.	Clin Genet [in press]	2015	国外
Duplication of the NPHP1 gene in patients with autism spectrum disorder and normal intellectual ability: a case series	Yasuda Y, Hashimoto R, Fukai R, Okamoto N, Hiraki Y, Yamamori H, Fujimoto M, Ohi K, Taniike M, Mohri I, Nakashima M, Tsurusaki Y, Saitsu H, Matsumoto N, Miyake N, Takeda M.	Ann Gen Psychiatry [in press]	2014	国外
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The spectrum of ZEB2 mutations causing the Mowat-Wilson syndrome in Japanese populations	Yamada Y, Nomura N, Yamada K, Matsuo M, Suzuki Y, Sameshima K, Kimura R, Yamamoto Y, Fukushi D, Fukuhara Y, Ishihara N, Nishi E, Imataka G, Suzumura H, Hamano SI, Shimizu K, Iwakoshi M, Ohama K, Ohta A, Wakamoto H, Kajita M, Miura K, Yokochi K, Kosaki K, Kuroda T, Kosaki R, Hiraki Y, Saito K, Mizuno S, Kurosawa K, Okamoto N, Wakamatsu N	Am J Med Genet A 164A; 1899-1908, 2014	2014	国外
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Aortic aneurysm and craniosynostosis in a family with Cantu syndrome	Hiraki Y, Miyatake S, Hayashidani M, Nishimura Y, Matsuura H, Kamada M, Kawagoe T, Yunoki K, Okamoto N, Yofune H, Nakashima M, Tsurusaki Y, Satisu H, Murakami A, Miyake N, Nishimura G, Matsumoto N	Am J Med Genet A 164A; 231-236, 2014	2014	国外

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Endocrinological Characteristics of 25 Japanese Patients with CHARGE Syndrome	Shoji Y, Ida S, Etani Y, Yamada H, Kayatani F, Suzuki Y, Kosaki K, Okamoto N.	Clin Pediatr Endocrinol 23; 45-51, 2014	2014	国外
A clinical study of patients with pericentromeric deletion and duplication within 16p12.2-p11.2	Okamoto N, Fujii T, Tanaka J, Saito K, Matsui T, Harada N	Am J Med Genet A 164A; 213-219, 2014	2014	国外
Microarray and FISH-based genotype-phenotype analysis of 22 Japanese patients with Wolf-Hirschhorn syndrome	Shimizu K, Wakui K, Kosho T, Okamoto N, Mizuno S, Itomi K, Hattori S, Nishio K, Samura O, Kobayashi Y, Kako Y, Arai T, Tsutomu OI, Kawame H, Narumi Y, Ohashi H, Fukushima Y	Am J Med Genet A 164A; 597-609, 2014	2014	国外
PIGN mutations cause congenital anomalies, developmental delay, hypotonia, epilepsy, and progressive cerebellar atrophy	Ohba C, Okamoto N, Murakami Y, Suzuki Y, Tsurusaki Y, Nakashima M, Miyake N, Tanaka F, Kinoshita T, Matsumoto N, Saitsu H	Neurogenetics 15; 85-92, 2014	2014	国外
Coffin-Siris syndrome is a SWI/SNF complex disorder	Tsurusaki Y, Okamoto N, Ohashi H, Mizuno S, Matsumoto N, Makita Y, Fukuda M, Isidor B, Perrier J, Aggarwal S, Dalal A, Al-Kindy A, Liebelt J, Mowat D, Nakashima M, Saitsu H, Miyake N, Matsumoto N	Clin Genet 85; 548-554, 2014	2014	国外

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De novo SOX11 mutations cause Coffin-Siris syndrome.	Tsurusaki Y, Koshimizu E, Ohashi H, Phadke S, Kou I, Shiina M, Suzuki T, Okamoto N, Imamura S, Yamashita M, Watanabe S, Yoshiura K, Kodera H, Miyatake S, Nakashima M, Saitsu H, Ogata K, Ikegawa S, Miyake N, Matsumoto N.	Nat Commun. Jun 2;5:4011. doi: 10.1038/ncomms5011. 2014	2014	国外
A hemizygous GYG2 mutation and Leigh syndrome: a possible link?	Imagawa E, Osaka H, Yamashita A, Shiina M, Takahashi E, Sugie H, Nakashima M, Tsurusaki Y, Saitsu H, Ogata K, Matsumoto N, Miyake N	Hum Genet 133; 225-34, 2014.	2014	国外
Early magnetic resonance detection of cortical necrosis and acute network injury associated with neonatal and infantile cerebral infarction	Okabe T, Aida N, Niwa T, Nozawa K, Shibasaki J, Osaka H	Pediatr Radiol 53; 448-58, 2014.	2014	国外
A Japanese adult case of guanidinoacetate methyltransferase deficiency	Akiyama T, Osaka H, Shimbo H, Nakajiri T, Kobayashi K, Oka M, Endo F, Yoshinaga H	JIMD Rep 12; 65-9, 2014.	2014	国外
A Novel two-nucleotide deletion in the ATP7A gene associated with delayed infantile onset of Menkes disease	Wada T, Haddad MR, Yi L, Murakami T, Sasaki A, Shimbo H, Kodama H, Osaka H, Kaler SG	Pediatr Neurol 50; 417-20, 2014.	2014	国外

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A rapid screening with direct sequencing from blood samples for the diagnosis of Leigh syndrome	Shimbo H, Takagi M, Okuda M, Tsuyusaki Y, Takano K, Iai M, Yamashita S, Murayama K, Ohtake A, Goto Y, AidaN, Osaka H	Mol Genet Metab Report 1; 133-138, 2014.	2014	国外
A three-year-old boy with glucose transporter type 1 deficiency syndrome presenting with episodic ataxia	Ohshiro-Sasaki A, Shimbo H, Takano K, Wada T, Osaka H	Pediatr Neurol 50; 99-100, 2014.	2014	国外
Causative novel PNKP mutations and concomitant PCDH15 mutations in a patient with microcephaly with early-onset seizures and developmental delay syndrome and hearing loss	Nakashima M, Takano K, Osaka H, Aida N, Tsurusaki Y, Miyake N, Saitsu H, Matsumoto N	J Hum Genet 59; 471-4, 2014.	2014	国外
Expanding the phenotypic spectrum of TUBB4A-associated hypomyelinating leukoencephalopathies	Miyatake S, Osaka H, Shiina M, Sasaki M, Takanashi J, Haginoya K, Wada T, Morimoto M, Ando N, Ikuta Y, Nakashima M, Tsurusaki Y, Miyake N, Ogata K, Matsumoto N, Saitsu H	Neurology 82; 2230-7, 2014.	2014	国外
Effect of CYP2C19 polymorphisms on stiripentol administration in Japanese cases of Dravet syndrome	Kouga T, Shimbo H, Iai M, Yamashita S, Ishii A, Ihara Y, Hirose S, Yamakawa K, Osaka H	Brain Dev. 37:243-9, 2015	2015	国外

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PIGA mutations cause early-onset epileptic encephalopathies and distinctive features	Kato M, Saitsu H, Murakami Y, Kikuchi K, Watanabe S, Iai M, Miya K, Matsuura R, Takayama R, Ohba C, Nakashima M, Tsurusaki Y, Miyake N, Hamano S, Osaka H, Hayasaka K, Kinoshita T, Matsumoto N	Neurology 82; 1587-96, 2014.	2014	国外
Genotype-phenotypecorrelation of contiguous gene deletions of SLC6A8, BCAP31 and ABCD1	van de Kamp J, Errami A, Howidi M, Anselm I, Winter S, Phalin-Roque J, Osaka H, van Dooren S, Mancini G, Steinberg S, Salomons G	Clin Genet 87:141-7, 2015	2015	国外
Epidemiological, clinical, and genetic landscapes ofhypomyelinating leukodystrophies	Numata Y, Gotoh L, Iwaki A, Kurosawa K, Takanashi J, Deguchi K, Yamamoto T, Osaka H, Inoue K	J Neurol 261; 752-8, 2014.	2014	国外
PIGO mutations in intractable epilepsy and severe developmental delay with mild elevation of alkaline phosphatase levels	Nakamura K, Osaka H, Murakami Y, Anzai R, Nishiyama K, Kodera H, Nakashima M, Tsurusaki Y, Miyake N, Kinoshita T, Matsumoto N, Saitsu H	Epilepsia 55; e13-7, 2014.	2014	国外
Involvement of ER stress in dysmyelination of Pelizaeus-Merzbacher disease with PLP1 missense mutations shown by iPSC-derived oligodendrocytes	Numasawa-Kuroiwa Y, Okada Y, Shibata S, Kishi N, Akamatsu W, Shoji M, Nakanishi A, Oyama M, Osaka H, Inoue K, Takahashi K, Yamanaka S, Kosaki K, Takahashi T, Okano H	Stem Cell Reports 2; 648-61, 2014.	2014	国外

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Seizure recurrence followingpyridoxine withdrawal in a patient with pyridoxine-dependent epilepsy	Tamaura M, Shimbo H, Iai M, Yamashita S, Osaka H	Brain Dev 37:442-5,2015	2015	国外
Mutations in the glutaminyl-tRNA synthetase gene cause early-onset epileptic encephalopathy	Kodera H, Osaka H, Iai M, Aida N, Yamashita A, Tsurusaki Y, Nakashima M,Miyake N, Saitsu H, Matsumoto N	J Hum Genet 60:97-101,2015	2015	国外
A Japanese girl with an early-infantile onset vanishing white matter disease resembling Creeleukoencephalopathy	Takano K, Tsuyusaki Y, Sato M, Takagi M, Anzai R, Okuda M, Iai M, Yamashita S, Okabe T, Aida N, Tsurusaki Y, Saitsu H, Matsumoto N, Osaka H	Brain Dev [in press]	2014	国外
Intracranial hemorrhage and tortuosity of veins detected on susceptibility-weighted imaging of a child with a type IV collagen α 1 mutation and schizencephaly	Niwa T, Aida N, Osaka H, Wada T, Saitsu H, Imai Y	Magn Reson Med Sci [in press]	2014	国外
大脳萎縮症	小坂　仁	編集　水澤秀洋、新領域別症候群シリーズNo.29「神経症候群（第２版）IV、日本臨牀社p.319-324.2014	2014	国内
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自閉性障害の多様な遺伝学的病態とシナプス関連病因遺伝子の解析	山形崇倫、松本歩、永田浩一	脳と発達　Vol.46 No.2 125-130, 2014	2014	国内

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Roles of Rho small GTPases in the tangentially migrating neurons	Ito H, Morishita R, Tabata H, Nagata K	Histol. Histopathol. 29: 871-879, 2014.	2014	国外
Expression of Drebrin, an actin binding protein, in basal cell carcinoma, trichoblastoma and trichoepithelioma	Mizutani Y, Iwamoto I, Kanoh H, Seishima M, Nagata K	Histol Histopathol 29, 757-766, 2014	2014	国外
SIL1, a causative cochaperone gene of Marinesco-Sj ö gren syndrome, plays an essential role in establishing the architecture of the developing cerebral cortex	Inaguma Y, Hamada N, Tabata H, Iwamoto I, Mizuno M, Nishimura VY, Ito H, Morishita R, Suzuki M, Ohno K, Kumagai T, Nagata K	EMBO Mol. Med. 6: 414-429, 2014	2014	国外
Phosphorylation of Drebrin by cyclin-dependent kinase 5 and its role in neuronal migration	Tanabe K, Yamazaki H, Inaguma Y, Asada A, Kimura T, Takahashi J, Taoka M, Ohshima T, Furuichi T, Isobe T, Nagata K, Shirao T, Hisanaga S	PLOS ONE 9(3): e92291, 2014.	2014	国外
Establishment of an in vivo electroporation method into postnatal newborn neurons in the dentate gyrus	Ito H, Morishita R, Iwamoto I, Nagata K	Hippocampus 24:1449-1457, 2014.	2014	国外
Cdk5 and its substrates, Dcx and p27kip1, regulate cytoplasmic dilation formation and nuclear elongation in migrating neurons	Nishimura YV, Shikanai M, Hoshino M, Ohshima T, Nabeshima Y, Mizutani K, Nagata K, Nakajima K, Kawauchi T	Development in press	2015	国外

掲載した論文（発表題目）	発表者氏名	発表した場所（学会誌・雑誌等名）	発表した時期	国内・外の別
Disrupted-in-schizophrenia 1 (DISC1) regulates dysbindin function by enhancing its stability	Lee S-A, Kim S-M, Suh B K, Sun H-Y, Park Y-U, Hong J-H, Park C, Nguyen M D, Nagata K, Yoo J-Y, Park S K	J. Biol. Chem. 290:7087-96, 2015	2015	国外
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Derivation of Mesenchymal Stromal Cells from Pluripotent Stem Cells through a Neural Crest Lineage using Small Molecule Compounds with Defined Media	Fukuta M, Nakai Y, Kirino K, Nakagawa M, Sekiguchi K, Nagata S, Matsumoto Y, Yamamoto T, Umeda K, Heike T, Okumura N, Koizumi N, Sato T, Nakahata T, Saito M, Otsuka T, Kinoshita S, Ueno M, Ikeya M, Toguchida J	PLoS One. 201; 9: e112291.2014	2014	国外
Enhanced chondrogenesis of iPS cells from neonatal-onset multisystem inflammatory disease occurs via the caspase-1-independent cAMP/PKA/CREB pathway	Yokoyama K, Ikeya M, Umeda K, Oda H, Nodomi S, Nasu A, Matsumoto Y, Izawa K, Horigome K, Kusaka T, Tanaka T, Saito MK, Yasumi T, Nishikomori R, Ohara O, Nakayama N, Nakahata T, Heike T, Toguchida J	Arthritis Rheumatol 67:302-14, 2015	2015	国外
Modeling the early phenotype at the neuromuscular junction of spinal muscular atrophy using patient-derived iPSCs	Yoshida M, Kitaoka S, Egawa N, Yamane M, Ikeda R, Tsukita K, Amano N, Watanabe A, Morimoto M, Takahashi J, Hosoi H, Nakahata T, Inoue H, Saito MK	Stem Cell Reports (in press)	2015	国外
明日の診療に役立つ細胞分子生物学再生医療-iPS細胞の応用	齋藤潤	日本呼吸器学会雑誌 3(5) 625-629, 2014	2014	国内
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Ⅳ. 研究成果の刊行物・別刷

RESEARCH ARTICLE

AMERICAN JOURNAL OF medical genetics PART B
Neuropsychiatric Genetics

Neuropsychological Profiles of Patients With 2q37.3 Deletion Associated With Developmental Dyspraxia

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Patients with 2q37 deletions manifest brachydactyly mental retardation syndrome (BDMR). Recent advances in human molecular research have revealed that alterations in the histone deacetylase 4 gene (*HDAC4*) are responsible for the clinical manifestations of BDMR. Here, we report two male patients with 2q37.3 deletions. One of the patients showed a typical BDMR phenotype, and *HDAC4* was included in the deletion region. *HDAC4* was preserved in the other patient, and he showed a normal intelligence level with the delayed learning of complex motor skills. Detailed neuropsychological examinations revealed similar neuropsychological profiles in these two patients (visuo-spatial dyspraxia) that suggested developmental dyspraxia. These observations suggested that some other candidate genes for neuronal development exist in the telomeric region of *HDAC4*. © 2014 Wiley Periodicals, Inc.

Key words: 2q37.3 deletion; intellectual disability; autism spectrum disorder; developmental dyspraxia; brachydactyly mental retardation syndrome (BDMR); Albright hereditary osteodystrophy-like syndrome (AHO-like); histone deacetylase 4 gene (HDAC4)

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INTRODUCTION

Chromosomal deletions in the 2q37 region have been identified in over 100 patients with brachydactyly mental retardation syndrome (BDMR), which is synonymous with Albright hereditary osteodystrophy-like syndrome (AHO-like) [Villavicencio-Lorini et al., 2013]. BDMR is a complex disorder that presents with a spectrum of clinical features, including developmental delay, obesity, autism spectrum disorder, and craniofacial and skeletal abnormalities, including brachydactyly type E in approximately 50% of the cases [Williams et al., 2010]. A recent genotype-phenotype correlation study has revealed that the histone deacetylase 4 gene (*HDAC4*) is located on the overlapping 2q37 deletions that have been reported in patients with BDMR and intragenic mutations of *HDAC4* have been identified in patients with BDMR who did not show 2q37 deletions, indicating that *HDAC4* is responsible for this clinical condition [Williams et al., 2010].

It is controversial whether additional telomeric regions of *HDAC4* are related to certain clinical features. In this study, we describe two patients with subtelomeric-invisible-small 2q37.3 deletions. One of the patients showed a typical BDMR phenotype, and the deletion region included *HDAC4*. This was consistent with

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previous knowledge. However, the deletion that was identified in the other patient did not include *HDAC4*. We evaluated these patients in order to gain a better understanding of the genotype-phenotype correlation regarding the neuropsychological manifestations of patients with 2q37 deletions.

MATERIALS AND METHODS

Materials

Peripheral blood samples were obtained from the patients and their families after obtaining informed consent from them and a permission from the institution's ethnical committee. DNA was extracted from the peripheral blood samples with a QIAamp DNA mini kit (QIAGEN, Venlo, The Netherlands). For the fluorescence in-situ hybridization (FISH) analysis, metaphase spreads were prepared from the peripheral blood samples with a standard method as previously described [Shimojima et al., 2009, 2011].

Molecular and Cytogenetic Analysis

Genomic copy numbers were analyzed with a SurePrint G3 Hmn CGH 60 k Oligo Microarray Kit (Agilent Technologies, Santa Clara, CA) according to a method described elsewhere [Shimojima

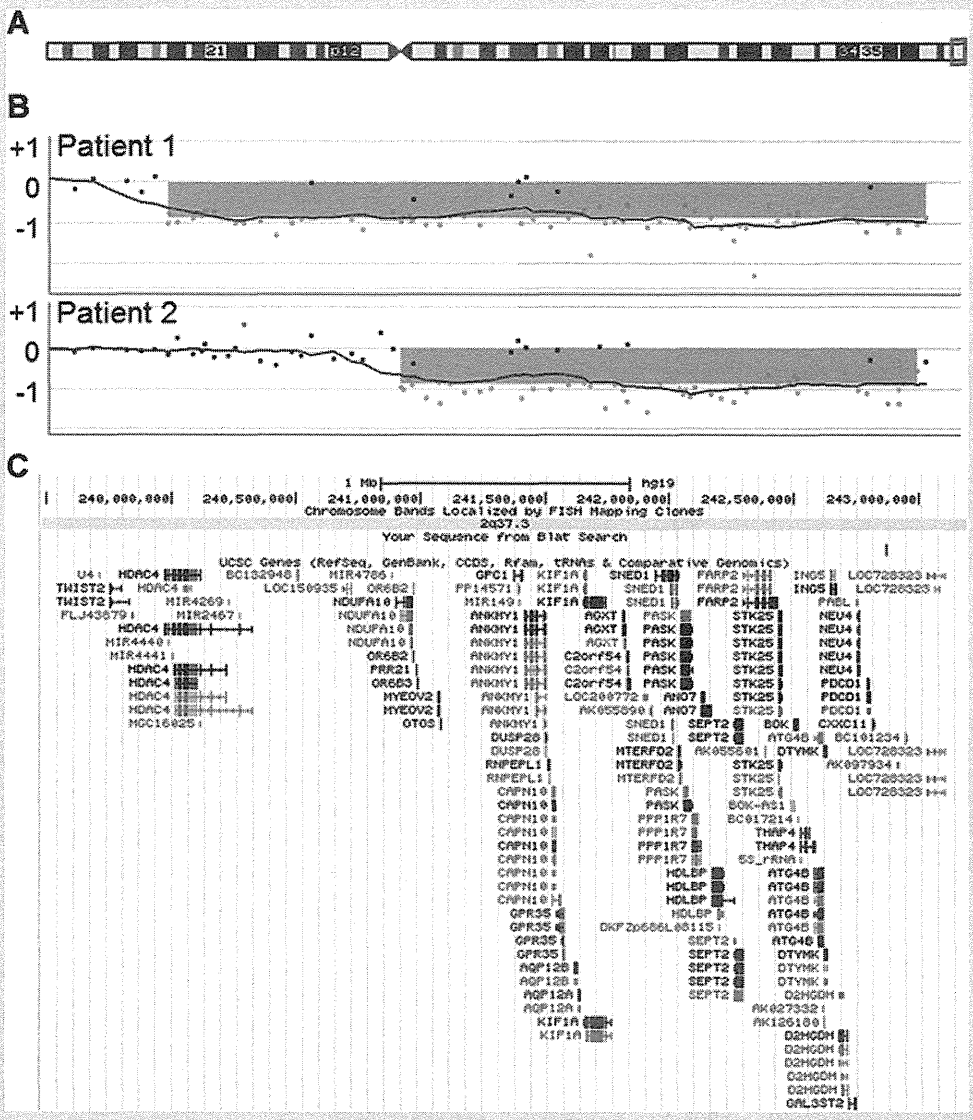

FIG. 1. Chromosomal microarray testing results and the genome map around 2q37. 3. A: A schematic representation of chromosome 2 that is downloaded from the UCSC genome browser. The red region is expanded in B and C. B: The results of chromosomal microarray testing are shown by Gene View, which is visualized by Agilent Genomic Workbench version 6.5 [Agilent Technologies]. The blue-translucent rectangles indicate the aberration region. The X- and Y-axes indicate the genomic location and signal $\log_2$ ratio. C: The gene map of 2q27.3 that was downloaded from the USCS genome-browser is shown at the same scale as the microarray testing. *HDAC4* is included in the aberration region in patient 1 but not in patient 2.

et al., 2009, 2011]. Genomic copy number aberrations were visualized with the Agilent Genomic Workbench version 6.5 (Agilent Technologies). In order to confirm the results of the chromosomal microarray testing, a FISH analysis was performed with the human bacterial artificial chromosomes (BACs) RP11–1077A16 (chr2q37.3:242,263,100–242,478,658), which was used as a target probe, and RP11–137A4 (chr2p22.2:37,211,567–37,360,171), which was used as a marker probe. Both probes were selected from the University of California Santa Cruz (UCSC) genome browser (http://www.gwenome.ucsc.edu). All of the genomic positions refer to the human reference genome build 19.

Neuropsychological Examinations

Both patients were evaluated with a comprehensive neuropsychological examination. The Wechsler Intelligence Scale for Children-III (WISC-III) was used to evaluate general intelligence. The Developmental Test of Visual Perception (DTVP) was administered to evaluate visual perception [Fazzi et al., 2004]. The Developmental Voluntary Movement Test-Revised (DVMT-R) was used to assess motor coordination and control [Yamane et al., 1990]. This test is a standardized childhood assessment for subjects who are 2–6 years old, and it consists of 40 items in three categories of voluntary movement: finger movement (three subscales: finger flexion, finger extension, and hand coordination), face and oral movement (three subscales: lips and cheeks, tongue, and speech), and upper and lower extremity movement (three subscales: jumping, balancing, and body coordination). The age at which 90% of the children acquire each tested movement is considered to determine their normal range of performance. Patients had to imitate the examiner's performance, and their performance was evaluated by the examiner as their developmental age, at which 90% of the children acquired each performance. We used the Childhood Autism Rating Scale (CARS) that was translated into Japanese to assess autistic symptoms [Schopler et al., 1980; Kanai et al., 2004; Taniai et al., 2008].

RESULTS

Genomic Copy Number Aberrations

Both patients showed genomic copy number aberrations in the region of 2q37.3. The genotype data are depicted in the genome map in Figure. 1. Patient 1 showed a genomic copy number loss of 3.2–Mb from the telomere, indicating arr 2q37.3(239,966,964–243,199,373) × 1. Patient 2 also showed a genomic copy number loss of 2.3 Mb from the telomere, indicating arr 2q37.3 (240,912,892–243,199,373) × 1. There were no other aberrations in either patient. The subsequent FISH analyses confirmed simple terminal deletions in the metaphase spreads in both patients (Fig. 2). Because all four parents declined to be genotyped, it is unknown whether these aberrations were de novo or inherited.

Clinical Description

Patient 1. A 16-year-old, right-handed, Japanese boy was born at 38 weeks gestation after an unremarkable pregnancy with a birth weight of 2,570 g (10–25th centile) to healthy non-consanguineous parents. His family history was not remarkable, and his younger brother was healthy. He had delayed developmental milestones, e.g., he did not walk until 19 months or talk until 24 months. He showed a moderate intellectual disability and attended a special school.

At present, his height is 165 cm (10–25th centile), and his weight is 58 kg (25–50th centile). The patient has a distinctive face with frontal bossing, down-slanting palpebral fissures, a wide and flat

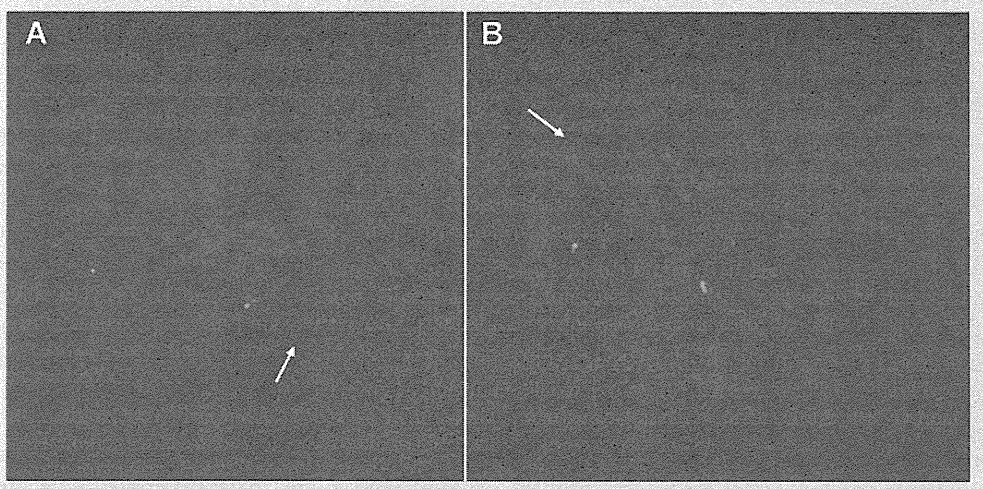

FIG. 2. The results of fluorescence in-situ hybridization analysis. A: Patient 1. B: Patient 2. The single green signal of RP11–1077A16 indicates the deletion of 2q37.3 in each patient. The white arrows indicate the deletion regions. The two red signals are the markers of chromosome 2.

nasal bridge, a thin upper lip vermilion, and micrognathia. Brachydactyly of the fourth finger is noted on both hands. Neurological examination revealed that he was alert and well oriented. He showed no cranial nerve deficits and had no hemiparesis or ataxia. The muscle tone of his arms and legs was normal, as were his muscle stretch reflexes, and no pathological reflexes were elicited. His sensitivities to light touch, pain, temperature, vibration, and joint motion were also normal. The findings of his brain magnetic resonance imaging (MRI) and electroencephalography (EEG) were within normal limits. His fluent speech indicated a higher expressive language ability than his comprehension level. His good interpersonal communication ability suggested a social personality; however, he showed clumsiness when using tools including scissors and toothbrushes. He had difficulty with clothing. Cognitive assessments revealed a moderate intellectual disability that was associated with mild impairments in attention, performance, and visual-perceptual cognition. High-performance capability testing revealed difficulty in finger imitation, left and right alternating movement, and mouth-opening operations.

Patient 2. A 5-year-old, right-handed, Japanese boy was born at 40 weeks of gestation to healthy non-consanguineous parents with a weight of 3,090 g (50–75th centile), a length of 48.2 cm (25–50th centile), and an occipito-frontal circumference of 33.5 cm (mean). His Apgar score was nine at 1 min. There was no remarkable family history, and his older brother was healthy. He started to walk independently and speak meaningful words at 18 months, indicating a mild delay. For this reason, he had opportunities to attend special intervention training. At that time, he was thought to have insufficient muscle power and clumsiness in his fingers.

At present, his height is 108.7 cm (50–75th centile) and his weight is 18.8 kg (50–75th centile). He does not show any dysmorphism. His behaviors are performed at his own pace, and he rarely follows our suggestions. He also shows obsessive tendencies and mild attention impairments. His fine motor skills are poor, and he has difficulty with left-right alternating movements. He shows difficulty using scissors. No abnormalities were observed on brain MRI or EEG.

TABLE I. Summary of Clinical and Neuropsychological Examination

	Patient 1	Patient 2
Clinical information		
Gender	Male	Male
Age (years [y] months [m])[a]	16y	5y 3m
Neuropsychological features		
Individual pace	−	+
Obsessive tendencies	+	+
Mild attention impairment	+	+
Clumsiness	+	+
BDMR features	+	−
Neuropsychological examination		
WISC-III		
Full IQ	44	90
Verbal IQ	56	99
Performance IQ	44	82
DTVP		
Eye-Hand coordination	7y 6m	4y 4m
Figure ground	5y 11m	3y 8m
Form constancy	4y 0m	NP
Position in space	6y 6m	4y 7m
Spatial relations	6y 6m	4y 10m
DVMT-R		
Flexion fingers	3y 0m	>3y 8m
Extension finger	>5y 8m	4y 7m
Hands coordination	<4y 8m	4y 8m
Lips and cheeks	3y 6m	3y 6m
Tongue	>3y 10m	3y 10m
Speech	>5y 0m	5y 0m
Jumping	>4y 9m	>4y 9m
Balancing	<3y 9m	3y 9m
Bodily coordination	4y 2m	3y 0m
CARS	24.5	25.5

WISC-III, Wechsler Intelligence Scale for Children-III; DTVP, Developmental Test of Visual Perception; DVMT-R, Developmental Voluntary Movement Test-Revised; CARS, Childhood Autism Rating Scale;IQ, intelligent quotiate; y, years; m, months; NP, not performed.
[a]All neuropsychological examinations have been performed at this age.

Neuropsychological Examinations

The detailed data for the standard neuropsychological tests are shown in Table I.

Although patient 2 showed a normal intelligence quotient (IQ), patient 1 showed moderate intellectual disability with a full IQ of 44. Both patients showed a discrepancy between verbal IQ (VIQ) and performance IQ (PIQ); the VIQ was higher than the PIQ in both. All of the results on the subsets of WISC-III are depicted in Figure 3. The patterns of the subset scores were quite similar to each other (Fig. 3). In particular, lower scores were common in the similarities and arithmetic of the verbal subsets and in the coding and picture arrangement of the performance subsets.

On the DTVP, both patients had low scores for finding hidden figures. Similarly, both patients also had low scores for movements of the lips and cheeks and body coordination on the DVMT-R. The CARS scores were similar in both patients and were not indicative of autism.

DISCUSSION

In this study, we identified 2q37.3 deletions in two unrelated male patients. One of the patients (patient 1) showed typical manifestations of BDMR with brachydactyly, moderate intellectual disability, and facial dysmorphism. The 2q37.3 deletion that was identified in this patient was 3.2 Mb in size and included *HDAC4* (Fig. 1, Supplemental Table SI). This was in accordance with the fact that his BDMR phenotype indicated *HDAC4* involvement, and haploinsufficiency of *HDAC4* has been recognized to be responsible for BDMR [Williams et al., 2010; Morris et al., 2012; Villavicencio-Lorini et al., 2013].

Williams et al. have suggested that the phenotypic features of a subject with a 2q37 deletion consist of a variety of findings that overlap with other syndromes, including Smith-Magenis syndrome (SMS) [Williams et al., 2010]. Patients with SMS show a variety of congenital anomalies, intellectual disabilities, and behavioral abnormalities. The retinoic acid-induced 1 gene (*RAI1*) that is located

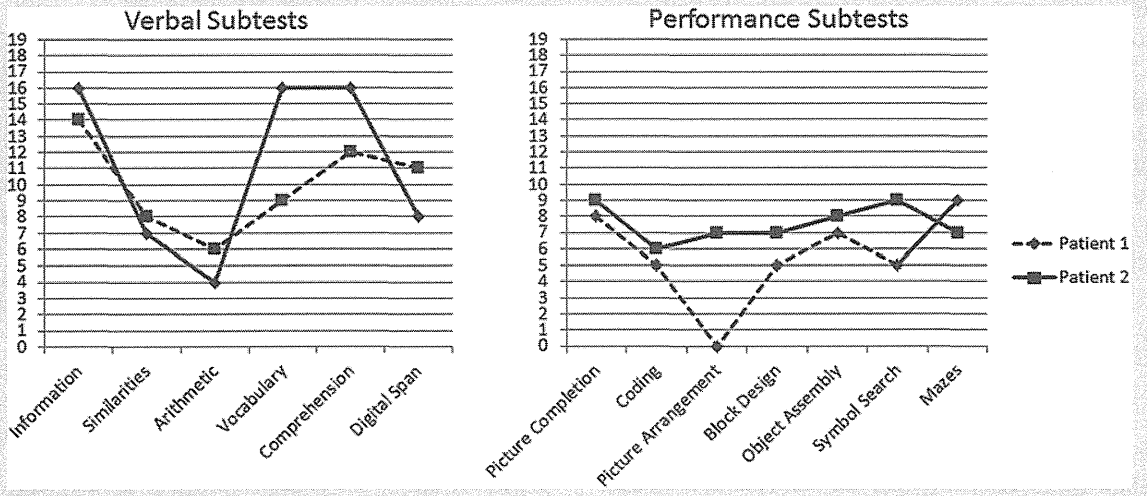

FIG. 3. The results of all of the subset scores in the WISC-III. Both patients show similar patterns in the subset results.

in the 17p11.2 deletion region is the gene that is responsible for SMS. Because *RAI1* expression is reduced in patients with *HDAC4* deletions and mutations, there is an expected interaction between *HDAC4* and *RAI1* [Williams et al., 2010]. Thus far, no findings have contradicted the notion that the behavioral abnormalities in patients with 2q27 deletions are derived from *HDAC4* haploinsufficiency.

In this study, we identified an additional patient (patient 2) with a 2q37.3 deletion who showed no BDMR phenotypes and no intellectual disability. This can be easily explained by the fact that the identified smaller deletion did not include *HDAC4* (Fig. 1). However, patient 2 showed some overlapping symptoms with patient 1, including obsessive tendencies, mild attention impairments, and clumsiness. Because there have been several reports on an association between 2q37 deletions and autistic behaviors [Ghaziuddin and Burmeister, 1999; Wolff et al., 2002; Lukusa et al., 2004; Galasso et al., 2008; Mazzone et al., 2012], we suspected that our patients might also be on the autism spectrum. Therefore, we performed neuropsychological examinations and compared the results of these two patients.

Although patient 2 did not show intellectual disability (IQ = 90), he exhibited a significant difference between his VIQ (99) and his PIQ (82). This pattern is the same in patient 1 who had a VIQ = 56 and a PIQ = 44. In the verbal subsets, both of the patients showed weakness in the similarities and arithmetic. In the performance subsets, both patients showed weakness in coding and picture arrangement. From these patterns, deficits in visual-motor skills were suspected. The DTVP and DVMT-R results also showed similar patterns. Both patients showed weakness in imitations of body movement, indicating disabilities of visuospatial perception. Based on these results, we suspected developmental dyspraxia [Dewey, 1995]. The CARS scores (24.5 and 25.5 in patient 1 and 2, respectively) were not suggestive of autism. Although intellectual disability was identified only in patient 1, the other neuropsychological characteristics were similar. Therefore, the intellectual disability that was observed in patient 1 can be explained by *HDAC4* involvement, but the other common neuropsychological abnormalities cannot because patient 2 did not show *HDAC4* involvement.

Williams et al., 2010 have described a similar patient who had a smaller deletion (#2282) and who showed autistic behavior [Williams et al., 2010]. However, the authors concluded that these phenotypic features were coincidental. Leroy et al., 2013 have also described a similar patient whose deletion did not include *HDAC4* but who had a duplication that included *HDAC4* [Leroy et al., 2013]. They suggested an alternative explanation that the other genes that are located in the telomeric region from *HDAC4* may be related to autistic behavior. Unfortunately, detailed neuropsychological examinations have never been performed in these patients. Therefore, an accumulation of the results of such examinations would disclose whether the telomeric region of 2q37 is related to autistic behaviors.

Missense mutations of the kinesin family member 1 A gene (*KIF1A*) have been revealed as the causative mutations for autosomal recessive spastic paraplegia [Erlich et al., 2011;Klebe et al., 2012]. Later, a de novo mutation in *KIF1A* was reported in a patient with nonsyndromic intellectual disability (MIM 601255) [Hamdan et al., 2011]. This indicated that a *KIF1A* haploinsufficiency could cause some neurological impairments, although behavior abnormalities were not described in those reports. The serine/threonine kinase 25 gene (*STK25*) is a genetic modifier of tau phosphorylation [Matsuki et al., 2012]. Because depletion of this molecule inhibits axon specification [Matsuki et al., 2010], a *STK25* haploinsufficiency may contribute to the behavioral abnormalities that have been observed in patients with 2q37.3 deletions [Shrimpton et al., 2004]. The glypican 1 gene (*GPC1*) has been reported as a good candidate gene for BDMR [Syrrou et al., 2002; Shrimpton et al., 2004; Chaabouni et al., 2006]. Recently, it has been reported that GPC1 controls brain size by

regulating fibroblast growth factor signaling in early neurogenesis [Jen et al., 2009], and there have been some reports that have described the contributions of GPC1 in neuronal functions [Abaskharoun et al., 2010; Wilson and Stoeckli, 2013].

The D-2-hydroxyglutarate dehydrogenase gene (*D2HGDH*) is involved in D-2-hydroxyglutaric aciduria (MIM 600721), which is a neurometabolic disorder that is characterized by developmental delays, epilepsy, hypotonia, and dysmorphic features. However, this disease is caused by an autosomal recessive trait [Struys et al., 2005]. Felder et al., 2009 have confirmed a downregulation of the FERM, RhoGEF, and pleckstrin domain protein 2 gene (*FARP2*); the high-density lipoprotein binding protein gene (*HDLBP*); and the PAS domain containing serine/threonine kinase gene (*PASK*) in the lymphoblastoid cell lines that were derived from a patient with a 2q37 deletion [Felder et al., 2009]. These three genes were selected as candidates because of their structural and functional relationships to the pathways that are involved in neuronal and/or skeletal development, and they have been shown to be considerably downregulated in a patient with a 2q37 deletion [Felder et al., 2009]. Because PASK is required for axonal ensheathment [Leiserson et al., 2000], deletion of this gene may be related to neuronal impairments in patients with 2q37 deletions.

As mentioned above, there are several genes that contribute to neuronal functions, and haploinsufficiencies of these genes may be related to the developmental dyspraxia or autistic features that are observed in patients with 2q37.3 deletions. Therefore, the identification of more patients with 2q37.3 deletions and detailed genotype-phenotype correlations should help to determine the gene(s) that are responsible for neuronal dysfunctions.

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