Curriculum Vitae Prof. Silverio Perrotta

Date and place of birth: 14/Nov/1962, Naples (Italy)

## Working Address:

Dipartimento della Donna, del Bambino e di Chirurgia Generale e Specialistica, University of Campania "L. Vanvitelli", Via Luigi De Crecchio, 4 - 80138 Naples (Italy)

## Education and degrees:

1992 – Postgraduate degree with honors in Pediatrics at the Second University of Naples (Italy) 1988 – Graduation degree (M.D.) in Medicine with honors from the School of Medicine of the University Federico II of Naples (Italy) (Medical Board number 23658 – Naples)

## Current positions and past appointments:

2018 to present: Full Professor of Pediatrics (ssd MED 38) at Dipartimento della Donna, del Bambino e di Chirurgia Generale e Specialistica, University of Campania "L. Vanvitelli", Naples (Italy);

2014 to 2018: Associate Professor of Pediatrics (ssd MED 38) at Dipartimento della Donna, del Bambino e di Chirurgia Generale e Specialistica, University of Campania "L. Vanvitelli", Naples (Italy);

2001 to 2014: Researcher at Dipartimento della Donna, del Bambino e di Chirurgia Generale e Specialistica, Second University of Naples (Italy);

1992 to 2018: Medical Doctor of Pediatric Hematology and of Rare blood disease center and Hemoglobinopathies of Hospital in University of Campania "L. Vanvitelli"

2018 to present: Medical Director of Center of Pediatric Hematology and Oncology, of Rare blood diseases, of Hemoglobinopathies and of Neufibromatosis, DAI Materno Infantile, University of Campania "L. Vanvitelli".

2017 to present: Director of the Graduate School in Pediatrics "L. Vanvitelli" Campania University

## Clinical Stages:

1992 Center of bone marrow transplantation, Hospital of Muraglia, Pesaro, Italy

He is an active member of the following clinical society and associations: Società Italiana di Emoglobinopatie e Talassemie (SITE) Italian Society of Pediatric Hematology/Oncology (AIEOP) European Cooperation in Science and Technology (COST) Hypoxia group

He has been the Chief of AIEOP Study Group of red blood cell for 8 years and the member of the AIEOP board from 2010 to 2012.

He is a Representative EuroBloodNet Member (the European reference network for rare hematological disorder). Healthcare Provider: AOU Università degli Studi della Campania "Luigi Vanvitelli".

He is a reviewer for the American Journal of Hematology, British Journal of Haematology, Haematologica and Blood.

He has been teaching Paediatrics since 2001 in several programs of the Medical School, and at the residency programs in Paediatrics at the University of Naples. He has been tutoring undergrads, residents and post-doctoral fellows being able to, always, mentor them to their maximum potentials.

Professor of Pediatric Hematology at Advanced Course of Pediatric Hematology, University of Sapienza, Roma (Italy)

He has been involved, as a Principal investigator, in 51 Clinical trials in the last 12 years, being the proponent and/or coordinator for some of them.

His contribution to the trials' design and the number of patients that were enrolled at his unit led to their success, both as scientific findings as well as new drugs' approvals for the market. All clinical trials were conducted according to the Good Clinical Practice (GCP) guidelines and he is a GCP-certified investigator.

During the years he has been funded with more than 20 regional, national and international grants as a member or a project coordinator that were successfully conducted leading to many ground-breaking scientific and medical discoveries.

In particular:

PRIN Projects (2002, 2004, 2006, 2008) total budget 170.000 Euros

Telethon Project: total budget 140.000 Euros

Valere Project 2019 (Università della Campania "L. Vanvitelli)": total budget 266.000 Euros

He is, indeed, author of 217 peer-reviewed articles with several national and international collaborations that include:

Prof. Joseph T. Prchal, University of Utah, Salt Lake City;

Prof. Victor R Gordeuk, Howard University, Washington;

Prof. Peter Robbins, University of Oxford, England;

Prof. Patrick Gallagher, Yale University School of Medicine, New Haven;

Prof. Narla Mohandas, New York Blood Center, New York;

Prof. Jean Delaunay, CNRS Lyon, France;

Prof. Franco Locatelli, Pediatric hospital Bambino Gesu', Italy;

Prof. Maria Domenica Cappellini, University of Milan, Italy;

Prof. Achille Iolascon, Federico II University, Italy;

Prof. Carlo Dufour, University of Genoa, Italy;

Prof. Carlo Balduini, University of Pavia, Italy;

He has a Total Impact Factor of 1407 with 6992 total citations by 4993 documents (by Scopus). His H-Index is 45

Scientific interests:

thalassaemia and hemoglobinopathies

iron metabolism

red cell membrane disorders (hereditary spherocytosis, hereditary elliptocytosis, congenital dyserythropoietic anaemias, stomatocytosis and related diseases)

Fanconi amemia

Gilbert's syndrome

oncogenes in pediatric tumors

Immune and hereditary thrombocytopenias neurohypophysial diabetes insipidus familial vesicoureteric reflux congenital erythrocytosis neurofibromatosis type 1

The overarching goal of his research program, which has been carried out at the Pediatric Hematology and Oncology of the University of Campania "L. Vanvitelli", has always been the elucidation of the genetic mutations and the molecular mechanisms underlying congenital paediatric diseases. His approach has always been a combination of molecular, translational and clinical researches.

In particular, most of his scientific work has been done on congenital haematological diseases such as inherited red cell membrane disorders (hereditary spherocytosis, hereditary elliptocytosis, congenital dyserythropoietic anaemias, stomatocytosis and related diseases), thalassaemia and hemoglobinopathies. He published seminal discoveries on new genetic and molecular basis of the hereditary spherocytosis on which he is considered a worldwide expert and has been invited for a review article by the journal Lancet.

Numerous other important clinical contributions were made regarding the incidence, the evaluation and the treatment of some of the most threatening complications of the Thalassemia, i.e. osteoporosis and iron overload.

On these topics, his findings greatly helped the management and treatment of thalassemic patients, improving considerably their quality of life. In 2020 he has been an author of article in New England Journal of Medicine on new drug in Thalassemia.

Moreover, he has been the national coordinator for the Italian guidelines on the management of the Sickle cell disease, Autoimmune Hemolytic Anemia and Bone disease in Thalassemia. Several studies were performed on immune and inherited thrombocytopenias, leading to the discovery of a new pro-oncogenic gene involved in the ethiopathogenesis of a subset of congenital thrombocytopenias. These studies were funded by Telethon.

In the contest of the interest for inherited paediatric diseases; molecular projects were developed and finalized on primary familial vesico-ureteral reflux and other inherited urological anomalies as well as on neurohypophysial diabetes insipidus

Lately, the main focus of his scientific interest has been dedicated to address the molecular basis of congenital erythrocytosis.

A major impact in this field was due to his work, published on Blood in 2006, where he described a cluster of Chuvash polycythemia (a form of inherited erythrocytosis due to alteration in the sensing of the oxygen level) in the island of Ischia. In this island the canonical mutation that causes this disease is more frequent (0.070) than it is in Chuvashia (0.057), region that named this form of erythrocytosis.

After this finding, he become a national and international referent on the topic and was able to start collaborations with the international leaders on adult and paediatric erythrocytosis as stated before.

He described some new mutations on HIF1-alpha and was involved in an international study group on the molecular basis of hypoxia sensing.

Moreover, he has been the person in charge for a public laboratory aimed to find Oxygen Sensing pathways' inhibitors to cure rare diseases.

During the years he collected samples and organized a personal national database on families with familiar erythrocytosis that led to the preliminary results described in the project proposal and is a great resource for future studies.

in 2020 he has published a prestigious article in New England Journal of Medicine on Effects of Germline VHL Deficiency on Growth, Metabolism, and Mitochondria.

Naples, 22/Mar/2022

Silverio PERROTTA

"I authorize the use of my personal data according to D.Lgs. n° 196/2003".