松本直通

[原著]

1. Okada I[#], Hamanoue H[#], ([#] denotes equal contribution) Terada K, Tohma T, Megarbane A, Chouery E, Abou-Ghoch J, Jalkh N, Cogulu O, Ozkinay F, Horie K, Takeda J, Furuichi T, Ikegawa S, Kiyomi Nishiyama K, Miyatake S, Nishimura A, Mizuguchi T, Niikawa N, Hirahara F, Kaname T, Yoshiura K-i, Tsurusaki Y, Doi H, Miyake N, Furukawa T, *Matsumoto N, *Saitsu H. *SMOC1* is essential for ocular and limb development in humans and mice**.** Am J Hum Genet 88(1): 30-41, 2011
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[著書・総説]

1. 土井宏・松本直通　常染色体劣性遺伝性疾患①遺伝形式と発症機序　遺伝子医学 MOOK 別冊「遺伝カウンセリングハンドブック」メディカルドウ社 p.88-89、2011
2. 鶴崎美徳・松本直通　研究から医療へ　遺伝子医学 MOOK 別冊「遺伝カウンセリングハンドブック」メディカルドウ社 p.274-275, 2011
3. 近藤有希子・松本直通　Beals 症候群　「症候群ハンドブック」、井村裕夫、辻省二、福井次矢編 p663, 2011
4. 村上翠・三宅紀子・松本直通　自閉症におけるコピー数異常研究　医学のあゆみ 239(6), 2011

[国際学会]

1. Matsumoto N, Megarbane A, Cogulu O, Tohma T, Okada I, Hmanoue H, Saitsu H. SMOC1 is essential for ocular and limb development in humans and mice. EHGC2011 May 28-31 at Amsterdam, The Netherland (RAI convention center)

[国内学会]

なし

[招聘講演・セミナー]

1. Naomichi Matsumoto “Identification of two epilepsy-related genes from a 2.25-Mb microdeletion in one patient” (Invited lecture at Department of Human Genetics, Leiden University, Leiden, The Netherland, May 26, 2011)
2. 大阪難症例脳血管疾患研究会・大阪もやもや病研究会（大阪・千里阪急ホテル 6 月 18 日）松本直通（特別講演）「もやもや病の遺伝学：最近わかってきたこと」

3. 講演会「次世代シーケンサーを用いた最先端研究」・松本直通「次世代シーケンサーを用いたヒト疾患ゲノム解析法」（徳島・徳島大学医学部臨床第一講堂 8 月 26 日）
4. 第一回サイトジェノミクスセミナー・松本直通「次世代シーケンス法による疾患研究の最前線」（三菱化学メディエンス志村事業所・東京 9 月 17 日）
5. 第 46 回産婦人科研究会（順天堂大学）松本直通「次世代シーケンサーを用いた疾患ゲノム解析の現状」（順天堂大学医学部・東京 9 月 20 日）
6. 第 18 回遺伝性疾患に関する出生前診断研究会　松本直通「次世代シーケンサーを用いた疾患ゲノム解析の現状」（佐賀大学医学部・佐賀 10 月 1 日）
7. 日本人類遺伝学会第 56 回大会　松本直通「ヒト遺伝性疾患の原因解明を目指して」学会賞受賞講演（於・幕張メッセ 11 月 11 日）
8. 日本人類遺伝学会第 56 回大会　松本直通「次世代シーケンサーを用いたヒト疾患ゲノム解析法」(シンポジスト)シンポジウム 11(超高速シークエンサーによる疾患ゲノム解析)（於・幕張メッセ 11 月 12 日）
9. 国立精神・神経医療研究センターTMC 棟／クラスター研究棟開棟記念講演会「遺伝性神経疾患のエクソーム解析」松本直通（招待講演）（国立精神・神経医療研究センター11 月 22 日）
10. The 34th annual meeting of the molecular biology society of Japan・Next generation sequencing technology enables a large scale medical genomic research (symposium)「Disease genome analysis using next generation sequencer」Naomichi Matsumoto (Invited speaker)(Dec 14, 2011 at Yokohama, Japan)

[その他]

新聞・マスコミ発表

2011 年 9 月 15 日　日刊工業新聞「常染色体劣性小脳変性症原遺伝子を発見」

2011 年 10 月 28 日　日経バイオテク ONLINE アカデミック版「横浜市大の松本直通教授ら、先天性大脳白質形成不全症の原因遺伝子を発見」

2011 年 10 月 31 日 Yahoo! Japan ニュース「横浜市立大、「先天性白質形成不全症」の 1 種「HCAHC」の原因遺伝子を特定」

特許出願状況

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

特願 2011-247457 才津浩智／松本直通・孔脳症および周産期脳出血の検出方法・平成 23 年 11 月 11 日

特願 2011-226488・才津浩智／松本直通・び慢性大脳白質形成不全症の検出方法・平成 23 年 10 月 14 日

特願 2011-136277・松本直通／土井宏・常染色体劣性遺伝性脊髄小脳変性症の検出方法・横浜市立大学・平成 23 年 6 月 20 日