Tsurusaki Y, *Saitoh S, Tomizawa K, Sudo A, Asahina N, Shiraishi H, Ito J, Tanaka H, Doi H, Saitsu H, Miyake N, *Matsumoto N (* denotes co-correponding)	A DYNC1H1 mutation causes a dominant spinal muscular atrophy with lower extremity predominance	Neurogenet			in press
Yoneda Y, Haginoya K, Kato M, Osaka H, Yokochi K, Arai H, Kakita A, Yamamoto T, Otsuki Y, Shimizu S, Wada T, Koyama N, Mino Y, Kondo N, Takahashi S, Hirabayashi S, Takanashi J, Okumura A, Kumagai T, Hirai S, Nabetani M, Saitoh S, Hattori F, Yamazaki A, Subo Y, Nishiyama K, Miyatake S, Tsurusaki Y, Doi H, Miyake N, Matsumoto N, *Saitsu H	Phenotype spectrum of COL4A1 mutations: porencephaly to schizencephaly	Ann Neurol			in press
Miyatake S, Murakami A, Okamoto N, Miyake N, Saitsu H, *Matsumoto N	A De Novo Deletion at 16q24.3 Involving ANKRD11 in a Japanese Patient With KBG Syndrome	Am J Med Genet Part A			in press
Miyatake S, Touho H, Miyake N, Ohba C, Doi H, Matsumoto N	Sibling cases of Moyamoya disease with different RNF213 genotypes and varying clinical course and severity	J Hum Genet			in press
Tsurusaki Y, Kobayashi Y, Hisano M, Ito S, Doi H, Nakashima M, Saitsu H, Matsumoto N, *Miyake N	The diagnostic utility of exome sequencing in Joubert syndrome related disorders	J Hum Genet			in press

Higashiyama Y, *Doi H, Wakabayashi M, Tsurusaki Y, Miyake N, Saitsu H, Ohba C, Fukai R, Miyatake S, Koyano S, Suzuki Y, Kuroiwa Y, Matsumoto N	A novel homozygous SCARB2 mutation causes late-onset progressive myoclonus epilepsy without renal failure	Mov disord			in press
*Miyake N, Mizuno S, Okamoto N, Ohashi H, Shiina M, Ogata K, Tsurusaki Y, Nakashima M, Saitsu H, *Matsumoto N (*: co-corresponding)	KDM6A point mutations cause Kabuki syndrome	Hum Mut	34 (1)	108-110	2012
Kimura-Ohba S, Kagitani-Shimono K, Hashimoto N, Nabatame S, Okinaga T, Murakami A, Miyake N, Matsumoto N, Osaka H, Hojo K, Tomita R, Taniike M, *Ozono K	A case of cerebral hypomyelination with spondylo-epi-metaphyseal dysplasia	Am J Med Genet Part A			in press
*Miyake N#, Yano S# (# denotes equal contribution), Sakai C, Hatakeyama H, Shiina M, Watanabe Y, Bartley J, Abdenur JE, Wang RY, Chang R, Tsurusaki Y, Doi H, Saitsu H, Ogata K, Goto Y, *Matsumoto N	Mitochondrial complex III deficiency caused by a homozygous UQCRC2 mutation presenting with neonatal-onset recurrent metabolic decompensation	Hum Mut			in press
Ken-ichi Matsumoto	Phosphorylation of extracellular matrix tenascin-X detected by differential mass tagging followed by nanoLC-MALDI-TOF/TOF-MS/MS using ProteinPilot software	*Connect. Tissue Res.*	53	106-116	2012

Ken-ichi Matsumoto, Tomoko Maniwa, Tetsuya Tanaka, Kazumi Satoh, Hideki Okunishi, and Teiji Oda	Proteomic analysis of calcified abdominal and thoracic aortic aneurysms	*Int. J. Mol. Med.*	30	417-429	2012
Teiji Oda, Kouji Shimizu, Akane Yamaguchi, Kazumi Satoh, and Ken-ichi Matsumoto	Hypothermia produces rat liver proteomic changes as in hibernating mammals but decreases endoplasmic reticulum chaperones	*Cryobiology*	65	104-112	2012
Ken-ichi Matsumoto, Kazumi Satoh, Tomoko Maniwa, Asuka Araki, Riruke Maruyama, and Teiji Oda	Noticeable decreased expression of tenascin-X in calcific aortic valves	*Connect. Tissue Res.*	53	460-468	2012
松本健一	エーラス・ダンロス症候群の原因遺伝子の一つのテネイシンX	臨床化学	42	22-29	2013
松本健一	大動脈弁石灰化と細胞外マトリックス・テネイシンX	Vascular Medicine	9	印刷中	2013

知的財産権の出願・登録状況

PCT/JP2012/83113 松本直通／鶴崎美徳／三宅紀子・コフィンーシリス症候群の検出方法
・平成 24 年 12 月 20 日

特願 2012-180356・松本直通／三宅紀子・ミトコンドリア複合体 III 欠乏症の確定診断法
・平成 24 年 8 月 16 日

特願 2012-136 松本直通／鶴崎美徳／三宅紀子・コフィンーシリス症候群の検出方法
・平成 24 年 1 月 4 日