Personal information

- **Born in Padua (PD), Italy on the 24/06/1972**

Current situation

- Biological Science in Department of Oncology, Family Cancer Clinic, Veneto Institute of Oncology IOV - IRCCS,Padua, Italy

Education

- Degree in Biological Science with grade 110/110 cum laude Specialist in Medical Genetics (University of Studies of Ferrara). Title of the thesis: 'Development of a recombinant virus based on the SV40 genome for in vitro gene transfer'
- Specialist in Medical Genetics (50/50) on genetic polymorphisms in folate metabolism enzymes in acute lymphoblastic leukaemia and non-Hodgkin's lymphomas: epidemiological and genetic drug studies. Faculty of Medicine and Surgery, University of Ferrara

Experience From: 2020 to date: Family Cancer Clinic -Veneto Oncology Institute-IOV

- Genetic Oncology Consultancy activities for hereditary neoplastic syndromes: in particular endocrine and neuroendocrine hereditary syndromes in a multidisciplinary team: about 20 visits/year and about 100 visits in the last 5-y of genetic counselling for Von Hippel Lindau Syndrome and 170/y of all other rare syndromes (MEN1-4, MEN2, pheocromocytoma and paraganglioma, hyperparathyroidism). Clinical personal and family history collection are proposal and management of genetic testing also through multi-genetic panels. Activities of management and interpretation of genetic data for taking charge of the high-risk family and cascade testing. Participation every 15 days every 15 days in multidisciplinary specialists groups to discuss a shared approach on VHL patients referred to in our Institute.
- **From 2009 to 2019** IOV- I.R.C. S Immunology and Molecular Oncology Diagnostics Unit: The multidisciplinary counselling activity related to genetic testing aimed at identifying families at high risk of breast and/or ovarian cancer has been integrated with genetic testing of BRCA1 and 2 and related genes in hereditary predisposition syndromes.
- Excellent knowledge of reporting guidelines (gene-specific guidelines, ENIGMA, ACMG, InSIGHT), interpretation and variant classification of genetic tests
- **2006** Holder of AIL (Associazione Italiana Leucemie) scholarship at the Ferrara Hospital, Department of Biomedical Sciences and Advanced Therapies
- Genetic/clinical/biomolecular and pharmacological activities, with follow-up of research projects on the main hereditary coagulopathies, cardiovascular and neoplastic pathologies in collaboration with the Haematology, Cardiology and Ophthalmology Departments. Prenatal diagnosis activities on Haemophilia A/B.
- Genetic/clinical/biomolecular and pharmacologicalstudies on tissue regeneration and repair mechanisms for the treatment of chronic lesions of the lower limbs in collaboration with Prof. Zamboni's Centre for Vascular Diseases. Awarded a 3rd prize at the European Venous Forum, Crete-2005.
- Epidemiological/pharmaceutical genetic studies in acute lymphoblastic leukaemias and non-Hodgkin's lymphomas.
- **2002-2006 Since** 2002 holder of a research grant at the Department of Biomedical Sciences

Advanced Therapies, Centre for Haemostasis and Thrombosis Studies, Ferrara University Hospital.

- Technical skills in molecular genetic:

  Nucleic acid manipulation techniques with extraction, amplification (PCR, PCRReal-Time, LongPCR, Nexted PCR), restriction, multi-capillary sequencing, ASO-Probe, Microarray/Nano-chip, DHPLC, mutational analysis with sequencing by Sanger, MLPA for analysis of large genomic rearrangements, NGS analysis with multigenic panels. Analysis of sequence profiles with specific software

  Biochemistry: Immunohistochemistry, Immunocytochemistry, Electrophoresis, immunoelectrophoresis, ELISA, Western Blot, SSCP, protein assays

  Cell biology: Primary and transformed cell cultures from biopsy samples, microscopy, bacterial plate cultures. Cell survival assays.

  Haemocoagulation techniques: biological and functional protein assays. Diagnostics: mutational analysis in BRCA1 and 2 genes. Prenatal diagnosis and linkage analysis for haemophilia A and B. Diagnosis of pro-thrombotic mutations.

- **Coauthor in 40 scientific publications**

Publication in the last 10y

1.Innella G. et al. Atypical cancer risk profile in carriers of Italian founder BRCA1 variant p.His1673del: Implications for classification and clinical management. Collaborators (Tognazzo et al.) Cancer Med. 2024 Aug;13(16)

2.Saccardi C., Tognazzo S., et al. Efficacy of risk-reducing salpingo-oophorectomy in BRCA1-2 variants and clinical outcomes of follow-up in patients with isolated serous tubal intraepithelial carcinoma (STIC) Gynecol Oncol 2021, Nov 163

3. Barnes DR, Tognazzo S., et al. Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. Genet Med. 2020 Oct;22(10):1653-1666

4. Agata S, Tognazzo S. et al.Segregation analysis of the BRCA2 c.9227G>T variant in multiple families suggests a pathogenic role in breast and ovarian cancer predisposition.Sci Rep. 2020 Aug 19;

5.Patel VL, Tognazzo S. et al. Association of Genomic Domains in *BRCA1* and *BRCA2* with Prostate Cancer Risk and Aggressiveness. Cancer Res. 2020 Feb 1;80(3):624-638.

6. Parson MT, Tognazzo S., et al. 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

7. Qian F, Tognazzo S. et al. Mendelian randomisation study of height and body mass index as modifiers of ovarian cancer risk in 22,588 BRCA1 and BRCA2 mutation carriers. Br J Cancer. 2019

8. Rebbeck TR,Tognazzo S. et al. Mutational spectrum in a worldwide study of 29,700 families with BRCA1 or BRCA2 mutations. Hum Mutat. 2018 May;39(5):593-620

9. Lecarpentier J., Tognazzo S. et al. Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. J Clin Oncol. 2017 Jul 10;35(20):2240-2250

10. Phelan CM, Tognazzo S. et al. Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nat Genet. 2017 May;49(5):680-691

11.Hamdi Y, Tognazzo S. Association of breast cancer risk in BRCA1 and BRCA2 mutation carriers with genetic variants showing differential allelic expression: identification of a modifier of breast cancer risk at locus 11q22.3. Breast Cancer Res Treat. 2017 Jan;161(1):117-134

12.Lawrenson K, Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. Nat Commun. 2016 Sep 7;7:12675.

13.Vigorito E, Tognazzo S. et al.Fine-Scale Mapping at 9p22.2 Identifies Candidate Causal Variants That Modify Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. PLoS One. 2016 Jul 27;

14.Peterlongo P, Tognazzo S. et al FANCM c.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor. Hum Mol Genet. 2015 Sep 15;24(18):5345-55

15.Kuchenbaecker KB, Tognazzo S., et alIdentification of six new susceptibility loci for invasive epithelial ovarian cancer. Nat Genet. 2015 Feb;47(2):164-71

16.Scaini MC., Tognazzo S. et al. CDKN2A unclassified variants in familial malignant melanoma: combining functional and computational approaches for their assessment. Hum Mutat. 2014 Jul;35(7):828-40.

17. Osorio A,Tognazzo S. et al. DNA glycosylases involved in base excision repair may be associated with cancer risk in BRCA1 and BRCA2 mutation carriers. PLoS Genet. 2014 Apr 3;10(4)

- **Attendance at courses and conferences in the last 5y**

16th International VHL medical Symposium 2024

Relatore BRCA in Urologia 2024

XXVII Congresso Nazionale SIGU 2024

34° Incontro di Genetica Oncologica Clinica 2023

Congresso Nazionale AIFET 2023

Malattia di Von Hippel Lindau (VHL): ricerca e terapia del carcinoma a cellule chiare del rene (ccRCC) nella malattia di VHL 2023

Tumori Ereditari: dai geni alle Sindromi Endocrine Multiple: Tumori neuroendocrini e melanoma familiare 2023

Congresso Nazionale di Neoplasia Endocrina Multipla di Tipo 1 2023

Relatore MASTERCLASS Gestione del paziente con Carcinoma della Prostata: la Prostate Unit 2023

Oncogenetica-nuove implicazioni cliniche, aspetti psicologici e bioetici 2023

Minicounseling: Definizione di un percorso nel paziente con carcinoma prostatico mutato 2023. Componente del gruppo di lavoro

Riflessione e approfondimento del proprio operato malattie genetiche: dalla diagnosi al trattamento 2022

Forum Nazionale OncoGenEtica 2022

Primo Congresso Associazione Italiana Familiarità Ereditarietà dei Tumori AIFET 2022

32° Incontro di Genetica Clinica 2022

Relatore Convegno Nazionale GISMA 2022

Relatore XX Convegno Nazionale A.G.E.O 2022

Relatore Padova Breast Meeting 2021

Prevenzione e presa in carico dei pazienti affetti da carcinoma non midollare familiare della tiroide FNMTC 2019

The Healthy BRCA-carrier-Sorveglianza e prevenzione del tumore della mammella e dell'ovaio 2020

- **Member of**

Multidisciplinary working group to update the clinical-diagnostic operational pathway for the identification, diagnosis, surveillance and prevention of healthy subjects carrying pathogenic variants for hereditary breast and ovarian cancer 2024

Multidisciplinary working group for Von Hippel Lindau Syndrome: discussion of clinical cases 2021-2024

Working group for the definition of the PDTA for breast-ovarian cancer patients (2017 and 2023)

Working Group 'Management of people at high risk of cancer -PPR Screening Member of the Breast Unit multidisciplinary group for the discussion of breast-ovarian clinical cases since 2009

Hereditary gastro-intestinal tumours multidisciplinary group

SIGU Oncology Genetics Group

Dichiaro di essere informato ai sensi e per gli effetti dell'art. 13 del D.lgs 196/2003, e a tal fine presto il consenso al trattamento dei dati personali ivi contenuti

Dichiaro "sotto la mia responsabilità, ai sensi degli artt. 46 e 47 del D.P.R. 445 del 28.12.2000 e consapevole delle sanzioni penali previste dall'art. 78 del citato D.P.R. per le ipotesi di falsità in atti e di dichiarazioni mendaci, dichiaro che quanto indicato nel presente curriculum corrisponde a verità"

In fede Dott. Silvia Tognazzo