

INFORMAZIONI PERSONALI Paola Concolino

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INCARICO ATTUALE

Dirigente Sanitario Biologo a tempo indeterminato con incarico di alta specialità presso la UOC di Chimica, Biochimica e Biologia Molecolare Clinica della Fondazione Policlinico Universitario A. Gemelli, IRCCS Roma.

ISTRUZIONE E FORMAZIONE

16/06/2014 Abilitazione Scientifica Nazionale Seconda Fascia Settore Disciplinare 05/E1 Biochimica Generale e Biochimica Clinica. (Validità: 16/06/2014-16/06/2025).

27/03/2015 Conseguimento del titolo di Dottore di Ricerca in Biochimica e Biologia Molecolare Clinica, Università Cattolica del Sacro Cuore, Roma. Voto: 50/50 e lode

Tesi: Concolino P, Mello E, Minucci A, Giardina B, Capoluongo E. Genes, pseudogenes and like genes: the case of 21-hydroxylase in Italian population. Clin Chim Acta. 2013 Sep 23;424:85-9. doi: 10.1016/j.cca.2013.05.019.

31/10/2009 Conseguimento del Diploma di Specializzazione in Patologia Clinica, Università degli Studi di Roma "La Sapienza" Voto: 70/70 e lode

Tesi: Concolino P. et al. A new CYP21A1P/CYP21A2 chimeric gene identified in an Italian woman suffering from classical congenital adrenal hyperplasia form. BMC Med Genet. 2009 Jul 22;10:72. doi: 10.1186/1471-2350-10-72.

16/01/2007 Abilitazione all'esercizio della Professione di Biologo, Università "La Tuscia", Viterbo.

18/07/2003 Laurea in Scienze Biologiche (indirizzo BIOTECNOLOGICO), Università degli Studi di Roma "La Sapienza". Voto: 110/110 e lode.

Tesi: Concolino P. et al. The unsolved enigma of CDH1 down-regulation in hereditary diffuse gastric cancer. J Surg Res. 2004 Sep;121(1):50-5. DOI: 10.1016/j.jss.2004.03.008

ESPERIENZA LAVORATIVA

01/01/2020-ad oggi Dirigente Biologo a tempo indeterminato con incarico di alta specialità presso la UOC di Chimica, Biochimica e Biologia Molecolare Clinica della Fondazione Policlinico Universitario A. Gemelli, IRCCS Roma.

01/06/2013-01/01/2020 Dirigente Biologo a tempo indeterminato presso la UOC di Chimica, Biochimica e Biologia Molecolare Clinica della Fondazione Policlinico Universitario A. Gemelli, IRCCS Roma.

01/11/2008-31/12/2012 Dirigente Biologo a tempo determinato, presso UOC di Chimica, Biochimica e Biologia Molecolare Clinica della Fondazione Policlinico Universitario A. Gemelli, IRCCS Roma.

ATTIVITA' DIDATTICA

Incarico di insegnamento- METABOLOMICA presso la FACOLTA' DI MEDICINA E CHIRURGIA "A.GEMELLI" nel Dottorato in SCIENZE BIOMEDICHE DI BASE E SANITA' PUBBLICA presso la sede di Roma dell'Università Cattolica del Sacro Cuore a.a. 2023/2024-2024/2025

Incarico di insegnamento- BIOCHIMICA E BIOLOGIA MOLECOLARE CLINICA presso la FACOLTA' DI SCIENZE nel Corso di Laurea magistrale in Biologia Molecolare, Cellulare E Della Salute presso Università degli Studi Roma Tre, Roma a.a. 2024/2025

Incarico di insegnamento - LABORATORIO DI BIOCHIMICA E BIOLOGIA MOLECOLARE CLINICA presso la FACOLTA' DI MEDICINA E CHIRURGIA "A. GEMELLI" nel Corso di Laurea magistrale in BIOTECNOLOGIE PER LAMEDICINA PERSONALIZZATA presso la sede di Roma dell'Università Cattolica del Sacro Cuore a.a. 2020/2021; 2021/2022; 2022/2023; 2023/2024; 2024/2025

Incarico di insegnamento - BIOLOGIA MOLECOLARE CLINICA presso la FACOLTA' DI MEDICINA E CHIRURGIA "A. GEMELLI" nel Corso di Laurea triennale in Tecniche di laboratorio biomedico (abilitante alla professione sanitaria di Tecnico di laboratorio biomedico) presso la sede di Roma dell'Università Cattolica del Sacro Cuore a.a 2023/2024; 2024/2025

Incarico insegnamento - NUOVE TECNOLOGIE DIAGNOSTICHE PER L'ANALISI GENOMICA presso la FACOLTA' DI MEDICINA E CHIRURGIA "A.GEMELLI" nel Corso di Laurea triennale in Biotecnologie sanitarie presso la sede di Roma dell'Università Cattolica del Sacro Cuore a.a. . 2018/2019; 2019/2020.

PUBBLICAZIONI SCIENTIFICHE

1: Concolino P, De Paolis E, Rinelli M, Maneri G, Brisighelli F, Trozzi R, Duranti S, Giacò L, Piane M, Preziosi A, Panfili A, Scambia G, Nero C, De Bonis M, Minucci A. Identification of a False-positive Multiplex Ligation dependent Probe Amplification Result in *BRCA1* Using a Copy Number Variation Algorithm Under Development for a Commercial Next-Generation Sequencing-based Homologous Recombination Deficiency Assay. Ann Lab Med. 2024 May 31. doi: 10.3343/alm.2024.0051. Epub ahead of print. PMID: 38826062.

2: Concolino P. Challenging Molecular Diagnosis of Congenital Adrenal Hyperplasia (CAH) Due to 21-Hydroxylase Deficiency: Case Series and Novel Variants of *CYP21A2* Gene. Curr Issues Mol Biol. 2024 May 16;46(5):4832-4844. doi: 10.3390/cimb46050291. PMID: 38785559; PMCID: PMC11119849.

3: Concolino P. Chimeric Genes Causing 11 β -Hydroxylase Deficiency: Implications in Clinical and Molecular Diagnosis. Mol Diagn Ther. 2024 Mar;28(2):215-224. doi: 10.1007/s40291-024-00697-y. Epub 2024 Feb 7. PMID: 38324138.

4: Pasquetti D, Gazzellone A, Rossi S, Orteschi D, L'Erario FF, Concolino P, Minucci A, Dionisi-Vici C, Genuardi M, Silvestri G, Chiurazzi P. Triple Genetic

Diagnosis in a Patient with Late-Onset Leukodystrophy and Mild Intellectual Disability. *Int J Mol Sci*. 2023 Dec 29;25(1):495. doi: 10.3390/ijms25010495. PMID: 38203665; PMCID: PMC10778870.

5: De Paolis E, Tilocca B, Lombardi C, De Bonis M, Concolino P, Onori ME, Ricciardi Tenore C, Perrucci A, Roncada P, Capoluongo E, Urbani A, Minucci A, Santonocito C. Next-Generation Sequencing for Screening Analysis of Cystic Fibrosis: Spectrum and Novel Variants in a South-Central Italian Cohort. *Genes (Basel)*. 2023 Aug 11;14(8):1608. doi: 10.3390/genes14081608. PMID: 37628659; PMCID: PMC10454170.

6: Concolino P, Perrucci A, Carrozza C, Urbani A. Genetic Characterization of a Cohort of Italian Patients with Congenital Adrenal Hyperplasia Due to 21-Hydroxylase Deficiency. *Mol Diagn Ther*. 2023 Sep;27(5):621-630. doi: 10.1007/s40291-023-00666-x. Epub 2023 Aug 7. PMID: 37548905.

7: Concolino P, De Paolis E, Moffa S, Onori ME, Soldovieri L, Ricciardi Tenore C, De Bonis M, Rabacchi C, Santonocito C, Rinelli M, Calandra S, Giaccari A, Urbani A, Minucci A. Identification and Molecular Characterization of a Novel Large-Scale Variant (Exons 4_18 Loss) in the LDLR Gene as a Cause of Familial Hypercholesterolaemia in an Italian Family. *Genes (Basel)*. 2023 Jun 16;14(6):1275. doi: 10.3390/genes14061275. PMID: 37372455; PMCID: PMC10298720.

8: Concolino P, Tartaglione L, De Paolis E, Carrozza C, Urbani A, Minucci A, Pitocco D, Santonocito C. A Novel *GCK* Large Genomic Rearrangement in a Patient with MODY-2 Detected by Clinical Exome Sequencing. *Genes (Basel)*. 2022 Nov 13;13(11):2104. doi: 10.3390/genes13112104. PMID: 36421779; PMCID: PMC9690203.

9: Rossi S, Concolino P, Di Natale D, Pasquetti D, Di Lella GM, Chiurazzi P, Silvestri G. Clinical Reasoning: A Young Man With Subacute Onset of Spastic Paraparesis. *Neurology*. 2023 Jan 24;100(4):199-205. doi: 10.1212/WNL.0000000000201516. Epub 2022 Oct 27. PMID: 36302663.

10: Cera G, Locantore P, Novizio R, Maggio E, Ramunno V, Corsello A, Policola C, Concolino P, Paragliola RM, Pontecorvi A. Pregnancy and Prenatal Management of Congenital Adrenal Hyperplasia. *J Clin Med*. 2022 Oct 19;11(20):6156. doi: 10.3390/jcm11206156. PMID: 36294476; PMCID: PMC9605322.

11: Pavese F, Capoluongo ED, Muratore M, Minucci A, Santonocito C, Fuso P, Concolino P, Di Stasio E, Carbognin L, Tiberi G, Garganese G, Corrado G, Di Leone A, Generali D, Fragomeni SM, D'Angelo T, Franceschini G, Masetti R, Fabi A, Mulè A, Santoro A, Belli P, Tortora G, Scambia G, Paris I. BRCA Mutation Status in Triple-Negative Breast Cancer Patients Treated with Neoadjuvant Chemotherapy: A Pivotal Role for Treatment Decision-Making. *Cancers (Basel)*. 2022 Sep 21;14(19):4571. doi: 10.3390/cancers14194571. PMID: 36230495; PMCID: PMC9559391.

12: Paragliola RM, Perrucci A, Foca L, Urbani A, Concolino P. Prevalence of CAH-X Syndrome in Italian Patients with Congenital Adrenal Hyperplasia (CAH) Due to 21-Hydroxylase Deficiency. *J Clin Med*. 2022 Jul 1;11(13):3818. doi: 10.3390/jcm11133818. PMID: 35807105; PMCID: PMC9267771.

13: Concolino P, Falhammar H. CAH-X Syndrome: Genetic and Clinical Profile. *Mol Diagn Ther*. 2022 May;26(3):293-300. doi: 10.1007/s40291-022-00588-0. Epub 2022 Apr 27. PMID: 35476220.

14: De Paolis E, Marchetti C, Concolino P, Scambia G, Urbani A, Fagotti A, Minucci A. A commentary on the discrepancy between blood and tumour BRCA testing: An open question. *BJOG*. 2022 Aug;129(9):1422-1426. doi: 10.1111/1471-0528.17158. Epub 2022 Apr 5. PMID: 35319826; PMCID: PMC9543799.

15: Kocova M, Concolino P, Falhammar H. Characteristics of In2G Variant in Congenital Adrenal Hyperplasia Due to 21-Hydroxylase Deficiency. *Front Endocrinol (Lausanne)*. 2022 Jan 24;12:788812. doi: 10.3389/fendo.2021.788812. PMID: 35140681; PMCID: PMC8818746.

16: De Paolis E, Concolino P, Onori ME, Santonocito C, Marchetti C, Fagotti A,

Scambia G, Urbani A, Minucci A. Tumor BRCA testing in ovarian cancer and EQA scheme: our experience of a critical evaluation. *Mol Biol Rep*. 2021 Dec;48(12):8203-8209. doi: 10.1007/s11033-021-06812-0. Epub 2021 Oct 13. PMID: 34643925; PMCID: PMC8604882.

17: Carrozza C, Foca L, De Paolis E, Concolino P. Genes and Pseudogenes: Complexity of the RCCX Locus and Disease. *Front Endocrinol (Lausanne)*. 2021 Jul 30;12:709758. doi: 10.3389/fendo.2021.709758. PMID: 34394006; PMCID: PMC8362596.

18: De Paolis E, Paragliola RM, Concolino P. Spectrum of *DICER1* Germline Pathogenic Variants in Ovarian Sertoli-Leydig Cell Tumor. *J Clin Med*. 2021 Apr 23;10(9):1845. doi: 10.3390/jcm10091845. PMID: 33922805; PMCID: PMC8123016.

19: De Angelis C, Nardelli C, Concolino P, Pagliuca M, Setaro M, De Paolis E, De Placido P, Forestieri V, Scaglione GL, Ranieri A, Lombardo B, Pastore L, De Placido S, Capoluongo E. Case Report: Detection of a Novel Germline *PALB2* Deletion in a Young Woman With Hereditary Breast Cancer: When the Patient's Phenotype History Doesn't Lie. *Front Oncol*. 2021 Feb 24;11:602523. doi: 10.3389/fonc.2021.602523. PMID: 33718150; PMCID: PMC7943848.

20: Concolino P, Paragliola RM. Molecular Analysis of 21-Hydroxylase Deficiency Reveals Two Novel Severe Genotypes in Affected Newborns. *Mol Diagn Ther*. 2021 May;25(3):327-337. doi: 10.1007/s40291-021-00520-y. Epub 2021 Mar 12. PMID: 33710594.

21: De Paolis E, Urbani A, Salvatore L, Foca L, Tortora G, Minucci A, Concolino P. A Novel *ATM* Pathogenic Variant in an Italian Woman with Gallbladder Cancer. *Genes (Basel)*. 2021 Feb 22;12(2):313. doi: 10.3390/genes12020313. PMID: 33671809; PMCID: PMC7926430.

22: Minucci A, Scambia G, De Bonis M, De Paolis E, Santonocito C, Fagotti A, Capoluongo E, Concolino P, Urbani A. BRCA testing delay during the COVID-19 pandemic: How to act? *Mol Biol Rep*. 2021 Jan;48(1):983-987. doi: 10.1007/s11033-020-06060-8. Epub 2020 Dec 12. PMID: 33313973; PMCID: PMC7733534.

23: De Paolis E, De Bonis M, Concolino P, Piermattei A, Fagotti A, Urbani A, Scambia G, Minucci A, Capoluongo E. Droplet digital PCR for large genomic rearrangements detection: A promising strategy in tissue BRCA1 testing. Clin Chim Acta. 2021 Feb;513:17-24. doi: 10.1016/j.cca.2020.12.001. Epub 2020 Dec 7. PMID: 33301768.

24: Corsello A, Bruno C, Rizza R, Concolino P, Papi G, Pontecorvi A, Rindi G, Paragliola RM. A novel MEN1 pathogenic variant in an Italian patient with multiple endocrine neoplasia type 1. Mol Biol Rep. 2020 Sep;47(9):7313-7316. doi: 10.1007/s11033-020-05730-x. Epub 2020 Aug 17. PMID: 32808116; PMCID: PMC7430936.

25: Paragliola RM, Costella A, Corsello A, Urbani A, Concolino P. A Novel Pathogenic Variant in the N-Terminal Domain of the Glucocorticoid Receptor, Causing Glucocorticoid Resistance. Mol Diagn Ther. 2020 Aug;24(4):473-485. doi: 10.1007/s40291-020-00480-9. PMID: 32607951.

26: Minucci A, Scambia G, Santonocito C, Concolino P, Urbani A. BRCA testing in a genomic diagnostics referral center during the COVID-19 pandemic. Mol Biol Rep. 2020 Jun;47(6):4857-4860. doi: 10.1007/s11033-020-05479-3. Epub 2020 May 9. PMID: 32388698; PMCID: PMC7210797.

27: Concolino P. A rare CYP21A2 haplotype clarifies the phenotype-genotype discrepancy in an Italian patient with Non Classical Congenital Adrenal Hyperplasia (NC-CAH). Mol Biol Rep. 2020 Apr;47(4):3049-3052. doi: 10.1007/s11033-020-05379-6. Epub 2020 Mar 17. PMID: 32185686.

28: Minucci A, Mazzuccato G, D'Indinosante M, Di Nardo L, Concolino P, De Bonis M, Urbani A, Scambia G, Fagotti A, Capoluongo E. Spliceogenic analysis of BRCA1 c.439T>C (rs794727800) variant by High Resolution Melting Analysis. Mol Biol Rep. 2020 Feb;47(2):1513-1520. doi: 10.1007/s11033-019-05199-3. Epub 2019 Dec 12. PMID: 31833030.

29: Paragliola RM, Corsello A, Concolino P, Ianni F, Papi G, Pontecorvi A, Corsello SM. Iodothyronine deiodinases and reduced sensitivity to thyroid hormones. *Front Biosci (Landmark Ed)*. 2020 Jan 1;25(2):201-228. doi: 10.2741/4803. PMID: 31585886.

30: Concolino P, Capoluongo E. Detection of *BRCA1/2* large genomic rearrangements in breast and ovarian cancer patients: an overview of the current methods. *Expert Rev Mol Diagn*. 2019 Sep;19(9):795-802. doi: 10.1080/14737159.2019.1657011. Epub 2019 Aug 28. PMID: 31429350.

31: Concolino P, Gelli G, Rizza R, Costella A, Scambia G, Capoluongo E. *BRCA1* and *BRCA2* Testing through Next Generation Sequencing in a Small Cohort of Italian Breast/Ovarian Cancer Patients: Novel Pathogenic and Unknown Clinical Significance Variants. *Int J Mol Sci*. 2019 Jul 12;20(14):3442. doi: 10.3390/ijms20143442. PMID: 31336956; PMCID: PMC6678297.

32: Concolino P. Issues with the Detection of Large Genomic Rearrangements in Molecular Diagnosis of 21-Hydroxylase Deficiency. *Mol Diagn Ther*. 2019 Oct;23(5):563-567. doi: 10.1007/s40291-019-00415-z. PMID: 31317337.

33: Chiloiro S, Capoluongo ED, Schinzari G, Concolino P, Rossi E, Martini M, Cocomazzi A, Grande G, Milardi D, Maiorano BA, Giampietro A, Rindi G, Pontecorvi A, De Marinis L, Bianchi A. First Case of Mature Teratoma and Yolk Sac Testis Tumor Associated to Inherited MEN-1 Syndrome. *Front Endocrinol (Lausanne)*. 2019 Jun 12;10:365. doi: 10.3389/fendo.2019.00365. PMID: 31249555; PMCID: PMC6582702.

34: Parsons MT, Tadini E, Li H, Hahnen E, Wappenschmidt B, Feliubadaló L, Aalfs CM, Agata S, Aittomäki K, Alducci E, Alonso-Cerezo MC, Arnold N, Auber B, Austin R, Azzollini J, Balmaña J, Barbieri E, Bartram CR, Blanco A, Blümcke B, Bonache S, Bonanni B, Borg Å, Bortesi B, Brunet J, Bruzzone C, Bucksch K, Cagnoli G, Caldés T, Caliebe A, Caligo MA, Calvello M, Capone GL, Caputo SM, Carnevali I, Carrasco E, Caux-Moncoutier V, Cavalli P, Cini G, Clarke EM, Concolino P, Cops EJ, Cortesi L, Couch FJ, Darder E, de la Hoya M, Dean M, Debatin I, Del Valle J, Delnatte C, Derive N, Diez O, Ditsch N, Domchek SM, Dutrannoy V, Eccles DM,

Ehrencrona H, Enders U, Evans DG, Farra C, Faust U, Felbor U, Feroce I, Fine M, Foulkes WD, Galvao HCR, Gambino G, Gehrig A, Gensini F, Gerdes AM, Germani A, Giesecke J, Gismondi V, Gómez C, Gómez Garcia EB, González S, Grau E, Grill S, Gross E, Guerrieri-Gonzaga A, Guillaud-Bataille M, Gutiérrez-Enríquez S, Haaf T, Hackmann K, Hansen TVO, Harris M, Hauke J, Heinrich T, Hellebrand H, Herold KN, Honisch E, Horvath J, Houdayer C, Hübbel V, Iglesias S, Izquierdo A, James PA, Janssen LAM, Jeschke U, Kaulfuß S, Keupp K, Kiechle M, Kölbl A, Krieger S, Kruse TA, Kvist A, Laloo F, Larsen M, Lattimore VL, Lautrup C, Ledig S, Leinert E, Lewis AL, Lim J, Loeffler M, López-Fernández A, Lucci-Cordisco E, Maass N, Manoukian S, Marabelli M, Matricardi L, Meindl A, Michelli RD, Moghadasi S, Moles-Fernández A, Montagna M, Montalban G, Monteiro AN, Montes E, Mori L, Moserle L, Müller CR, Mundhenke C, Naldi N, Nathanson KL, Navarro M, Nevanlinna H, Nichols CB, Niederacher D, Nielsen HR, Ong KR, Pachter N, Palmero EI, Papi L, Pedersen IS, Peissel B, Perez-Segura P, Pfeifer K, Pineda M, Pohl-Rescigno E, Poplawski NK, Porfirio B, Quante AS, Ramser J, Reis RM, Revillion F, Rhiem K, Riboli B, Ritter J, Rivera D, Rofes P, Rump A, Salinas M, Sánchez de Abajo AM, Schmidt G, Schoenwiese U, Seggewiß J, Solanes A, Steinemann D, Stiller M, Stoppa-Lyonnet D, Sullivan KJ, Susman R, Sutter C, Tavtigian SV, Teo SH, Teulé A, Thomassen M, Tibiletti MG, Tischkowitz M, Tognazzo S, Toland AE, Tornero E, Törngren T, Torres-Esquius S, Toss A, Trainer AH, Tucker KM, van Asperen CJ, van Mackelenbergh MT, Varesco L, Vargas-Parra G, Varon R, Vega A, Velasco Á, Vesper AS, Viel A, Vreeswijk MPG, Wagner SA, Waha A, Walker LC, Walters RJ, Wang-Gohrke S, Weber BHF, Weichert W, Wieland K, Wiesmüller L, Witzel I, Wöckel A, Woodward ER, Zachariae S, Zampiga V, Zeder-Göß C; KConFab Investigators; Lázaro C, De Nicolo A, Radice P, Engel C, Schmutzler RK, Goldgar DE, Spurdle AB. Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. *Hum Mutat.* 2019 Sep;40(9):1557-1578. doi: 10.1002/humu.23818. PMID: 31131967; PMCID: PMC6772163.

35: Concolino P, Costella A, Paragliola RM. Mutational Landscape of Resistance to Thyroid Hormone Beta (RTH β). *Mol Diagn Ther.* 2019 Jun;23(3):353-368. doi: 10.1007/s40291-019-00399-w. PMID: 30976996.

36: Rizza R, Hackmann K, Paris I, Minucci A, De Leo R, Schrock E, Urbani A,

Capoluongo E, Gelli G, Concolino P. Novel BRCA1 Large Genomic Rearrangements in Italian Breast/Ovarian Cancer Patients. *Mol Diagn Ther*. 2019 Feb;23(1):121-126. doi: 10.1007/s40291-018-0376-2. PMID: 30506513.

37: Minucci A, Lalle M, De Leo R, Mazzuccato G, Scambia G, Urbani A, Fagotti A, Concolino P, Capoluongo E. Additional molecular and clinical evidence open the way to definitive IARC classification of the BRCA1 c.5332G > A (p.Asp1778Asn) variant. *Clin Biochem*. 2019 Jan;63:54-58. doi: 10.1016/j.clinbiochem.2018.10.004. Epub 2018 Oct 10. PMID: 30315757.

38: Palermo A, Capoluongo E, Del Toro R, Manfrini S, Pozzilli P, Maggi D, Defeudis G, Pantano F, Coppola R, Di Matteo FM, Raffaelli M, Concolino P, Falchetti A. A novel germline mutation at exon 10 of MEN1 gene: a clinical survey and positive genotype-phenotype analysis of a MEN1 Italian family, including monozygotic twins. *Hormones (Athens)*. 2018 Sep;17(3):427-435. doi: 10.1007/s42000-018-0044-2. Epub 2018 Aug 6. PMID: 30083881.

39: Costella A, De Leo R, Guarino D, D'Indinosante M, Concolino P, Mazzuccato G, Urbani A, Scambia G, Capoluongo E, Fagotti A, Minucci A. High-resolution melting analysis coupled with next-generation sequencing as a simple tool for the identification of a novel somatic BRCA2 variant: a case report. *Hum Genome Var*. 2018 Jun 8;5:10. doi: 10.1038/s41439-018-0006-x. PMID: 29899995; PMCID: PMC5993729.

40: Papi G, Paragliola RM, Concolino P, Di Donato C, Pontecorvi A, Corsello SM. 46,XY Disorder of Sex Development Caused by 17 α -Hydroxylase/17,20-Lyase Deficiency due to Homozygous Mutation of *CYP17A1* Gene: Consequences of Late Diagnosis. *Case Rep Endocrinol*. 2018 Apr 24;2018:2086861. doi: 10.1155/2018/2086861. PMID: 29854486; PMCID: PMC5941809.

41: Minucci A, Concolino P, De Bonis M, Costella A, Paris I, Scambia G, Capoluongo E. Preliminary molecular evidence associating a novel BRCA1 synonymous variant with hereditary ovarian cancer syndrome. *Hum Genome Var*. 2018 Apr 20;5:2. doi: 10.1038/s41439-018-0003-0. PMID: 29760936; PMCID: PMC5938031.

42: Scaglione GL, Concolino P, De Bonis M, De Paolis E, Minucci A, Ferrandina G, Scambia G, Capoluongo E. A Whole Germline BRCA2 Gene Deletion: How to Learn from CNV In Silico Analysis. *Int J Mol Sci*. 2018 Mar 23;19(4):961. doi: 10.3390/ijms19040961. PMID: 29570666; PMCID: PMC5979302.

43: Concolino P, Rizza R, Mignone F, Costella A, Guarino D, Carboni I, Capoluongo E, Santonocito C, Urbani A, Minucci A. A comprehensive BRCA1/2 NGS pipeline for an immediate Copy Number Variation (CNV) detection in breast and ovarian cancer molecular diagnosis. *Clin Chim Acta*. 2018 May;480:173-179. doi: 10.1016/j.cca.2018.02.012. Epub 2018 Feb 16. PMID: 29458049.

44: Concolino P, Costella A. Congenital Adrenal Hyperplasia (CAH) due to 21-Hydroxylase Deficiency: A Comprehensive Focus on 233 Pathogenic Variants of CYP21A2 Gene. *Mol Diagn Ther*. 2018 Jun;22(3):261-280. doi: 10.1007/s40291-018-0319-y. PMID: 29450859.

45: Santonocito C, Scapaticci M, Guarino D, Bartolini A, Minucci A, Concolino P, Scambia G, Paris I, Capoluongo E. Identification of twenty-nine novel germline unclassified variants of BRCA1 and BRCA2 genes in 1400 Italian individuals. *Breast*. 2017 Dec;36:74-78. doi: 10.1016/j.breast.2017.09.007. Epub 2017 Oct 8. PMID: 29020660.

46: Concolino P, Rizza R, Hackmann K, Minucci A, Scaglione GL, De Bonis M, Costella A, Zuppi C, Schrock E, Capoluongo E. Identification and Characterization of a New BRCA2 Rearrangement in an Italian Family with Hereditary Breast and Ovarian Cancer Syndrome. *Mol Diagn Ther*. 2017 Oct;21(5):539-545. doi: 10.1007/s40291-017-0288-6. PMID: 28620890.

47: Concolino P, Rizza R, Costella A, Carrozza C, Zuppi C, Capoluongo E. CYP21A2 intronic variants causing 21-hydroxylase deficiency. *Metabolism*. 2017 Jun;71:46-51. doi: 10.1016/j.metabol.2017.03.003. Epub 2017 Mar 9. PMID: 28521877.

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ABSTRACT

Autore di numerosi Abstracts presentati a Congressi Nazionali ed Internazionali.

Il sottoscritto, consapevole che – ai sensi dell’art. 76 del D.P.R. 445/2000 – le dichiarazioni mendaci, la falsità negli atti e l’uso di atti falsi sono puniti ai sensi del Codice penale e delle leggi speciali, dichiara che le informazioni rispondono a verità.

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Firma
