

**ROSELLINA M MANCINA, PHD**

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**1. POSIZIONE ATTUALE:**

-Dicembre 2023- presente: *Professore Associato* (S.S.D. MED/50 – S.C. 06/N1) presso il Dipartimento di Scienze della Vita, della Salute e delle Professioni Sanitarie, Università degli studi Link , Roma, Italia.

-Luglio 2022- presente: consulente scientifico.

**2. POSIZIONI PREGRESSE:**

-Maggio 2018- Aprile 2022: *Ricercatore* presso il Dipartimento di Medicina Clinica e Sperimentale, Università di Goteborg, Goteborg, Svezia.

-Maggio 2014- Maggio 2018: *Post Doc* presso il Dipartimento di Medicina Clinica e Sperimentale, Università di Goteborg, Goteborg, Svezia.

-Febbraio 2011- Aprile 2014: *studente di Dottorato (PhD student)* in Scienze endocrino-metaboliche e della nutrizione presso l'Università "Magna Græcia" di Catanzaro, Italia e il Dipartimento di Medicina Clinica e Sperimentale, Università di Goteborg, Goteborg, Svezia. Titolo della tesi: "*Molecular genetics of hepatic steatosis: the role of PNPLA3*".

-Maggio 2009- Maggio 2010: *Master student* presso l'Istituto di Scienze Neurologiche -CNR c.da Burga – 87050 - Mangone (CS), Italia. Titolo della tesi: "*Molecular analysis of patients with Huntington-like disease*".

**3. TITOLI DI STUDIO:**

-Febbraio 2011- Aprile 2014: *Dottorato di Ricerca* in Scienze endocrino-metaboliche e della nutrizione presso l'Università "Magna Græcia" di Catanzaro, Italia. Titolo della tesi: "*Molecular genetics of hepatic steatosis: the role of PNPLA3*".

-Settembre 2007- Maggio 2010: *Laurea specialistica in Scienze Biologiche (MSc)* presso l'università della Calabria, Cosenza, Italia. Titolo della tesi: "*Molecular analysis of patients with Huntington-like disease*". Score: 110/110 cum laude

-Settembre 2004- Ottobre 2007: *Laurea triennale in Scienze Biologiche (MSc)* presso l'università della Calabria, Cosenza, Italia. Titolo della tesi: "*Successful aging: from phenotype to genotype*".

**4. RICONOSCIMENTI SCIENTIFICI:**

- Febbraio 2023- Febbraio 2033: "Abilitazione scientifica nazionale (ASN), fascia II" SC 06/A1 – *Genetica Medica*.

- Maggio 2021- Maggio 2031: "Abilitazione scientifica nazionale (ASN), fascia II" SC 06/N1 - *Scienze delle Professioni Sanitarie e delle Tecnologie Mediche Applicate*.

- 2016: Articolo premiato con la copertina della rivista *Gastroenterology*: Mancina RM et al. The MBOAT7-TMC4 Variant rs641738 Increases Risk of Nonalcoholic Fatty Liver Disease in Individuals

of European Descent.

#### **4. FINANZIAMENTI PER PROGETTI DI RICERCA COME PRINCIPAL INVESTIGATOR:**

- "Wilhelm and Martina Lundgren Science Fund" project grant 2016. Titolo del progetto: *MBOAT7 rs641738 genetic variant and the risk of hepatic steatosis in obese children.*
- "Wilhelm and Martina Lundgren Science Fund" project grant 2015. Titolo del progetto: *Genetic risk factors for insulin resistance and type 2 diabetes in obese children.*
- "Nilsson-Ehle-donationerna" project grant 2014. Titolo del progetto: *Genetic risk factors for insulin resistance and type 2 diabetes in obese children.*

#### **5. SUPERVISIONE:**

Co-supervisore per 2 studenti di dottorato:

- OJ (discussione delle tesi 16-Gennaio-2023), Dipartimento di Medicina Clinica e Sperimentale, Università di Goteborg, Goteborg, Svezia; PhD project title: *Identifying new gene targets to treat hyperlipidemia.*
- KS (discussione delle tesi 3-Marzo-2023), Dipartimento di Medicina Clinica e Sperimentale, Università di Goteborg, Goteborg, Svezia.; PhD project title: *Understanding the genetic architecture of non-viral chronic liver disease.*

#### **6. ATTIVITÀ DIDATTICHE INTERNAZIONALI:**

- 2018-2022: "Neurofisiologia" all'interno del Corso di "Anatomia e Fisiologia", Università di Goteborg, Goteborg, Svezia
- 2019-2020: "Lipogenesi e fatty liver disease" all'interno del Corso di "Biomedicina", Università di Goteborg, Goteborg, Svezia
- 2018-2019: "Obesity and Inflammation" all'interno del Corso di "Biomedicina", Università di Goteborg, Goteborg, Svezia

#### **7. ORGANIZZAZIONE DI CONGRESSI SCIENTIFICI:**

- Organizzatore: EASD NAFLD Study Group, Gothenburg Sweden, 8-9 Maggio 2017. *Sezione: genetica della malattia epatica.*
- Organizzatore: Atheroma Club, Marstrand, Gothenburg Sweden, 27-29 Settembre 2017. *Sezione: genetica dell'ipercolesterolemia familiare.*
- Organizzatore: Atheroma Club, Marstrand, Gothenburg Sweden, 21-23 September 2016. *Sezione: genetica dell'ipercolesterolemia familiare.*

#### **8. COMMISSIONS OF TRUST:**

- 2021-2022: Guest Editor per la rivista internazionale International Journal of Molecular Sciences per lo Special Issue dal titolo: "Molecular Genetics of fatty liver disease"
- 2019- presente: Membro dell' Editorial Board di Molecular Medicine, parte del giornale internazionale "Frontiers in Cell and Developmental Biology"

- 2018-presente: Membro dell' Editorial Board della rivista scientifica internazionale "Journal of Endocrinological Investigation"
- 2017-presente: Reviewer per importanti riviste scientifiche internazionali quali *Liver International; Cellular, Molecular Gastroenterology and Hepatology* (CMGH), *Surgery for Obesity and Related Diseases*, *Journal of Hepatology and Gastroenterology*, e *Human Immunology*

#### 9. PARTECIPAZIONE A CONFERENZE E WORKSHOP INTERNAZIONALI COME RELATORE INVITATO:

- Conferenza: The NASH-TAG Conference (THERAPEUTIC AGENTS FOR NON-ALCOHOLIC STEATOHEPATITIS AND LIVER FIBROSIS), Gennaio 4-6, 2024 in Park City, Utah (USA); Titolo: PSD3 in NASH.
- Conferenza: Genomic Medicine 2018 Nordic 1-3 Ottobre 2018 a Odense (DK); Titolo: *Genetic diagnosis of familial hypercholesterolaemia by targeted next-generation sequencing*
- Workshop all'interno della conferenza "Liver Days (Leverdagarna)" 5 Ottobre 2017, Gothenburg: Current research in Swedish hepatology; Titolo: *MBOAT7 rs641738 variant increases the risk of nonalcoholic fatty liver disease*.

#### 10. TRACK RECORD SCIENTIFICO E PUBBLICAZIONI (Marzo 2024):

Secondo Scopus (ID 55248202600), numero totale di citazioni: 4,064.

- **h-index 32**
- **62 pubblicazioni su riviste scientifiche internazionali**, includendo 4 come corresponding author, 13 come primo autore (includendo uno come primo e corresponding author, 2 invited editorials e una lettera), 2 invited editorials, 2 reviews e una lettera.
- >10 articoli con impact factor  $\geq 15$ , includendo articoli su *Nature Metabolism* (IF 20, first author), *Gastroenterology* (IF 23, first author e uno come co-author), *Cell metabolism* (IF 27), *Gut* (IF 23; x2), *Hepatology* (IF 17; x3), *Journal of Hepatology* (IF 25; x2).
- >30 articoli con impact factor  $\geq 5$ , includendo articoli su *Cell Reports Medicine* (IF 14), *Journal of Internal Medicine* (IF 9; x3, incluso uno come first author), *Human Molecular Genetics* (IF 6; x2) e *The Journal of Clinical Endocrinology & Metabolism* (IF 6; x2 incluso uno come first author).

#### Lista delle pubblicazioni

##### 10 PUBBLICAZIONI SELEZIONATE COME FIRST, LAST E/O CORRESPONDING AUTHOR

		Impact Factor
1	<b>Mancina RM*</b> , Sasidharan K*, Lindblom A, Wei Y, Ciociola E, Jamialahmadi O, Pingitore P, Andréasson AC, Pellegrini G, Baselli G, Männistö V, Pihlajamäki J, Kärjä V, Grimaudo S, Marini I, Maggioni M, Becattini B, Tavaglione F, Dix C, Castaldo M, Klein S, Perelis M, Pattou F, Thuillier D, Raverdy V, Dongiovanni P, Fracanzani AL, Stickel F, Hampe J, Buch S, Luukkonen PK, Prati D, Yki-Järvinen H, Petta S, Xing C, Schafmayer C, Aigner E, Datz C, Lee RG, Valenti L, Lindén D, Romeo S. PSD3 downregulation confers protection against fatty liver disease. <i>Nat Metab.</i> 2022 Jan;4(1):60-75. doi: 10.1038/s42255-021-00518-0. PMID: 35102341 * co-first authors	20
2	Raja AM, Ciociola E, Ahmad IN, Dar FS, Naqvi SMS, Moaen-Ud-Din M, Kaukab Raja G, Romeo S, <b>Mancina RM</b> . Genetic Susceptibility to Chronic Liver Disease in Individuals from Pakistan. <i>Int J Mol Sci.</i> 2020 May 18;21(10):3558. doi:	6

- 10.3390/ijms21103558. PMID: 32443539
- 3 Pavanello C, Pirazzi C, Bjorkman K, Sandstedt J, Tarlarini C, Mosca L, Romeo S\*, Calabresi L\*, **Mancina RM\***. Individuals with familial hypercholesterolemia and cardiovascular events have higher circulating Lp(a) levels. *J Clin Lipidol*. 2019 Jul 4. pii: S1933-2874(19)30231-4. doi: 10.1016/j.jacl.2019.06.011. PMID: 31371270 \*co-corresponding 5
  - 4 Pirazzi C\*, Håkansson L, Gustafsson C, Omerovic E, Wiklund O, **Mancina RM\***. High prevalence of genetic determined familial hypercholesterolemia in premature coronary artery disease. *Appl Clin Genet*. 2019 May 24;12:71-78. doi: 10.2147/TACG.S202942. PMID: 31213876 \*co-corresponding 2
  - 5 **Mancina RM\***, Ferri F, Farcomeni A, Molinaro A, Maffongelli A, Mischitelli M, Poli E, Parlati L, Burza MA, De Santis A, Attilia F, Rotondo C, Rando MM, Attilia ML, Ceccanti M, Ginanni Corradini S\*. A two gene-based risk score predicts alcoholic cirrhosis development in males with at-risk alcohol consumption. *Appl Clin Genet*. 2019 Jan 10;12:1-10. doi: 10.2147/TACG.S187922. PMID: 30666147 \*co-corresponding 2
  - 6 **Mancina RM\***, Dongiovanni P\*, Petta S, Pingitore P, Meroni M, Rametta R, Borén J, Montalcini T, Pujia A, Wiklund O, Hindy G, Spagnuolo R, Motta BM, Pipitone RM, Craxi A, Fargion S, Nobili V, Käkelä P, Kärjä V, Männistö V, Pihlajamäki J, Reilly DF, Castro-Perez J, Kozlitina J, Valenti L, Romeo S. The MBOAT7-TMC4 Variant rs641738 Increases Risk of Nonalcoholic Fatty Liver Disease in Individuals of European Descent. *Gastroenterology*. 2016 Feb 2. pii: S0016-5085(16)00131-1. doi: 10.1053/j.gastro.2016.01.032 PMID: 26850495 \*co-first authors This paper has been awarded the Gastroenterology journal cover 34
  - 7 **Mancina RM\***, Sentinelli F\*, Incani M, Bertocchini L, Russo C, Romeo S, Baroni MG. Transmembrane-6 superfamily member 2 (TM6SF2) E167K variant increases susceptibility to hepatic steatosis in obese children. *Dig Liver Dis*. 2016 Jan;48(1):100-1. doi: 10.1016/j.dld.2015.10.003. PMID: 26520056 \*co-first authors 5
  - 8 **Mancina RM\***, Spagnuolo R\*, Milano M, Brogneri S, Morrone A, Cosco C, Lazzaro V, Russo C, Ferro Y, Pingitore P, Pujia A, Montalcini T, Doldo P, Garieri P, Piodi L, Caprioli F, Valenti L, Romeo S. PNPLA3 148M Carriers with Inflammatory Bowel Diseases Have Higher Susceptibility to Hepatic Steatosis and Higher Liver Enzymes. *Inflamm Bowel Dis*. 2016 Jan;22(1):134-40. doi: 10.1097/MIB.0000000000000569. PMID: 26355465 \*co-first authors 7
  - 9 Mondul A\*, **Mancina RM\***, Merlo A\*, Dongiovanni P, Rametta R, Montalcini T, Valenti L, Albanes D, Romeo S. PNPLA3 I148M Variant Influences Circulating Retinol in Adults with Nonalcoholic Fatty Liver Disease or Obesity. *J Nutr*. 2015 Aug;145(8):1687-91. doi: 10.3945/jn.115.210633. PMID: 26136587 \*co-first authors 5
  - 10 **Mancina RM\***, Matikainen N\*, Maglio C, Söderlund S, Lundbom N, Hakkarainen A, Rametta R, Mozzi E, Fargion S, Valenti L, Romeo S, Taskinen MR, Borén J. Paradoxical Dissociation Between Hepatic Fat Content and De Novo Lipogenesis Due to PNPLA3 Sequence Variant. *J Clin Endocrinol Metab*. 2015 May;100(5):E821-5. doi: 10.1210/jc.2014-4464. PMID: 25763607 \*co-first authors 6

#### ALTRI ARTICOLI COME FIRST, LAST E/O CORRESPONDING AUTHOR

- 11 **Mancina RM**, De Bonis D, Pagnotta R, Cosco C, Cosco V, Montalcini T, Pujia A, Doldo P, Spagnuolo R. Ulcerative Colitis as an Independent Risk Factor for Hepatic Steatosis. *Gastroenterol Nurs*. 2020 Jul/Aug;43(4):292-297. doi: 10.1097/SGA.0000000000000461. PMID: 32665525 1
- 12 **Mancina RM**, Pagnotta R, Pagliuso C, Albi V, Bruno D, Garieri P, Doldo P, Spagnuolo R. Gastrointestinal Symptoms of and Psychosocial Changes in Inflammatory Bowel Disease: A Nursing-Led Cross-Sectional Study of Patients in Clinical Remission. *Medicina (Kaunas)*. 2020 Jan 20;56(1):45. doi: 10.3390/medicina56010045. PMID: 31968710 3

- 13 Dongiovanni P\*, Stender S\*, Pietrelli A\*, **Mancina RM\***, Cespiati A, Petta S, Pelusi S, Pingitore P, Badiali S, Maggioni M, Mannisto V, Grimaudo S, Pipitone RM, Pihlajamaki J, Craxi A, Taube M, Carlsson L, Fargion S, Romeo S, Kozlitina J, and Valenti L. Causal relationship of hepatic fat with liver damage and insulin resistance in nonalcoholic fatty liver. *J Intern Med.* 2018 Apr;283(4):356-370. doi: 10.1111/joim.12719. PMID: 29280273 \*co-first authors 13
- 14 **Mancina RM**, Burza MA, Maglio C, Pirazzi C, Sentinelli F, Incani M, Montalcini T, Pujia A, Congiu T, Loche S, Pilia S, Wiklund O, Borén J, Romeo S, Baroni MG. The COBLL1 C allele is associated with lower serum insulin levels and lower insulin resistance in overweight and obese children. *Diabetes Metab Res Rev.* 2013 Jul;29(5):413-6. doi: 10.1002/dmrr.2408. PMID: 23463496 8

## ARTICOLI COME CO-AUTHOR:

- 15 Pennisi G, Maurotti S, Ciociola E, Jamialahmadi O, Bertolazzi G, Mirarchi A, Bergh PO, Scionti F, **Mancina RM**, Spagnuolo R, Tripodo C, Boren J, Petta S, Romeo S. *ANGPTL3* Downregulation Increases Intracellular Lipids by Reducing Energy Utilization. *Arterioscler Thromb Vasc Biol.* 2024 Feb 22. doi: 10.1161/ATVBAHA.123.319789. Online ahead of print. PMID: 38385290 8.7
- 16 Dutta T, Sasidharan K, Ciociola E, Pennisi G, Noto FR, Kovooru L, Kroon T, Lindblom A, Du Y, Pirmoradian M, Wallin S, **Mancina RM**, Lindén D, Romeo S. Mitochondrial amidoxime-reducing component 1 p.Ala165Thr increases protein degradation mediated by the proteasome. *Liver Int.* 2024 Feb 20. doi: 10.1111/liv.15857. PMID: 38375985 6.7
- 17 Sasidharan K, Caddeo A, Jamialahmadi O, Noto FR, Tomasi M, Malvestiti F, Ciociola E, Tavaglione F, **Mancina RM**, Cherubini A, Bianco C, Mirarchi A, Männistö V, Pihlajamäki J, Kärjä V, Grimaudo S, Luukkonen PK, Qadri S, Yki-Järvinen H, Petta S, Manfrini S, Vespasiani-Gentilucci U, Bruni V, Valenti L, Romeo S. *IL32* downregulation lowers triglycerides and type I collagen in di-lineage human primary liver organoids. *Cell Rep Med.* 2024 Jan 16;5(1):101352. doi: 10.1016/j.xcrm.2023.101352. PMID: 38232700 14.3
- 18 Albuquerque J, Medeiros AM, Alves AC, Jannes CE, **Mancina RM**, Pavanello C, Chora JR, Mombelli G, Calabresi L, Pereira ADC, Krieger JE, Romeo S, Bourbon M, Antunes M. Generation and validation of a classification model to diagnose familial hypercholesterolaemia in adults. *Atherosclerosis.* 2023 Oct;383:117314. doi: 10.1016/j.atherosclerosis.2023.117314. Epub 2023 Sep 28. PMID: 37813054 5.3
- 19 Mischitelli M, Spagnoli A, Abbatecola A, Codazzo C, Giacomelli M, Parisse S, **Mancina RM**, Rotondo C, Attilia F, Ginanni Corradini S, Ferri F. New Diagnostic and Prognostic Models for the Development of Alcoholic Cirrhosis Based on Genetic Predisposition and Alcohol History. *Biomedicines.* 2023 Jul 28;11(8):2132. doi: 10.3390/biomedicines11082132. PMID: 37626629 4.7
- 20 Jamialahmadi O, Salehabadi E, Hashemi-Najafabadi S, Motamedian E, Bagheri F, **Mancina RM**, Romeo S. Cellular Genome-Scale Metabolic Modeling Identifies New Potential Drug Targets Against Hepatocellular Carcinoma. *OMICS.* 2022 Dec;26(12):671-682. doi: 10.1089/omi.2022.0122. PMID: 36508280 4
- 21 Spagnuolo R, Iaquina FS, Mauro D, Pantano I, Dastoli S, Naty S, Cosco C, **Mancina RM**, Iacono D, Gaggiano E, Ruggiero A, Nisticò SP, Ciccio F, Grembiale RD, Patrizia D. Satisfaction in Social Roles and Physical Function in Immune-mediated Inflammatory Diseases: A Cross-Sectional Study. *Rev Recent Clin Trials.* 2022 May 31. doi: 10.2174/1574887117666220531162104. Online ahead of print. PMID: 35642115 0
- 22 Govaere O, Petersen SK, Martinez-Lopez N, Wouters J, Van Haele M, **Mancina RM**, Jamialahmadi O, Bilkei-Gorzo O, Lassen PB, Darlay R, Peltier J, Palmer JM, Younes R, Tiniakos D, Aithal GP, Allison M, Vacca M, Göransson M, Berlinguer-Palmini R, Clark JE, Drinnan MJ, Yki-Järvinen H, Dufour JF, Ekstedt M, Francque S, Petta S, Bugianesi E, Schattenberg JM, Day CP, Cordell HJ, Topal B, Clément K, Romeo S, Ratzu V, 30

- Roskams T, Daly AK, Anstee QM, Trost M, Härtlova A. Macrophage scavenger receptor 1 mediates lipid-induced inflammation in non-alcoholic fatty liver disease. *J Hepatol.* 2022 May;76(5):1001-1012. doi: 10.1016/j.jhep.2021.12.012. PMID: 34942286
- 23** Tavaglione F, Jamialahmadi O, De Vincentis A, Qadri S, Mowlaei ME, **Mancina RM**, Ciociola E, Carotti S, Perrone G, Bruni V, Gallo IF, Tuccinardi D, Bianco C, Prati D, Manfrini S, Pozzilli P, Picardi A, Caricato M, Yki-Järvinen H, Valenti L, Vespasiani-Gentilucci U, Romeo S. Development and Validation of a Score for Fibrotic Non-Alcoholic Steatohepatitis. *Clin Gastroenterol Hepatol.* 2022 Apr 11:S1542-3565(22)00385-8. doi: 10.1016/j.cgh.2022.03.044. Online ahead of print. PMID: 35421583
- 24** Baselli GA, Jamialahmadi O, Pelusi S, Ciociola E, Malvestiti F, Saracino M, Santoro L, Cherubini A, Dongiovanni P, Maggioni M, Bianco C, Tavaglione F, Cespiati A, **Mancina RM**, D'Ambrosio R, Vaira V, Petta S, Miele L, Vespasiani-Gentilucci U, Federico A, Pihlajamäki J, Bugianesi E, Fracanzani AL, Reeves HL, Soardo G, Prati D, Romeo S, Valenti LV; EPIDEMIC Study Investigators. Rare ATG7 genetic variants predispose patients to severe fatty liver disease. *J Hepatol.* 2022 Apr 8:S0168-8278(22)00207-0. doi: 10.1016/j.jhep.2022.03.031. Online ahead of print. PMID: 35405176
- 25** Tavaglione F, De Vincentis A, Bruni V, Gallo IF, Carotti S, Tuccinardi D, Spagnolo G, Ciociola E, **Mancina RM**, Jamialahmadi O, D'Alessio R, Bottazzi B, Manfrini S, Picardi A, Perrone G, Pozzilli P, Caricato M, Vespasiani-Gentilucci U, Romeo S. Accuracy of controlled attenuation parameter for assessing liver steatosis in individuals with morbid obesity before bariatric surgery. *Liver Int.* 2022 Feb;42(2):374-383. doi: 10.1111/liv.15127. PMID: 34890093
- 26** Borén J, Adiels M, Björnson E, Matikainen N, Söderlund S, Rämö J, Ståhlman M, Ripatti P, Ripatti S, Palotie A, **Mancina RM**, Ainola M, Hakkarainen A, Romeo S, Packard CJ, Taskinen MR. Effects of PNPLA3 I148M on hepatic lipid and very low-density lipoprotein metabolism in humans. *J Intern Med.* 2021 Aug 19. doi: 10.1111/joim.13375. Online ahead of print. PMID: 34411351
- 27** Tanaka Y, Shimanaka Y, Caddeo A, Kubo T, Mao Y, Kubota T, Kubota N, Yamauchi T, **Mancina RM**, Baselli G, Luukkonen P, Pihlajamäki J, Yki-Järvinen H, Valenti L, Arai H, Romeo S, Kono N. LPIAT1/MBOAT7 depletion increases triglyceride synthesis fueled by high phosphatidylinositol turnover. *Gut.* 2021 Jan;70(1):180-193. doi: 10.1136/gutjnl-2020-320646. PMID: 32253259
- 28** Jamialahmadi O, **Mancina RM**, Ciociola E, Tavaglione F, Luukkonen PK, Baselli G, Malvestiti F, Thuillier D, Raverdy V, Männistö V, Pipitone RM, Pennisi G, Prati D, Spagnuolo R, Petta S, Pihlajamäki J, Pattou F, Yki-Järvinen H, Valenti L, Romeo S. Exome-wide Association Study on Alanine Aminotransferase Identifies Sequence Variants in the GPAM and APOE Associated with Fatty Liver Disease. *Gastroenterology.* 2020 Dec 18:S0016-5085(20)35562-1. doi: 10.1053/j.gastro.2020.12.023. Online ahead of print. PMID: 33347879
- 29** Borén J, Adiels M, Björnson E, Matikainen N, Söderlund S, Rämö J, Ståhlman M, Ripatti P, Ripatti S, Palotie A, **Mancina RM**, Hakkarainen A, Romeo S, Packard CJ, Taskinen MR. Effects of TM6SF2 E167K on hepatic lipid and very low-density lipoprotein metabolism in humans. *JCI Insight.* 2020 Dec 17;5(24):144079. doi: 10.1172/jci.insight.144079. PMID: 33170809
- 30** Spagnuolo R, Corea A, Blumetti M, Giovinazzo A, Serafino M, Pagliuso C, Pagnotta R, Curto G, Cosco C, Cosco V, **Mancina RM**, Garieri P, Papaleo A, Grande L, Barilaro A, Garofalo E, Bruni A, Doldo P. Effects of listening to music in digestive endoscopy: A prospective intervention study led by nursing. *J Adv Nurs.* 2020 Nov;76(11):2993-3002. doi: 10.1111/jan.14516. PMID: 32901972
- 31** Pina A, Helgadottir S, **Mancina RM**, Pavanello C, Pirazzi C, Montalcini T, Henriques R, Calabresi L, Wiklund O, Macedo MP, Valenti L, Volpe G, Romeo S. Virtual genetic diagnosis for familial hypercholesterolemia powered by machine learning. *Eur J Prev*

- Cardiol.* 2020 Oct;27(15):1639-1646. doi: 10.1177/2047487319898951. PMID: 32019371
- 32 Baselli GA, Dongiovanni P, Rametta R, Meroni M, Pelusi S, Maggioni M, Badiali S, Pingitore P, Maurotti S, Montalcini T, Taliento AE, Prati D, Rossi G, Fracanzani AL, **Mancina RM**, Romeo S, Valenti L. Liver transcriptomics highlights interleukin-32 as novel NAFLD-related cytokine and candidate biomarker. *Gut.* 2020 Oct;69(10):1855-1866. doi: 10.1136/gutjnl-2019-319226. PMID: 32001554 32
- 33 De Vincentis A, **Mancina RM**, Pihlajamäki J, Männistö V, Petta S, Dongiovanni P, Fracanzani AL, Valenti L, Tavaglione F, Romeo S, Vespasiani-Gentilucci U. Genetic variants in the MTHFR are not associated with fatty liver disease. *Liver Int.* 2020 Aug;40(8):1934-1940. doi: 10.1111/liv.14543. PMID: 32460399 9
- 34 Prill S, Caddeo A, Baselli G, Jamialahmadi O, Dongiovanni P, Rametta R, Kanebratt KP, Pujia A, Pingitore P, **Mancina RM**, Lindén D, Whatling C, Janefeldt A, Kozyra M, Ingelman-Sundberg M, Valenti L, Andersson TB, Romeo S. The TM6SF2 E167K genetic variant induces lipid biosynthesis and reduces apolipoprotein B secretion in human hepatic 3D spheroids. *Sci Rep.* 2019 Aug 12;9(1):11585. doi: 10.1038/s41598-019-47737-w. PMID: 31406127 5
- 35 Caddeo A, Jamialahmadi O, Solinas G, Pujia A, **Mancina RM**, Pingitore P, Romeo S. MBOAT7 is anchored to endomembranes by six transmembrane domains. *J Struct Biol.* 2019 Jun 1;206(3):349-360. doi: 10.1016/j.jsb.2019.04.006. PMID: 30959108 3
- 36 Dongiovanni P, Meroni M, Baselli G, **Mancina RM**, Ruscica M, Longo M, Rametta R, Cespiati A, Pelusi S, Ferri N, Ranzani V, Nobili V, Pihlajamäki J, Fracanzani AL, Badiali S, Petta S, Fargion S, Romeo S, Kozlitina J, Valenti L. PCSK7 gene variation bridges atherogenic dyslipidemia with hepatic inflammation in NAFLD patients. *J Lipid Res.* 2019 Jun;60(6):1144-1153. doi: 10.1194/jlr.P090449. 7
- 37 Lindén D, Ahnmark A, Pingitore P, Ciociola E, Ahlstedt I, Andréasson AC, Sasidharan K, Madeyski-Bengtson K, Zurek M, **Mancina RM**, Lindblom A, Bjursell M, Böttcher G, Ståhlman M, Bohlooly-Y M, Haynes WG, Carlsson B, Graham M, Lee R, Murray S, Valenti L, Bhanot S, Åkerblad P, Romeo S. Pnpla3 silencing with antisense oligonucleotides ameliorates nonalcoholic steatohepatitis and fibrosis in Pnpla3 I148M knock-in mice. *Mol Metab.* 2019 Apr;22:49-61. doi: 10.1016/j.molmet.2019.01.013. 9
- 38 Bailetti D, Bertocchini L, **Mancina RM**, Barchetta I, Capoccia D, Cossu E, Pujia A, Lenzi A, Leonetti F, Cavallo MG, Romeo S, Baroni MG. ANGPTL4 gene E40K variation protects against obesity-associated dyslipidemia in participants with obesity. *Obes Sci Pract.* 2018 Dec 26;5(1):83-90. doi: 10.1002/osp4.311. 0
- 39 Spagnuolo R, Dattilo V, D'Antona L, Cosco C, Talerico R, Ventura V, Conforti F, Camastra C, **Mancina RM**, Catalogna G, Cosco V, Iuliano R, Carbone E, Perrotti N, Amato R, Doldo P. Deregulation of SGK1 in Ulcerative Colitis: A Paradoxical Relationship Between Immune Cells and Colonic Epithelial Cells. *Inflamm Bowel Dis.* 2018 May 18. doi: 10.1093/ibd/izy158. 7
- 40 Dongiovanni P, Meroni M, **Mancina RM**, Baselli G, Rametta R, Pelusi S, Männistö V, Fracanzani AL, Badiali S, Miele L, Grimaudo S, Petta S, Bugianesi E, Soardo G, Fargion S, Pihlajamäki J, Romeo S, Valenti L. Protein phosphatase 1 regulatory subunit 3B gene variation protects against hepatic fat accumulation and fibrosis in individuals at high risk of nonalcoholic fatty liver disease. *Hepatol Commun.* 2018 Apr 25;2(6):666-675. doi: 10.1002/hep4.1192. 6
- 41 Mardinoglu A, Wu H, Bjornson E, Zhang C, Hakkarainen A, Räsänen SM, Lee S, **Mancina RM**, Bergentall M, Pietiläinen KH, Söderlund S, Matikainen N, Ståhlman M, Bergh PO, Adiels M, Piening BD, Granér M, Lundbom N, Williams KJ, Romeo S, Nielsen J, Snyder M, Uhlén M, Bergström G, Perkins R, Marschall HU, Bäckhed F, Taskinen MR, Borén J. An Integrated Understanding of the Rapid Metabolic Benefits of a Carbohydrate-Restricted Diet on Hepatic Steatosis in Humans. *Cell Metab.* 2018 Mar 6;27(3):559-571.e5. doi: 10.1016/j.cmet.2018.01.005. Epub 2018 Feb 15. 31

- 42 Caddeo A, **Mancina RM**, Pirazzi C, Russo C, Sasidharan K, Sandstedt J, Maurotti S, Montalcini T, Pujia A, Leren TP, Romeo S, Pingitore P. Molecular analysis of three known and one novel LPL variants in patients with type I hyperlipoproteinemia. *Nutr Metab Cardiovasc Dis*. 2018 Feb;28(2):158-164. doi: 10.1016/j.numecd.2017.11.003. 5
- 43 Donati B, Pietrelli A, Pingitore P, Dongiovanni P, Caddeo A, Walker L, Baselli G, Pelusi S, Rosso C, Vanni E, Daly A, **Mancina RM**, Grieco A, Miele L, Grimaudo S, Craxi A, Petta S, De Luca L, Maier S, Soardo G, Bugianesi E, Colli F, Romagnoli R, Anstee QM, Reeves HL, Fracanzani AL, Fargion S, Romeo S, Valenti L. Telomerase reverse transcriptase germline mutations and hepatocellular carcinoma in patients with nonalcoholic fatty liver disease. *Cancer Med*. 2017 Aug;6(8):1930-1940. doi: 10.1002/cam4.1078. 5
- 44 Taskinen MR, Soderlund S, Bogl LH, Hakkarainen A, Matikainen N, Pietilainen KH, Rasanen S, Lundbom N, Bjornson E, Eliasson B, **Mancina RM**, Romeo S, Almeras N, Della Pepa G, Vetrani C, Prinster A, Annuzzi G, Rivellese A, Despres JP and Boren J. Adverse effects of fructose on cardiometabolic risk factors and hepatic lipid metabolism in subjects with abdominal obesity. *J Intern Med*. 2017 Aug;282(2):187-201. doi: 10.1111/joim.12632. 13
- 45 Donati B, Dongiovanni P, Romeo S, Meroni M, McCain M, Miele L, Petta S, Maier S, Rosso C, De Luca L, Vanni E, Grimaudo S, Romagnoli R, Colli F, Ferri F, **Mancina RM**, Iruzubieta P, Craxi A, Fracanzani AL, Grieco A, Corradini SG, Aghemo A, Colombo M, Soardo G, Bugianesi E, Reeves H, Anstee QM, Fargion S, Valenti L. MBOAT7 rs641738 variant and hepatocellular carcinoma in non-cirrhotic individuals. *Sci Rep*. 2017 Jul 3;7(1):4492. doi: 10.1038/s41598-017-04991-0. 5
- 46 Spagnuolo R, Cosco C, **Mancina RM**, Ruggiero G, Garieri P, Cosco V, Doldo P. Beta-glucan, inositol and digestive enzymes improve quality of life of patients with inflammatory bowel disease and irritable bowel syndrome. *Eur Rev Med Pharmacol Sci*. 2017 Jun;21(2 Suppl):102-107. 4
- 47 Pingitore P, Dongiovanni P, Motta BM, Meroni M, Lepore SM, **Mancina RM**, Pelusi S, Russo C, Caddeo A, Rossi G, Montalcini T, Pujia A, Wiklund O, Valenti L, Romeo S. PNPLA3 overexpression results in reduction of proteins predisposing to fibrosis. *Hum Mol Genet*. 2016 Dec 1;25(23):5212-5222. doi: 10.1093/hmg/ddw341. 5
- 48 Pingitore P, Lepore SM, Pirazzi C, **Mancina RM**, Motta BM, Valenti L, Berge KE, Retterstøl K, Leren TP, Wiklund O, Romeo S. Identification and characterization of two novel mutations in the LPL gene causing Type I hyperlipoproteinemia. *Journal of Clinical Lipidology*. 2016 Jul-Aug;10(4):816-23. doi: 10.1016/j.jacl.2016.02.015. 5
- 49 Burza MA, Motta BM, **Mancina RM**, Pingitore P, Pirazzi C, Lepore SM, Spagnuolo R, Doldo P, Russo C, Lazzaro V, Fischer J, Berg T, Aghemo A, Cheroni C, De Francesco R, Fargion S, Colombo M, Datz C, Stickel F, Valenti L, Romeo S. DEPDC5 variants increase fibrosis progression in Europeans with chronic hepatitis C virus infection. *Hepatology*. 2016 Feb;63(2):418-27. doi: 10.1002/hep.28322. 17
- 50 Donati B, Motta BM, Pingitore P, Meroni M, Pietrelli A, Alisi A, Petta S, Xing C, Dongiovanni P, del Menico B, Rametta R, **Mancina RM**, Badiali S, Fracanzani AL, Craxi A, Fargion S, Nobili V, Romeo S, Valenti L. The rs2294918 E434K variant modulates PNPLA3 expression and liver damage. *Hepatology*. 2015 Nov 25. doi: 10.1002/hep.28370. 17
- 51 Dongiovanni P, Petta S, Mannisto V, **Mancina RM**, Pipitone R, Karja V, Maggioni M, Kakela P, Wiklund O, Mozzi E, Grimaudo S, Kaminska D, Rametta R, Craxi A, Fargion S, Nobili V, Romeo S, Pihlajamaki J, Valenti L. Statin use and non-alcoholic steatohepatitis in at risk individuals. *J Hepatol*. 2015 Sep;63(3):705-12. doi: 10.1016/j.jhep.2015.05.006. 30
- 52 Milano M, Aghemo A, **Mancina RM**, Fischer J, Dongiovanni P, De Nicola S, Fracanzani AL, D'Ambrosio R, Maggioni M, De Francesco R, Fargion S, Berg T, Stickel F, Hampe J, Romeo S, Colombo M, Valenti L. Transmembrane 6 superfamily member 2 gene E167K variant impacts on steatosis and liver damage in chronic hepatitis C patients. 17



*Hepatology*. 2015 Mar 29. doi: 10.1002/hep.27811

- 53** Maglio C, **Mancina RM**, Motta BM, Stef M, Pirazzi C, Palacios L, Askaryar N, Borén J, Wiklund O, Romeo S. Genetic diagnosis of familial hypercholesterolaemia by targeted next-generation sequencing. *J Intern Med*. 2014 May 1. doi: 10.1111/joim.12263. 13
- 54** Pingitore P, Pirazzi C, **Mancina RM**, Motta BM, Indiveri C, Pujia A, Montalcini T, Hedfalk K, Romeo S. Recombinant PNPLA3 protein shows triglyceride hydrolase activity and its I148M mutation results in loss of function. *Biochim Biophys Acta*. 2014 Apr;1841(4):574-80. doi: 10.1016/j.bbaliip.2013.12.006. 3
- 55** Pirazzi C, Valenti L, Motta BM, Pingitore P, Hedfalk K, **Mancina RM**, Burza MA, Indiveri C, Ferro Y, Montalcini T, Maglio C, Dongiovanni P, Fargion S, Rametta R, Pujia A, Andersson L, Ghosal S, Levin M, Wiklund O, Iacovino M, Borén J, Romeo S. PNPLA3 has retinyl-palmitate lipase activity in human hepatic stellate cells. *Hum Mol Genet*. 2014 Mar 30. PMID: 24670599 5
- 56** Maglio C, Ericson U, Burza MA, **Mancina RM**, Pirazzi C, Assarsson JA, Sjöholm K, Baroni MG, Svensson PA, Montalcini T, Pujia A, Sjöström L, Wiklund O, Carlsson LM, Borén J, Orho-Melander M, Romeo S. The IRS1 rs2943641 variant and risk of future cancer among morbidly obese individuals. *J Clin Endocrinol Metab*. 2013 Apr;98(4):E785-9. doi: 10.1210/jc.2012-2831. 6
- 57** Pirazzi C, Adiels M, Burza MA, **Mancina RM**, Levin M, Ståhlman M, Taskinen MR, Orho-Melander M, Perman J, Pujia A, Andersson L, Maglio C, Montalcini T, Wiklund O, Borén J, Romeo S. Patatin-like phospholipase domain-containing 3 (PNPLA3) I148M (rs738409) affects hepatic VLDL secretion in humans and in vitro. *J Hepatol*. 2012 Dec;57(6):1276-82. doi: 10.1016/j.jhep.2012.07.030. 30
- 58** Burza MA, Pirazzi C, Maglio C, Sjöholm K, **Mancina RM**, Svensson PA, Jacobson P, Adiels M, Baroni MG, Borén J, Ginanni Corradini S, Montalcini T, Sjöström L, Carlsson LMS, Romeo S. PNPLA3 I148M (rs738409) genetic variant is associated with hepatocellular carcinoma in obese individuals. *Dig Liver Dis*. 2012 Dec;44(12):1037-41. doi: 10.1016/j.dld.2012.05.006. 5

#### INVITED EDITORIALS

- 59** **Mancina RM**, Spagnuolo R. Cross talk between liver and adipose tissue: A new role for PNPLA3? *Liver Int*. 2020 Sep;40(9):2074-2075. doi: 10.1111/liv.14561. PMID: 32930522 9
- 60** **Mancina RM**, Pingitore P. Natural Extracts as Modifiers of Intracellular Lipid Handling. *Ann Hepatol*. 2018 Mar 1;17(2):180-181. doi: 10.5604/01.3001.0010.8630. PMID: 29469050 3

#### REVIEW

- 61** Sadiea RZ, Sultana S, Chaki BM, Islam T, Dash S, Akter S, Islam MS, Kazi T, Nagata A, Spagnuolo R, **Mancina RM**, Hossain MG. Phytomedicines to Target Hepatitis B Virus DNA Replication: Current Limitations and Future Approaches. *Int J Mol Sci*. 2022 Jan 30;23(3):1617. doi: 10.3390/ijms23031617. PMID: 35163539. 6
- 62** Dongiovanni P, Donati B, Fares R, Lombardi R, **Mancina RM**, Romeo S, Valenti L. PNPLA3 I148M polymorphism and progressive liver disease. *World J Gastroenterol*. 2013 Nov 7;19(41):6969-6978. PMID: 24222941 5