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MEETING ABSTRACTS AND CONFERENCE PROCEEDINGS

1. Csonka M, Schönholzer MT, Ferenczi S, Kovács KJ, Meienberg J, **Mátyás G** (2023) Dose-effect of celiprolol in mice modelling vEDS. ITINERARE 9th Rare Disease Summer School, Kartause Ittingen, Warth TG, July 4-7, 2023.
2. Csonka M, Schönholzer MT, Ferenczi S, Kovács KJ, Meienberg J, **Mátyás G** (2023) Dose-effect of celiprolol in mice modelling vEDS. ESHG Hybrid Conference, Glasgow, June 10-13, 2023.
3. Meienberg J, Caspar SM, **Mátyás G** (2023) Two in one: Pharmacogenetic profiling from whole genomes. ESHG Hybrid Conference, Glasgow, June 10-13, 2023.
4. Mroczek M, Mensova L, Meienberg J, Rejmer P, Parmova O, Henggeler C, **Mátyás G** (2023) Three heterozygous GAA cases mimicking late-onset Pompe disease. ESHG Hybrid Conference, Glasgow, June 10-13, 2023.
5. Tomio G, Saurer N, Meienberg J, **Mátyás G** (2023) The dark side of Moon and other commercial genome analysis or interpretation tools. ESHG Hybrid Conference, Glasgow, June 10-13, 2023.
6. Meienberg J*, Dubacher N, Münger J, Schönholzer MT, Ferenczi S, Kovács KJ, **Mátyás G** (2023) Added value of statins in the drug therapy of vascular Ehlers-Danlos syndrome. SGMG Annual Meeting with Young Investigator Day, Basel, April 27-28, 2023. *oral presentation
7. Meienberg J, Caspar SM, **Mátyás G** (2023) WGS-based personalized PGx profiling. SGMG Annual Meeting with Young Investigator Day, Basel, April 27-28, 2023.
8. Chittim N, Dubacher N, **Mátyás G**, Snedeker JG (2023) Tendinopathic changes to the murine patellar tendon in response to ciprofloxacin treatment. Orthopaedic Research Society Conference, Dallas, Texas, February 10-14, 2023.
9. Mroczek M, Meienberg J, Rejmer P, Henggeler C, Baumgartner MR, **Mátyás G** (2022) A case of atypical inheritance in late-onset Pompe disease. ASHG 2022, Los Angeles, October 25-29, 2022.
10. Dubacher N, Münger J, Ferenczi S, Kovács KJ, Schönholzer MT*, Meienberg J, **Mátyás G** (2022) Added value of statins in the treatment of vascular Ehlers-Danlos syndrome. International Scientific Symposium on the Ehlers-Danlos Syndromes and Hypermobility Spectrum Disorders, Rome, September 14-17, 2022. *oral presentation
11. Dubacher N, Sugiyama K, Smith JD, Nussbaumer V, Caspar SM, Schönholzer MT, Meienberg J*, Yanagisawa H, Sheppard MB, **Mátyás G** (2022) Mechanical integrity of the thoracic aorta in mice modelling vEDS and related aortic diseases. International Scientific Symposium on the Ehlers-Danlos Syndromes and Hypermobility Spectrum Disorders, Rome, September 14-17, 2022. *oral presentation
12. Stengl R, Dubacher N, Schönholzer MT, Ferenczi S, Meienberg J, Benke K, Szabolcs Z, **Mátyás G** (2022) Novel burst pressure read-out system to assess the biomechanical integrity of the murine aorta. International Scientific Symposium on the Ehlers-Danlos Syndromes and Hypermobility Spectrum Disorders, Rome, September 14-17, 2022.
13. Dubacher N, Sugiyama K, Smith JD, Nussbaumer V, Caspar SM, Schönholzer MT, Meienberg J*, Yanagisawa H, Sheppard MB, **Mátyás G** (2022) Insights into the biomechanical integrity of the aorta in mice modeling hereditary aortic diseases. Science in Paris featuring the VEDS Scientific Meeting, the International Symposium on Marfan Syndrome, LDS, and Related Conditions, and the GenTAC Aortic Summit, Paris, August 29 – September 1, 2022. *oral presentation
14. Meienberg J, Caspar SM, Stoll P, Fritzmann S, Gut G, Salerno D, **Mátyás G** (2022) ClinVar and HGMD variants in TAAD genes: You cannot interpret what you do not detect. Science in Paris featuring the VEDS Scientific Meeting, the International Symposium on Marfan Syndrome, LDS, and Related Conditions, and the GenTAC Aortic Summit, Paris, August 29 – September 1, 2022.
15. Schönholzer MT*, Dubacher N, Münger J, Ferenczi S, Kovács KJ, Meienberg J, **Mátyás G** (2022) Added value of statins in vascular Ehlers-Danlos syndrome. Science in Paris featuring the VEDS

Scientific Meeting, the International Symposium on Marfan Syndrome, LDS, and Related Conditions, and the GenTAC Aortic Summit, Paris, August 29 – September 1, 2022. *oral presentation

16. Stengl R*, Dubacher N, Schönholzer MT, Ferenczi S, Meienberg J, Benke K, Szabolcs Z, **Mátyás G** (2022) Novel assay to assess the aortic rupture of mice modeling aortic diseases. Science in Paris featuring the VEDS Scientific Meeting, the International Symposium on Marfan Syndrome, LDS, and Related Conditions, and the GenTAC Aortic Summit, Paris, August 29 – September 1, 2022. *oral presentation
17. Brühwiler B, Koller S, Baehr L, Rohrbach M, Steiner B, Baumer A, **Mátyás G**, Kivrak Pfiffner F, Berger W, Gerth-Kahlert C (2022) Genotype-phenotype spectrum in patients with novel variants in the ADAMTSL4 gene. SOG Jahreskongress 2022, Basel, August 24-26, 2022.
18. Caspar SM, Stoll P, Fritzmann S, Gut G, Salerno D, Meienberg J, **Mátyás G** (2022) Whole-genome sequencing: The long and the short of it. ESHG Hybrid Conference, Vienna, June 11-14, 2022.
19. Dubacher N, Sugiyama K, Smith JD, Nussbaumer V, Caspar SM, Schönholzer MT, Meienberg J, Yanagisawa H, Sheppard MB, **Mátyás G** (2022) Aortic rupture force in mice modelling hereditary aortic diseases. ESHG Hybrid Conference, Vienna, June 11-14, 2022.
20. Caspar SM, Stoll P, Fritzmann S, Gut G, Meienberg J, **Mátyás G** (2022) You cannot interpret what you do not detect: How not to miss clinically-relevant ClinVar and HGMD variants. SGMG Annual Meeting 2022 and Young Investigator Day, Online Venue, April 7-8, 2022.
21. Caspar SM, Stoll P, Meienberg J, **Mátyás G** (2021) Need for speed in whole-genome sequencing data analysis: Benchmarking the new generation of alignment and variant calling tools. ESHG Virtual Conference, August 28-31, 2021.
22. Dubacher N*, Caspar SM, Meienberg J, **Mátyás G** (2021) New uses for old drugs: Added value of celiprolol and pravastatin in vascular EDS. ESHG Virtual Conference, August 28-31, 2021. *oral presentation
23. Caspar SM, Stoll P, Meienberg J, **Mátyás G** (2021) Whole-genome sequencing data analysis: Performance of novel ultra-fast alignment and variant calling tools. 5th Curating the Clinical Genome, Virtual Conference, May 12-14, 2021.
24. Dubacher N*, Caspar SM, Meienberg J, Mátyás G (2021) Drug repositioning: Added value of celiprolol and pravastatin in vascular EDS. 4th Scientific Meeting on Vascular Ehlers-Danlos Syndrome, Virtual Meeting, April 9, 2021. *oral presentation
25. Caspar SM*, Najafi A, Meienberg J, **Mátyás G** (2021) Clinical whole-genome sequencing: Co-occurring rare diseases and pharmacogenetic profiling. The Virtual World Conference on Rare Diseases (RARE2021), February 22-23, 2021. *oral and poster presentation
26. Dubacher N, Caspar SM, Meienberg J, **Mátyás G** (2021) From diagnosis to therapy: Novel approach reveals celiprolol as medical therapy of choice for vascular Ehlers-Danlos syndrome. The Virtual World Conference on Rare Diseases (RARE2021), February 22-23, 2021.
27. Dubacher N, Münger J, Gorosabel MC, Crabb J, Ksiazek AA, Caspar SM, Bakker E NTP, van Bavel E, Ziegler U, Carrel T, Steinmann B, Zeisberger S, Meienberg J, **Mátyás G** (2021) From diagnosis to therapy: Novel approach reveals celiprolol but not losartan and bisoprolol as medical therapy of choice for vascular Ehlers-Danlos syndrome. RE(ACT) Congress and IRDiRC Conference 2021, International Congress on Research of Rare and Orphan Diseases, Virtual Meeting, January 13-15, 2021.
28. Caspar SM, Najafi A, Meienberg J, **Mátyás G** (2021) New insights into clinical whole-genome sequencing: co-occurring rare diseases and pharmacogenetic profiling. SGMG Annual Meeting 2021, Online Venue, January 21-22, 2021.
29. Dubacher N*, Münger J, Gorosabel MC, Meienberg J, **Mátyás G** (2021) Medical therapy of vascular Ehlers-Danlos syndrome: challenging the paradigm of interchangeable antihypertensive drugs.

SGMG Annual Meeting 2021, Online Venue, January 21-22, 2021. *oral presentation as finalist for the young investigator award.

30. Caspar SM*, Najafi A, Meienberg J, **Mátyás G** (2020) Clinical Whole-Genome Sequencing: Co-Occurring Fibrillinopathies and Pharmacogenetic Profiling. GenTAC Aortic Summit 2020 Virtual Conference, September 29, October 8-10, 2020. *oral presentation.
31. Dubacher N*, Münger J, Meienberg J, **Mátyás G** (2020) Novel Approach Reveals Celiprolol but not Losartan and Bisoprolol as Medical Therapy of Choice for Vascular Ehlers-Danlos Syndrome. GenTAC Aortic Summit 2020 Virtual Conference, September 29, October 8-10, 2020. *oral presentation.
32. Meienberg J, Caspar SM, Schneider T, **Mátyás G** (2020) Zusatznutzen der Ganzgenomsequenzierung: Pharmakogenetisches Profil. Careum Forum 2020, Zürich, 27. August, 2020.
33. Caspar SM, Najafi A, Meienberg J, Schneider T, Henggeler C, **Mátyás G** (2020) Added value of clinical whole-genome sequencing: Co-occurring rare diseases and pharmacogenetic profiling. ESHG 2020.2 Virtual Conference, June 6-9, 2020.
34. Dubacher N, Caspar SM, Meienberg J, **Mátyás G** (2020) Medical therapy of vascular Ehlers-Danlos syndrome: Challenging the paradigm of interchangeable antihypertensive drugs. ESHG 2020.2 Virtual Conference, June 6-9, 2020.
35. Caspar SM*, Niederberger D, **Mátyás G** (2020) Whole-Genome-Sequencing-Based Pharmaco-genetic Profiling for Precision Medicine. Life Science Zurich Impact Conference, Zurich, February 3, 2020. *oral presentation.
36. Caspar SM*, Niederberger D, **Mátyás G** (2019) Pharmacogenetics from Whole Genomes: At the Leading Edge of Precision Medicine Using the Medication Safety Card. Swiss Genomics Forum Geneve, September 27, 2019. *oral presentation.
37. Caspar SM*, Niederberger D, **Mátyás G** (2019) Whole-Genome-Sequencing-Based Pharmaco-genetic Profiling – Leading Edge of Precision Medicine. 15th Symposium of the ZIHP, Zurich, August 23, 2019. *oral presentation.
38. Dubacher N*, Münger J, Gorosabel MC, Crabb J, Ksiazek AA, Caspar SM, Bakker E NTP, van Bavel E, Ziegler U, Carrel T, Steinmann B, Zeisberger S, Meienberg J, **Mátyás G** (2019) Novel approach reveals celiprolol but not losartan as medical therapy for vascular Ehlers-Danlos syndrome − Lessons from a mouse model. ISACB + ISVTE 2019, Zurich, June 19-21, 2019. * Poster teaser.
39. Dubacher N, Münger J, Gorosabel MC, Crabb J, Ksiazek AA, Caspar SM, Bakker E NTP, van Bavel E, Ziegler U, Carrel T, Steinmann B, Zeisberger S, Meienberg J, **Mátyás G** (2019) Novel approach reveals celiprolol but not losartan as medical therapy for vascular Ehlers-Danlos syndrome. ESHG 2019, Gothenburg, June 15-18, 2019.
40. Caspar SM*, Meienberg J, Plüss M, Dubacher N, **Mátyás G** (2019) New Insights into the molecular diagnostics of rare (aortic) disorders in the current genomics era. SGMG General Assembly, Lausanne, April 4-5, 2019. *oral presentation.
41. Dubacher N*, Münger J, Gorosabel MC, Crabb J, Ksiazek AA, Caspar SM, Bakker E NTP, van Bavel E, Ziegler U, Carrel T, Steinmann B, Zeisberger S, Meienberg J, **Mátyás G** (2019) From diagnosis to therapy: novel approach reveals celiprolol but not losartan as medical therapy for vascular Ehlers-Danlos syndrome. SGMG General Assembly, Lausanne, April 4-5, 2019. *oral presentation.
42. Caspar SM, Najafi A, Dubacher N, Meienberg J, **Mátyás G** (2018) ExAC/gnomAD and the largest Swiss database of Marfan genomes reveal challenges in the diagnosis of fibrillinopathies. Central-Eastern European Marfan Symposium, Budapest, Hungary, October 26, 2018.
43. Meienberg J*, Dubacher N, Münger J, Gorosabel MC, Crabb J, Bakker E NTP, van Bavel E, Ziegler U, Zeisberger S, **Mátyás G** (2018) Candidate drug testing in a murine model of vascular Ehlers-

Danlos syndrome using biomechanical integrity as novel read-out. International Symposium of the Ehlers-Danlos Syndromes, Ghent, Belgium, September 26-29, 2018. *oral presentation.

44. Meienberg J, Najafi A, Caspar SM, Rohrbach M, Steinmann B, **Mátyás G** (2018) New Insights into Fibrillinopathies and Pitfalls of Variant Filtering in the Current Genomics Era. 6th International Meeting on Aortic Diseases, Liège, Belgium, September 12-14, 2018.
45. Dubacher N, Meienberg J, Münger J, Gorosabel MC, Crabb J, Bakker E NTP, van Bavel E, Ziegler U, Zeisberger S, **Mátyás G** (2018) Candidate drug testing in murine models of aortic diseases using biomechanical integrity as novel read-out. 6th International Meeting on Aortic Diseases, Liège, Belgium, September 12-14, 2018.
46. Dubacher N, Münger J, Meienberg J, Crabb J, Bakker E NTP, van Bavel E, Ziegler U, Zeisberger S, **Mátyás G** (2018). Paving the way for assessing the effect of candidate drugs on the mechanical integrity of the murine aorta: A hitherto unprecedented read-out system. 14th Symposium of the ZIHP, Zurich, August 31, 2018.
47. Dubacher N, Münger J, Meienberg J, Crabb J, Bakker E NTP, van Bavel E, Ziegler U, Zeisberger S, **Mátyás G** (2018).Novel read-out system paves the way for assessing the effect of candidate drugs on the mechanical integrity of the aorta in murine models. 6th Rare Diseases Summer School, Kartause Ittingen, Warth TG, July 11-13, 2018.
48. Caspar SM*, Meienberg J, Kopps AM, Plüss M, Dubacher N, **Mátyás G** (2018) Chances and challenges of high-throughput sequencing. Personalized Health Technologies and Translational Research Conference, Zurich, June 18-21, 2018. *oral presentation.
49. Najafi A, Caspar SM, Meienberg J, Rohrbach M, Steinmann B, **Mátyás G** (2018) Pitfalls of variant filtering in the current genomics era. Personalized Health Technologies and Translational Research Conference, Zurich, June 18-21, 2018.
50. Bors A, Kövy P, Stengl R, Benke K, Ágg B, Daradics N, **Mátyás G**, Szabolcs Z, Andrikovics H (2018) Genotype-phenotype correlations in Marfan syndrome for the prediction of severe cardiovascular manifestations. ESHG 2018, Milan, June 16-19, 2018.
51. Meienberg J, Kopps AM, Plüss M, Caspar SM, Dubacher N, **Mátyás G** (2018) High-throughput sequencing of Mendelian disorders: From raw data to diagnosis with lifetime value. ESHG 2018, Milan, June 16-19, 2018.
52. Bonassin Tempesta F, Attenhofer Jost C, Possner M, Meier L, Gruner C, Oxenius A, Seeliger T, **Mátyás G**, Rohrbach M, Pavicevic J, Greutmann M (2018) Mitral valve involvement in genetically confirmed connective tissue disease: data from a large Swiss cohort. Jahrestagung der Schweizerischen Gesellschaft für Kardiologie, Basel, June 6-8, 2018.
53. Caspar SM, Najafi A, Meienberg J, **Mátyás G** (2018) Pitfalls of gnomAD-frequency-based variant filtering in the current NGS era. Keystone Symposia on Molecular and Cellular Biology − One Million Genomes: From Discovery to Health, Hannover, June 4-8, 2018.
54. Dubacher N, Plüss M, Kopps AM, Keller I, Meienberg J, Caspar SM, Bruggmann R, Vogel M, **Mátyás G** (2018) High-throughput sequencing data analysis: full throttle! Keystone Symposia on Molecular and Cellular Biology − One Million Genomes: From Discovery to Health, Hannover, June 4-8, 2018.
55. Meienberg J, Kopps AM, Plüss M, Caspar SM, Dubacher N, **Mátyás G** (2018) Chances and challenges of high-throughput sequencing of Mendelian disorders. 3rd Curating the Clinical Genome, Wellcome Genome Campus, Cambridge, May 23-25, 2018.
56. Caspar SM, Najafi A, Meienberg J, **Mátyás G** (2018) New insights into fibrillinopathies in the current genomics era. 10th International Research Symposium on Marfan Syndrome and Related Disorders, Amsterdam, May 3-5, 2018.

57. Dubacher N, Münger J, Meienberg J, Crabb J, Bakker E NTP, van Bavel E, Ziegler U, Zeisberger S, **Mátyás G** (2018) Novel read-out system to assess the mechanical integrity of the thoracic aorta in murine models. 10th International Research Symposium on Marfan Syndrome and Related Disorders, Amsterdam, May 3-5, 2018.
58. Meienberg J, Kopps AM, Caspar SM, Plüss M, Dubacher N, **Mátyás G** (2018) Chances and challenges of high-throughput sequencing in genetic testing of Marfan syndrome and related disorders. 10th International Research Symposium on Marfan Syndrome and Related Disorders, Amsterdam, May 3-5, 2018.
59. **Mátyás G**, Odavic D, Henggeler C, Ntinopoulos V*, Dzemali O, Löblein H, Häussler A, Gruszczynski M, Zientara A, Genoni M (2018) Older patient age at replacement surgery of the ascending aorta does not exclude a hereditary aortopathy. 47. Jahrestagung der Deutschen Gesellschaft für Thorax-, Herz- und Gefässchirurgie (DGTHG), Leipzig, Februar 17-20, 2018. *oral presentation.
60. Caspar SM, Plüss M, Kopps AM, Dubacher N, Keller I, Meienberg J, Bruggmann R, Vogel M, **Mátyás G** (2017) Benchmarking of new ultra-fast genome-scale short-read alignment and variant calling pipelines. Target Validation using Genomics and Informatics Conference, Wellcome Genome Campus, Cambridge, December 6-8, 2017.
61. Caspar SM, Plüss M, Kopps AM, Keller I, Meienberg J, Bruggmann R, Vogel M, **Mátyás G** (2017) Whole-genome sequencing (WGS): Fast and reliable data analysis. 13th Symposium of the ZIHP, Zurich, September 1, 2017.
62. Dubacher N, Münger J, Meienberg J, Crabb J, Bakker E NTP, van Bavel E, Ziegler U, Zeisberger S, **Mátyás G** (2017) Read-out system to assess drug effects on the mechanical integrity of the thoracic aorta in a murine model of Ehlers Danlos syndrome vascular type. 13th Symposium of the ZIHP, Zurich, September 1, 2017.
63. Kopps AM, Fattorini N, Meienberg J, **Mátyás G** (2017) Characterising large deletions using MinION nanopore sequencing. 13th Symposium of the ZIHP, Zurich, September 1, 2017.
64. Odavic D, **Mátyás G**, Henggeler C, Ntinopulos V, Dzemali O, Genoni M (2017) The Importance of Genetic Testing After Replacement Surgery of the Ascending Aorta. Annual Meeting of the Swiss Society of Cardiology and the Swiss Society of Cardiac Surgery, Baden, June 7-9, 2017.
65. Plüss M, Caspar SM, Meienberg J, Kopps AM, Keller I, Bruggmann R, Vogel M, **Mátyás G** (2017) Need for speed in high-throughput sequencing data analysis. ESHG 2017, Copenhagen, May 27-30, 2017.
66. Kopps AM, Meienberg J, Bruggmann R, **Mátyás G** (2017) Whole does not equal whole in WES and WGS. Genomics of Rare Disease, Cambridge UK, April 5-7, 2017.
67. Meienberg J, Kopps AM, Plüss M, Caspar SM, Bruggmann R, **Mátyás G** (2017) Whole-genome sequencing for clinical applications. International Conference on Genomic Medicine, Baltimore USA, February 22-24, 2017.
68. Kopps A, Meienberg J, Bruggmann R, Oexle K, **Mátyás G** (2016) The most comprehensive genomic testing of Mendelian disorders. Swiss MedLab, Berne, June 14-16, 2016.
69. Meienberg J, Bruggmann R, Oexle K, **Mátyás G** (2016) Clinical sequencing: WGS is the better WES. ESHG 2016, Barcelona, May 21-24, 2016.
70. Meienberg J, Münger J, Crabb J, Bakker E NTP, van Bavel E, Ziegler U, Carrel T, Zeisberger S, **Mátyás G** (2016) Ehlers-Danlos syndrome vascular type: Read-out system to assess drug effects on the mechanical property of the thoracic aorta of a *Col3a1* mouse model. EDS International Symposium, New York, May 3-6, 2016.
71. Meienberg J, Bruggmann R, Oexle K, **Mátyás G** (2016) WGS is the better WES for clinical sequencing. Genomics of Rare Disease: Beyond the Exome, Cambridge UK, April 13-15, 2016.

72. Meienberg J, Münger J, Crabb J, Mauri A, Kaiser C, Barmettler G, Giunta C, Ziegler U, Zeisberger S, Mazza E, **Mátyás G** (2015) Mechanical Stability of the Aorta in a Col3a1 Mouse Model. ASHG 65th Annual Meeting, Baltimore MD, October 6-10, 2015.

73. Oexle K, Meienberg J, Zerjavic K, Röthlisberger B, Bruggmann R, **Mátyás G** (2015) Genome versus exome sequencing: Is WGS the better WES? ASHG 65th Annual Meeting, Baltimore MD, October 6-10, 2015.

74. Meienberg J, Münger J, Crabb J, Mauri A, Kaiser C, Barmettler G, Giunta C, Ziegler U, Zeisberger S, Mazza E, **Mátyás** G (2015) Mechanical Stability of the Aorta in a Murine Model of EDS Type IV. 11th Symposium of the Zurich Center for Integrative Human Physiology, Zurich, August 21, 2015.

75. Meienberg J, Zerjavic K, Keller I, Okoniewski M, Patrignani A, Ludin K, Xu Z, Steinmann B, Carrel T, Röthlisberger B, Schlapbach R, Bruggmann R, **Mátyás G** (2015) Is whole genome sequencing (WGS) the better whole exome sequencing (WES)? Evolutionary Medicine Conference 2015, *Journal of Evolutionary Medicine* 3: 34-35.

76. Münger J, Meienberg J, Crabb J, Mauri A, Kaiser C, Barmettler G, Giunta C, Ziegler U, Zeisberger S, Mazza E, **Mátyás G** (2015) Assessment of the mechanical stability of the aorta in a mouse model of Ehlers-Danlos syndrome vascular type (EDS IV). ESHG 2015, *Eur J Hum Genet* 23, Suppl. 1:119.

77. Münger J, Meienberg J, Crabb J, Mauri A, Gysi S, Kaiser C, Barmetler G, de Vos J, Bhattacharya I, Courseau J, Giunta C, Bakker EN, Battegay EJ, Jaeger R, van Bavel E, Haas E, Ziegler U, Kopf M, Zeisberger S, Mazza E, **Mátyás G** (2015) Assessment of the mechanical stability of the aorta in a *Col3a1* mouse model. 14th Day of Clinical Research (DCR), Zurich, April 9, 2015.

78. Münger J, Meienberg J, Crabb J, Mauri A, Kaiser C, Barmettler G, Giunta C, Ziegler U, Zeisberger S, Mazza E, **Mátyás G** (2015) Bestimmung der mechanischen Stabilität der Aorta in einem Mausmodell für Ehlers-Danlos Syndrom vaskulärer Typ (EDS IV). 29. Jahrestagung der Arbeitsgemeinschaft für Pädiatrische Stoffwechsel-Störungen, Fulda, Germany, March 4-6, 2015, *Monatsschrift Kinderheilkunde* 163:647.

79. **Mátyás G***, Naef P, Tollens Martin, Oexle K (2014) De novo *TGFB3* mutation in a patient with overgrowth and Loeys-Dietz syndrome features. 9th International Research Symposium on Marfan Syndrome and Related Disorders, Paris, France, September 25-27, 2014; *oral presentation.

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90. Meienberg J, Okoniewski M, Patrignani A, Szabelska A, Tsai Y-C, Turner SW, Korlach J, Schlapbach R, **Mátyás G** (2013) Breakpoint characterization of large deletions using PacBio and Illumina sequencing technologies. ESHG 2013, *Eur J Hum Genet* 21, Suppl. 1:360.
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93. Meienberg J, Alonso S, Patrignani A, Okoniewski M, Arnold E, Henggeler C, Carrel T, Steinmann B, **Mátyás G** (2012) Characterisation of large hemizygous *FBN1* deletions causing Marfan syndrome. 7th European Elastin Meeting, Ghent, Belgium, September 1-4, 2012.
94. Meienberg J, Patrignani A, Okoniewski M, Henggeler C, Arnold E, Perez R, Mahlberg N, Amstutz N, Burri H, Dutly F, Carrel T, Steinmann B, **Mátyás G** (2012) Evaluation of exome sequencing in genes associated with aortic connective tissue disorders. XXIIIrd FECTS Meeting, Katowice, Poland, August 25-29, 2012.
95. Meienberg J, Patrignani A, Okoniewski M, Henggeler C, Arnold E, Perez R, Mahlberg N, Amstutz N, Burri H, Dutly F, Carrel T, Steinmann B, **Mátyás G** (2012) Evaluation of exome sequencing in genes associated with aortic diseases. ESHG 2012, *Eur J Hum Genet* 20, Suppl. 1:306.
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97. Meienberg J, Patrignani A, Okoniewski M, Henggeler C, Arnold E, Perez R, Mahlberg N, Amstutz N, Burri H, Dutly F, Carrel T, Steinmann B, **Mátyás G** (2012) Evaluation of exome sequencing with different types of sequence variations in genes associated with aortic diseases. RE(ACT) International Congress on Research of Rare and Orphan Diseases, Basel, Switzerland, February 29 – March 2, 2012; *Mol Syndromol* 2:271.

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100. **Mátyás G**, Arnold E, Alonso S, Patrignani A, Magyar I, Henggeler C, Carrel T, Berger W, Steinmann B (2010) *FBN1*, *TGFBR1*, *TGFBR2*, and *SLC2A10* Mutation Analyses in Patients with Suspected Marfan Syndrome: A Swiss Study. 8th International Research Symposium on the Marfan Syndrome and Related Disorders, Warrenton (VA), September 11-14, 2010.

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102. Meienberg J, Neuenschwander S, Rohrbach M, Giunta C, Alonso S, Arnold E, Henggeler C, Spanaus K, Regenass S, Azzarello-Burri S, Berger W, Steinmann B, **Mátyás G** (2010) True haploinsufficiency of *COL3A1* causes aortic dissection. XXIInd FECTS Meeting, Davos, July 3-7, 2010 (selected for oral presentation by Meienberg J).

103. Spanaus K, Meienberg J, Rohrbach M, Neuenschwander S, Giunta C, Alonso S, Henggeler C, Azzarello-Burri S, Steiner B, Berger W, Steinmann B, **Mátyás G** (2010) No evidence for hemochromatosis type 4 in hemizygous *SLC40A1* deletion carriers. ESHG 2010, *Eur J Hum Genet* 18, Suppl. 1:343.

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107. Magyar I, Colman D, Arnold E, Baumgartner D, Bottani A, Fokstuen S, Addor M-C, Berger W, Carrel T, Steinmann B, **Mátyás G** (2009) Quantitative sequence analysis of *FBN1* premature termination codons provides evidence for incomplete NMD in leukocytes. ESHG 2009, *Eur J Hum Genet* 17, Suppl. 1:333.

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110. Stiller R[#], **Mátyás G[#]**, Macas E, Reuge Ph, Berger W, Imthurn B (2008) Successful pregnancy following polar body biopsy and vitrification in a woman with myotonic dystrophy type 1 (Curschmann-Steinert). Women's Health Kongress 2008, Zurich, September 4-5, 2008.
[#]equally contributing authors

111. **Mátyás G**, Arnold E, Alonso S, Patrignani A, Magyar I, Henggeler C, Carrel T, Berger W, Steinmann B (2008) *FBN1*, *TGFBR1*, *TGFBR2*, and *SLC2A10* Mutation Analyses in Patients with Suspected Marfan Syndrome: A Swiss Study. GfH 2008, *Medizinische Genetik* 20:124.

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117. **Mátyás G***, Arnold E, Alonso S, Magyar I, Carrel T, Berger W, Steinmann B (2006) Genetic heterogeneity in Marfan syndrome (MFS): Mutations in *TGFBR1*, *TGFBR2*, and *SLC2A10* cause MFS-like phenotypes. Swiss Pediatric Research Meeting, Bern, November 14, 2006 – "NESTLE RESEARCH PRIZE"; *oral presentation.

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119. **Mátyás G**, Arnold E, Carrel T, Berger W, Steinmann B (2006) Identification and in silico analyses of novel *TGFBR1* and *TGFBR2* mutations in Marfan syndrome-related disorders. 5th Day of clinical research, Zurich, March 23-24, 2006.

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121. Imboden M, Nicod L, Nieters A, Glaus E, **Mátyás G**, Bircher AJ, Ackermann-Liebrich U, Berger W, Probst-Hensch NM, SAPALDIA Team (2006) IL18 single nucleotide polymorphism as genetic risk factor for atopic asthma. The SAPALDIA Cohort Study. Annual Congress of SGAI (Schweizerische Gesellschaft für Allergologie und Immunologie), Zurich, March 30-31, 2006.

122. Zeitz C, van Genderen M, Neidhardt J, Luhmann UFO, Hoeben F, Forster U, Wycisk K, **Mátyás G**, Hoyng CB, Riemslag F, Meire F, Cremers FPM, Berger W (2005) Mutations in *GRM6* cause autosomal recessive congenital stationary night blindness (CSNB) with a distinctive scotopic 15 Hz flicker electroretinogram (ERG). ZNZ Symposium 2005, ETH Zurich, October 21, 2005.

123. Baumgartner C, Baumgartner D, Eberle M, **Mátyás G**, Steinmann B (2005) Aufbau einer Bioinformatik-Plattform für Mutationen im *FBN1*-Gen zur Genotyp-Phänotyp Korrelation beim Marfan-Syndrom. 50. Jahrestagung der Deutschen Gesellschaft für Medizinische Informatik, Biometrie und Epidemiologie (gmds), September 11-15, 2005, *German Medical Science*: Doc 05gmds058, http://www.egms.de/en/meetings/gmds2005/05gmds404.shtml.

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126. **Mátyás G***, Arnold E, Berger W, Steinmann B (2004) Mutation analysis of the TGT beta receptor 2 gene (*TGFBR2*) in patients with Marfan syndrome. Swiss Pediatric Research Meeting, Bern, November 18, 2004; *oral presentation.

127. **Mátyás G***, Imboden M, Probst-Hensch N, Steinmann B, Berger W (2004) Recent tools for DNA extraction, mutation detection and SNP genotyping. International Swiss MedLab and 8th Alps Adria Congress, Lucerne, Switzerland, October 5-9, 2004. *Pipette - Swiss Laboratory Medicine*, Suppl.:26; *oral presentation.

128. Lo B, Maegawa G, Satodia P, Whyte H, Semer M, Hutchinson J , Wilson G, Gross G, **Mátyás G**, Hinek A, Unger S, Chitayat D (2004) Neonatal Marfan Syndrome: Two cases with some unusual clinical findings. ASHG 2004, Abstract no. 580.

129. Maegawa GB, Lo B, Satodia P, Whyte H, Sermer M, Hutchinson J, Wilson G, Gross G, **Mátyás G,** Hinek A, Chitayat D, Unger S (2004) Neonatal Marfan Syndrome: Two cases with some unusual clinical findings. *Eur J Hum Genet* 12, Suppl. 1:118.

130. **Mátyás G**, Arnold E, Maudrich M, Berger W, Steinmann B (2004) Application of DHPLC for *FBN1* mutation detection: Identification of 58 novel mutations. ESHG 2004, *Eur J Hum Genet* 12, Suppl. 1:256-257.

131. **Mátyás G*** (2004) Molekulare Diagnostik des Marfan-Syndroms - Suche nach der Nadel im Heuhaufen. 1. Schweizerische Stoffwechseltagung, Zurich, January 28, 2004; *oral presentation.

132. Dutly F, Goldenberger D, **Mátyás G**, Frischknecht H (2003) Identification of *Neisseria gonorrhoeae* and Herpes Simplex Virus DNA by Denaturing High-Performance Liquid Chromatography. 3th European Meeting on Molecular Diagnostics, Scheveningen, The Netherlands, October 16-17, 2003.

133. Gugerli F, Sperisen C, **Mátyás G**, Senn J, Anzidei M, Vendramin GG (2001) Molecular markers help to reveal postglacial re-colonisation routes of forest tree species in the Alpine Region. Interdisciplinary Mountain Research, Young Scientists Conference, Stilfserjoch/Stelvio National Park, Italy, September 25-28, 2001, Abstract no. 62.

134. **Mátyás G***, De Paepe A, Halliday D, Boileau C, Pals G, Steinmann B (2001) Evaluation and application of denaturing HPLC for mutation detection in Marfan syndrome. 6th International Symposium on Marfan Syndrome. Seattle, USA, August 12-13, 2001; *oral presentation.

135. **Mátyás G**, De Paepe A, Boileau C, Handford P, Pals G, Steinmann B (2001) Marfan syndrome: Mutation detection in the *FBN1* gene by DHPLC – A sensitivity study. 6th International Symposium on Marfan Syndrome. Seattle, USA, August 12-13, 2001.

136. **Mátyás G**, De Paepe A, Boileau C, Handford P, Pals G, Steinmann B (2000) Marfan syndrome: Mutation detection in the *FBN1* gene by DHPLC – A sensitivity study. Swiss Pediatric Research Meeting, Lausanne, Switzerland, November 30, 2000.
137. **Mátyás G,** Nuytinck L, De Paepe A, Halliday D, Handford P, Steinmann B (2000) Where is the Marfan mutation? Sensitivity study of denaturing HPLC (DHPLC) for routine mutation detection in the Marfan syndrome. EMBO course «Advanced Techniques in Molecular Medicine», Uppsala, Sweden, August 24-30, 2000.

INVITED ORAL PRESENTATIONS

1. **Mátyás G** (2019) Personalisierte Medizin und Gendiagnostik. Health Business School 2019, St. Gallen, März 6, 2019.
2. **Mátyás G** (2018) Differential Diagnostics of Marfan Syndrome and other Aortopathies with Genetic Background. Central-Eastern European Marfan Symposium, Budapest, Hungary, October 26, 2018.
3. **Mátyás G** (2017) Cardiac Genomics. Live-Symposium «Moderne Herzmedizin» des Herzzentrums Hirslanden, Zürich, September 21, 2017.
4. **Mátyás G** (2017) Genetic Evaluation of Aortopathies. 15. Zürcher Review Kurs in Klinischer Kardiologie, Zurich, April 8, 2017.
5. **Mátyás G** (2017) Rare Genomics am Zentrum für Kardiovaskuläre Genetik und Gendiagnostik. Genomics Board Hirslanden, Zürich, March 6, 2017.
6. **Mátyás G** (2016) Personalisierte Medizin und Gendiagnostik. Health Business School 2016, Schaffhausen, November 9, 2016.
7. **Mátyás G** (2016) Loeys-Dietz Syndrom Typ 5 (TGFB3-Mutation): Eine Differenzialdiagnose des Marfan Syndroms. Universitätsspital, Zürich, October 31, 2016.
8. **Mátyás G** (2016) Genetik beim Marfan-Syndrom: Hilfe für Diagnose und Differentialdiagnose. Echoforum Hirslanden, Zürich, February 2, 2016.
9. **Mátyás G** (2015) Mechanische Stabilität der Aorta im Mausmodell für EDS vaskulärer Typ (EDS IV). Universitätsspital, Zürich, October 19, 2015.
10. **Mátyás G** (2015) Verdacht auf MFS: Ein Update über die genetische Abklärung. Universitätsspital, Zürich, April 20, 2015.
11. **Mátyás G** (2014) Genetische Untersuchungen und Risikoprüfung anhand von Fallbeispielen. ASA/SVV Fortbildungsreihe «Genetik 2014», Modul 3: Umsetzung für Versicherungsmediziner, Underwriter und Versicherungsfachleute aus der Risiko- und Leistungsprüfung. Olten, November 20, 2014.
12. **Mátyás G** (2014) Rare Genomics at the Genetic Center of the Foundation for People with Rare Diseases. Interdisziplinäres Kolloquium Regenerative Medizin I, Universitätsspital, Zürich, October 28, 2014.
13. **Mátyás G** (2014) Aortenkrankheiten und Orphan Diseases. ASA/SVV Fortbildungsreihe «Genetik 2014», Modul 2: Lesen und Interpretation von genetischen Informationen. Olten, August 28, 2014.
14. **Mátyás G** (2014) Erfassung, Beschreibung und Bedeutung von Sequenzvarianten sowie Umgang mit Datenbanken. ASA/SVV Fortbildungsreihe «Genetik 2014», Modul 2: Lesen und Interpretation von genetischen Informationen. Olten, August 28, 2014.
15. **Mátyás G** (2014) Life Science Zurich Young Scientist Network (LSZYSN). Company Visit: Bio-Technopark. Schlieren, August 21, 2014.
16. **Mátyás G** (2014) Grundlagen und Terminologie von genetischen Untersuchungen. ASA/SVV Fortbildungsreihe «Genetik 2014», Modul 1: Basiswissen zur Genetik. Olten, May 22, 2014.
17. **Mátyás G** (2014) Ehlers-Danlos-Syndrom (EDS IV). Fortbildung Gefässmedizin, Universitätsspital, Zürich, May 13, 2014.
18. **Mátyás G** (2014) Genetische Aortenkrankheiten. Angiologische Kolloquien, Universitätsspital, Basel, April 29, 2014.

19. **Mátyás G** (2014) Marfan oder nicht Marfan, das ist hier die Frage. Echoforum Hirslanden, Zürich, March 18, 2014.
20. **Mátyás G** (2014) Genetische Abklärung zu medizinischen Zwecken. SIM Jahrestagung 2014. Olten, March 13, 2014.
21. **Mátyás G** (2013) Gendiagnostik, aller Anfang ist Forschung. Weiterbildung GSK, UCB "Orphan Disease". Münchenbuchsee, December 4, 2013.
22. **Mátyás G** (2013) Genetic Aortic Diseases – Diagnosis and Therapy of Marfan Syndrome (MFS) and Related Aortic Diseases (AD). 16. Dreiländertagung, Angiologie 2013, Graz, Austria, September 17, 2013.
23. **Mátyás G** (2013) Novel insights into the molecular bases of Marfan syndrome (MFS) and related aortic diseases (AD). EMSN conference and Swiss Marfan Days, Zurich, September 3, 2013.
24. **Mátyás G** (2013) Genetische Abklärungen zu medizinischen Zwecken. 3. Forum Risikoprüfung in der Personenversicherung, Zurich-Adliswil, June 6, 2013.
25. **Mátyás G** (2013) Gendiagnostik von Marfan-Syndrom und verwandten Aortenkrankheiten. Fortbildung «Herzchirurgie», Basel, February 4, 2013.
26. **Mátyás G** (2013) Genetische Untersuchungen beim Menschen - eine gesellschaftliche Herausforderung. Einführung in die Gendiagnostik. ProGenom Forum, Zurich, January 22-23, 2013.
27. **Mátyás G** (2012) The role of geneticists in aortic diseases: Molecular genetic testing of aortic diseases in Switzerland. 2nd Swiss Russian Health Forum, Basel, September 11, 2012.
28. **Mátyás G** (2012) Gendiagnostik der Aortendissektion/Aortenerkrankungen. Symposium Erkrankungen der Aorta, University Hospital, Zurich, September 6, 2012.
29. **Mátyás G** (2012) Interdisziplinarität und Nachhaltigkeit am Zentrum für Kardiovaskuläre Genetik und Gendiagnostik. Strickhof Lindau, July 11, 2012.
30. **Mátyás G** (2012) Simplified characterization of large deletions using the PacBio sequencing platform (Workshop – Revealing the Genome through SMRT Biology: Discoveries and Applications). European Human Genetics Conference 2012, Nurnberg, Germany, June 23, 2012.
31. **Mátyás G** (2012) Genetic assessment in patients with a large aorta. 10. Zürcher Review Kurs in Klinischer Kardiologie, Zurich, April 20, 2012.
32. **Mátyás G** (2012) Gendiagnostik von Marfan-Syndrom und verwandten Aortenkrankheiten. Weiter- und Fortbildungs-Symposium «Molekulare Diagnostik», Zurich, March 8, 2012.
33. **Mátyás G** (2011) Genetik und Zukunft. Marfan-Tag, Aarau, October 1, 2011.
34. **Mátyás G** (2010) Genetische Abklärungsmöglichkeiten in der Schweiz. Ärzte Gesellschaft des Kantons Zürich AGZ, Zürich, December 6, 2010.
35. **Mátyás G** (2010) Gendiagnostik bei Marfan-Syndrom und ähnlichen Aortenaneurysma-Syndromen. Kinderspital Zürich, July 27, 2010.
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