GIANCARLO PARENTI

ACADEMIC POSITION
Full Professor of Pediatrics

PERSONAL INFORMATION

Place and date of birth Naples, Aug 23, 1956

Nationality: Italian

Professional Address: Department of Translational Medicine, Federico II University

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Biosketch

Education
Medical degree in 1980 (School of Medicine, Federico II University, Naples)
Residency in Pediatrics (1981-84) (Department of Pediatrics, Federico II University, Naples)

Research training
1979-1985 and 1987-1989: Department of Pediatrics, Federico II University, Naples.
1985-1986: Dept. of Cell Biology and Genetics, Erasmus University, Rotterdam, the Netherlands.
September-December 1991 and July-November 1994: Institute of Medical Genetics, Baylor College of Medicine, Houston, Texas, USA.

Membership in scientific societies: Member of the European Reference Network for Metabolic diseases (MetabERN), co-coordinator of the lysosomal storage diseases subnetwork. Member of SIMMESN (Italian Society for the Study of Inherited Metabolic Diseases and neonatal screenings), SSIEM (Society for the Study of the of Inborn Errors of Metabolism), European Pompe Disease Consortium (EPoC).

Main scientific and research interests: Inborn errors of metabolism.

Contribution to science
- Identification of genes responsible for metabolic disorders, such as *X-linked ichthyosis* due to the deficiency of steroid sulfatase (Ballabio A, et al. *Proc Natl Acad Sci USA*. 1987); *X-linked recessive chondrodysplasia punctata* (CDPX) (Franco B, et al. *Cell*. 1995); *Multiple sulfatase deficiency* (Cosma MP, et al. *Cell*. 2003); *Lathosterolosis*, due to deficiency of 3beta-hydroxysteroid-delta5-desaturase (Brunetti-Pierri N, *Am J Hum Genet*. 2002).
- *Lysosomal storage diseases*, with a particular focus on the characterization of their pathophysiology (Sardiello M, et al. *Science*. 2009; Spampanato C, et al. *EMBO Mol Med*. 2013; Gatto F, et al. *Sci Rep*. 2017; Tarallo A, et al. *EMBO Mol Med*. 2021; Parenti G, et al *EMBO Mol Med*. 2021; Parenti G, et al *Annu Rev Med*. 2015).
- D*evelopment of innovative therapies for lysosomal storage disorders*, particularly pharmacological "chaperones" (Parenti G, et al. *Mol Ther*. 2007; Parenti G. *EMBO Mol Med*. 2009; Parenti G, et al. *Mol Ther.* 2015). Studies on the synergy between enzyme replacement therapy and chaperones (Porto C, et al. *Mol Ther*. 2009) have been translated into a clinical trial (Parenti G, et al *Mol Ther*. 2014). Studies on other therapeutic approaches, such as gene therapy have been conducted in mucopolysaccharidoses IIIB (Tardieu M, *Lancet Neurol*. 2017) and VI.

Prof. Parenti has published approximately 220 papers (Scopus) in international, peer-reviewed journals.

Link to scopus profile: https://www.scopus.com/authid/detail.uri?authorId=7005315253