（報告様式４）

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I. 基本情報

事　業　名：（日本語）難治性疾患実用化研究事業
（英　語）Practical Research Project for Rare/Intractable diseases

研究開発課題名：（日本語）遺伝性難治疾患の網羅的遺伝子解析拠点研究
（英　語）NGS center for rare genetic diseases

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実 施 期 間：平成２６年６月２日　～　平成２９年３月３１日

分担研究開発課題名：（日本語）遺伝性難治疾患の網羅的遺伝子解析拠点研究
（英　語）NGS center for rare genetic diseases

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分担研究開発課題名：（日本語）ゲノム研究倫理と網羅的エクソーム解析の対象症例の集積
（英　語）Implication of genomics research and selection of appropriate patients for whole exome sequence

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分担研究　　（日本語）次世代シーケンサーのデータ解析研究
開発課題名：　（英　語）Research of Data Analysis for Next Generation Sequencer

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II. 成果の概要（総括研究報告）

・ 研究開発代表者による報告の場合

和文

I. 希少遺伝性難病の新規遺伝子異常同定

様々な原因不明の遺伝性希少難病を全国の研究協力者および難治性疾患等克服研究事業（難治性疾患等実用化研究事業（難治性疾患実用化研究事業)）の他の拠点班と各連携研究班、あるいは共同研究の要請に応じて諸外国の病院等から症例検体を集積し全遺伝子を対象とした全エクソーム解析を中心とした解析を行い、原因となる遺伝子異常の特定を進めた。3 年間で 6483 サンプルのエクソーム解析を行った。遺伝学的解析結果から、新規の責任遺伝子を 15 疾患で確立した。Coffin-Siris 症候群（SOX11, Nat Commun 2014）, Leigh 脳症（GYG2, Hum Genet 2014)、グリコシル化異常症（COG2, Clin Genet 2015)、限局性皮質形成異常症（MTOR の体細胞変異、Ann Neurol 2015)、ステロイド抵抗性ネフローゼ症候群（NUP107, Am J Hum Genet 2015)、脊椎硬膜外くも膜嚢腫（HOXD4, Plos One 2015)、軸性脊椎骨幹端異形成症（C12orf65, Plos One 2016)、視床下部過誤腫（OFD1 と GLI3 の体細胞変異、Ann Clin Transl Neurol 2016)、先天性糖鎖異常症（PIGG, Am J Hum Genet 2016)、小児早期神経変性脳症（TBCD, Am J Hum Genet 2016)、視神経萎縮を伴う難治性てんかん性脳症（AP3P2, Am J Hum Genet 2016)、MIRAGE 症候群（SAMD9, Nat Genet 2016)、橋小脳低形成（TOE1, Nat Genet 2017)、ネマリンミオパチー（MYPD, Am J Hum Genet 2017)、Weaver 症候群（SUZ12, Hum Mut 2017）である。論文は 2014 年に 42 本、2015 年に 37 本、2016 年以降には 67 本が印刷された(計 146 本)。
拠点としてのインフォーマティクス解析を強化するため、解析サーバーを新たに導入した。さらに遺伝子変異同定率を向上させるためインフォーマティクスプロトコールの改良や次世代シーケンス解析の手法についても検討を加えた。全ゲノム解析において Short read sequencing 技術で補えない CNV 検出法の開発を目的とした long read sequencing 技術を導入し、解析系の確立を進めた。

II. 難病に関連する遺伝子診断

難治性疾患等克服研究事業（難治性疾患等実用化研究事業（難治性疾患実用化研究事業)）の他の拠点班と連携・分担し、難病症例の遺伝子診断の体制づくりと診断業務を協力して進めた。遺伝子診断結果の返却に関しては、診断委員会を得て研究室内の合意（診断委員会での了承）を得て適切な手続きを踏んだ上で診断書を郵送する手順で進めた。本研究の遺伝学的解析により 2014 年～2016 年の 3 年で独立した（家族例でない）2815 例中 1182 例（42.0%)が確定診断となり、臨床現場における診断に大きく貢献した。

英文

I. Identification of novel mutated genes for rare genetic diseases

Through broad collaborations with clinical doctors and Practical Research Projects for Rare/Intractable diseases as well as oversea's clinicians, we analyzed a total of 6483 exomes in these three years (fiscal 2014~2016). Through this project, we could successfully find novel culprit genes mutated in a total of 15 different diseases as follows.

2014 Leigh syndrome: GYG2 (Hum Genet)
Coffin-Siris syndrome: SOX11 (Nat Commun)
2015 Glycosylation disorder: COG2 (Clin Genet)
FCD Type IIb: MTOR (somatic) (Ann Neurol)
Steroid-registant nephrotic syndrome: NUP107 (Am J Hum Genet)
Spinal extradural arachnoid cyst: HOXD4 (Plos One)
Axial spondylometaphyseal dysplaia: C21orf2 (Plos One)
Hypothalamic Hamartoma: OFD1 & GLI3 (somatic) (Annals of Clinical and Translational Neurology)
2016 Congenital glycosylation disorder: PIGG (Am J Hum Genet)
MIRAGE syndrome: SAMD9 (Nat Genet)
Progressive neurodegeneration: TBCD (Am J Hum Genet)
EOEE with optic atrophy: AP3P2 (Am J Hum Genet)
2017 Nemaline myppathy: MYPN (Am J Hum Genet)
Pontocerebellar Hypoplasia 7: TOE1 (Nat Genet)
Weaver syndrome: SUZ12 (Hum Mut)

We have published a total of 146 scientific papers: 42 (2014), 37 (2015) and 67 (2016~).
To strengthen informatics analysis power, we newly installed servers. Using these, we improved analytical protocols. Furthermore, we also newly installed a long-read sequencer (Sequel) for improved detection of relatively small-sized CNVs which short-read sequencers may easily miss.

II. Genetic diagnosis of rare/intractable diseases

Through collaboration with Practical Research Projects for Rare/Intractable diseases, we have done genetic diagnosis in 1182 out of 2815 independent cases. Diagnostic rate was 42.0%.

・ 研究開発分担者による報告の場合
研究開発代表者：公立大学法人横浜市立大学・医学研究科遺伝学・松本直通　総括研究報告を参照。

III. 成果の外部への発表

（１）学会誌・雑誌等における論文一覧（国内誌 10 件、国際誌 167 件）

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（2）学会・シンポジウム等における口頭・ポスター発表

松本直通

1 第 37 回日本小児遺伝学会学術集会・シンポジウム・松本直通「次世代シーケンサー解析のポテンシャルと小児医療」平成 26 年 4 月 10 日名古屋市立大学桜山キャンパス・名古屋

2 The 9th Cherry Blossom Symposium. Symposium 4 “Clinical testing and automation utilizing new technology”Naomichi Matsumoto “Next generation sequencing dissecting human genetic diseases”Yokohama Red Brick Warehouse No. 1, April 19, 2014

3 第 382 回医学研究の基礎を語り合う集い・松本直通「遺伝性疾患におけるエクソーム解析の有用性」平成 26 年 4 月 21 日東京慈恵医科大学

4 九州大学産婦人科セミナー・松本直通「ゲノム解析と医療：1000 ドルゲノム時代を迎えて」平成 26 年 5 月 21 日第二博多偕成ビルアイクレオ

5 信州大学先鋭領域融合研究群バイオメディカル研究所設立記念講演会・記念講演会・松本直通「ゲノム解析と医療：1000 ドルゲノムシーケンス時代を迎えて」平成 26 年 5 月 25 日・信州大学医学部・松本

6 第 55 回日本神経病理学会総会学術集会・松本直通（シンポジスト）「WDR45 mutations cause static encephalopathy of childhood with neurodegeneration in adulthood」@シンポジウム 1 （English）「Neurodegeneration with Brain Iron Accumulation (NBIA): 急速に広がる疾患概念」平成 26 年 6 月 7 日学術総合センター

7 藤田保健衛生大学遺伝子網羅的解析センター開所記念セミナー・松本直通　特別講演「ゲノム解析と医療：1000 ドルゲノム時代を迎えて」平成 26 年 6 月 21 日@藤田保健衛生大学・豊橋

8 第 110 回日本精神神経学会学術総会・松本直通（シンポジスト）「発達障害のゲノム解析」シンポジウム 50「精神疾患における分子と臨床の架け橋～病因と治療」2014 年 6 月 27 日パシフィコ横浜、横浜

9 がん研臨床試験・研究センター　第 12 回臨床研究セミナー松本直通「遺伝性疾患のゲノム解析---1000 ドルゲノム時代を迎えて---」2014 年 7 月 3 日がん研究会・東京

10 文部科学省イノベーションシステム整備事業先端融合領域イノベーション創出拠点形成プログラム「翻訳後修飾プロテオミクす医療研究拠点の形成」第 5 回公開シンポジウム・松本直通「ヒト発生・発達異常の分子探索と診断法の開発」2014 年 8 月 19 日・県民共済みなとみらいホール・横浜

11 KOBE 内分泌・代謝スキルアップセミナー特別講演・松本直通・「ゲノム解析と医療--1000 ドルゲノム時代を迎えて--」平成 26 年 8 月 23 日＠ホテルクラウンパレス神戸

12 第 17 回胎児遺伝子診断研究会・セミナー１・松本直通「次世代シーケンサーの活用法」平成 26 年 11 月 7 日＠ハウステンボス・佐世保

13 第 56 回日本先天代謝異常学会総会・松本直通・シンポジスト・「次世代シーケンサーによる遺伝子解析」平成 26 年 11 月 14 日＠江陽グランドホテル・仙台

14 日本人類遺伝学会第 59 回大会・松本直通・シンポジスト「次世代シーケンサーを用いた染色体・ゲノム構造異常解析」平成 26 年 11 月 21 日＠タワーホール船堀・東京

15 日本人類遺伝学会第 59 回大会・松本直通・NPG ランチョンセミナー「人類遺伝学研究におけるデータベースの重要性」平成 26 年 11 月 22 日＠タワーホール船堀・東京

16 The 37th annual meeting of the molecular biology society of Japan. Matsumoto N (as a symposist).“Congenital intellectual disability syndromes arising from mutations of epigenetic regulators”in the symposium for epigenetic dysregulation and disease. Pacifico Yokohama, Yokohama, Nov 27, 2014.

17 日本遺伝子診療学会：遺伝子診断・検査技術推進フォーラム公開シンポジウム 2014「個別化医療を進めるために」ランチョンセミナー・松本直通「臨床に役立つ次世代シーケンサーの活用法」・平成 26 年 12 月 12 日・コクヨホール（品川）

18 順天堂大学医学部神経学講演会・松本直通「疾患解析における次世代シーケンサーの活用法」平成 27 年 1 月 16 日・順天堂大学

19 International symposium on genome science 2015, “Expanding Frontiers of Genome Science II”. Matsumoto N (Invited speaker). “De novo mutations cause early onset epileptic encephalopathy” @ National Center of Sciences, Tokyo, Jan 20, 2015

20 群馬成育医療 UPDATE2015-遺伝と医療を考える-松本直通「遺伝性疾患解析の現状」平成 27 年 1 月 28 日・群馬大学＠前橋

21 平成 26 年度厚生労働科学研究障害者対策総合研究事業（神経・筋疾患分野）研究成果等普及啓発事業研究成果発表会（研究者向け）・松本直通「統合的遺伝子解析システムを用いたヒト発達障害研究」・平成 27 年 2 月 5 日＠八重洲ホール・東京

22 第 55 回神奈川小児神経懇話会・特別講演・松本直通「発達障害の遺伝学的研究」2015 年 2 月 7 日ホテルキャメロットジャパン・横浜

23 平成 26 年度厚生労働科学研究費委託費事業（難治性疾患実用化研究）・成果報告会・松本直通「遺伝性難治疾患の網羅的遺伝子解析拠点研究」2015 年 3 月 13 日於東京コンベンションホール・東京

24 愛知学院大学先天異常遺伝学・言語学講座・講演会・松本直通「遺伝性疾患の次世代シーケンサー解析」・平成 27 年 3 月 14 日愛知学院大学楠元学舎＠名古屋

25 産総研 Computational Biology Research Center (CBRC)セミナー・松本直通「遺伝性疾患の次世代シーケンス解析」@産総研・臨海副都心センター（別館）平成 27 年 4 月 23 日

26 European Human Genetics Conference 2015・Naomichi Matsumoto, Eriko Koshimizu, Satoko Miyatake, “Deep sequencing detects very low-grade somatic mosaicism in the unaffected mother of siblings with nemaline myopathy”(poster), @ Glasgow, Scotland, United Kingdom, June 6-9, 2017

27 2015 アジレントゲノミクスフォーラム・招聘講演・松本直通「全エクソーム解析データの様々な活用法」@ヒューリックホール浅草橋・東京・平成 27 年 6 月 16 日

28 第 33 回内分泌代謝学サマーセミナー・続・内分泌至上主義・松本直通（シンポジスト（招聘））「ヒト疾患と rare variants」@柳川藩主立花邸・柳川（福岡）・平成 27 年 7 月 9 日

29 第 22 回日本遺伝子診療学会大会・プレシジョン医療と診断情報の質保証（シンポジウム 3）松本直通「遺伝性疾患における話題提供」@かながわ労働プラザ・横浜・平成 27 年 7 月 19 日

30 浜松医科大学大学院特別講演・松本直通「ヒト疾患と rare variants」@浜松医科大学・浜松・平成 27 年 9 月 11 日

31 日本人類遺伝学会第 60 回大会・シンポジウム 9 「ヒト疾患に関わる体細胞モザイク変異とその検出法」・松本直通「低頻度体細胞モザイク変異が惹起するヒト疾患」（シンポジスト）@京王プラザホテル・東京・平成 27 年 10 月 16 日

32 The VI Croatian Congress of Human Genetics, Naomichi Matsumoto, “Next Generation Sequencing Dissecting Human “Genetic” Diseases” (invited) Nov 6, 2015@Hotel President Split, Split, Croatia.

33 Invited lecture, Naomichi Matsumoto, “Rare variants in human diseases” (invited) Nov 9, 2015@University of Split, Split, Croatia.

34 Biochemistry and Molecular Biology (BMB) 2015, Workshop, Naomichi Matsumoto, “Somatic mutations in the MTOR gene cause focal cortical dysplasia type IIb” Dec 1, 2015@Kobe portpia hotel, Kobe.

35 第 38 回日本分子生物学会年会(第 88 回日本生化学会大会)BMB2015, ランチョンセミナー, 松本直通（講師）“遺伝子解析におけるドロップレットディジタル PCR の活用”@神戸ポートピアホテル・神戸・平成 27 年 12 月 1 日

36 第 23 回食細胞機能異常症研究会、特別講演・松本直通「ヒト疾患と rare variants」@東京慈恵医科大学・東京・平成 27 年 12 月 12 日

37 第 1 回日本産婦人科遺伝診療学会学術講演会・教育講演・松本直通「最先端テクノロジーによる疾患ゲノム解析」H27 年 12 月 18 日@長崎ブリックホール・長崎

38 平成 27 年度 IRUD-A 研究班班会議・松本直通「成人未診断疾患のゲノム解析」H28 年 1 月 21 日・@シェーンバッハサボー・東京

39 平成 28 年横浜市立大学医学部呼吸器病学勉強会・松本直通「ヒト疾患と rare variants」H28 年 2 月 15 日@横浜市立大学医学部・横浜

40 第 35 回小児成長研究会・松本直通「ヒト疾患と rare rariants」H28 年 2 月 27 日・@霞山会館・東京

41 ゲノムテクノロジー第 164 委員会第 51 回研究会「ゲノム医療」講師・松本直通「難治性疾患の遺伝子解析の現状」H28 年 3 月 24 日@コンファレンススクウェアエムプラス・東京

42 International Congress of Human Genetics 2016 (ICHG2016) Concurrent Invited Session 21. Naomichi Matsumoto (Invited speaker) “Next Generation Sequencing Dissecting Human Genetic Diseases”. Apr 6, 2016@Kyoto International Conference Center, Kyoto, Japan.

43 The 11th Asian & Oceanian Epilepsy Congress (AOEC), Naomichi Matsumoto (Invited speaker) “Somatic mutation in Sturge Weber syndrome” May 16th, 2016 @ Hong Kong Convention & Exhibition Centre, Hong Kong

44 International Symposium on Genomic Medicine 2016, Naomichi Matsumoto (invited speaker) “Rare variants in human diseases” June 24, 2016@Samsung Medical Center, Seoul, Korea

45 The 22nd Annual Meeting of Japan Society of Gene and Cell Therapy (JSGCT), Naomichi Matsumoto (Education Session), “Rare variants in human diseases”. July 29, 2016 @ Toranomon Hills Forum, Tokyo

46 第 2 回日本筋学会学術集会・最新テクノロジーによる骨格筋研究の新展開・松本直通（シンポジスト）「Rare variants とヒト疾患」H28 年 8 月 5 日国立精神・神経研究センター・小平・東京

47 H28 年 9 月 9 日・Ingenuity ユーザーミーティング・松本直通「HGMD で加速するヒト遺伝子変異探索」@大手町サンケイプラザ・東京

48 H28 年 9 月 13 日・北海道大学臨床遺伝子診療部講演・松本直通「Rare variants とヒト疾患」@北海道大学病院・札幌

49 H28 年 10 月 8 日・第 50 回日本てんかん学会学術集会アドバンストコース 7／松本直通「エクソーム解析の進歩」@グランシップ・静岡

50 LMCE2016, Symposium 13: Applications to disease gene identification & diagnosis using NGS. Naomichi Matsumoto (Invited speaker), “Mendelian Exome in Japan”@The-K Hotel, Oct 28, 2016, Seoul, Korea

51 H28 年 11 月 1 日・東京大学医学部腎臓内分泌内科講演・松本直通「ヒト疾患と Rare Variants」東京大学附属病院・東京

52 A Lecture: Naomichi Matsumoto “Rare variants in human diseases” @ Central South University, Changsha, China on Nov 3, 2016

53 The 2016 Annual Meeting of The Chinese Society of Medical Genetics (CSMG), Keynote Lecture, Naomichi Matsumoto, “Rare variants in human diseases”. Hanghzou, China on Nov 6, 2016

54 The 4th International Conference on Rare and Undiagnosed Diseases, Naomichi Matsumoto, Invited speaker, “Rare variants in human diseases”@Gakushikaikan, Tokyo, Nov 16, 2016

55 2017 Educational Seminar of Genetic & Metabolic Diseases for Young Pediatricians. Naomichi Matsumoto (Invited Lecturer), “NGS dissecting pediatric neurological disorders” @Awards Inn Hankyu, Tokyo on Jan 22, 2017

56 H29 年 2 月 23 日・広島大学原爆放射線医科学研究所セミナー・松本直通「ヒト疾患における Rare variants 探索の現状と展望」@広島大学・広島

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該当なし

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