

Anna Aspesi

Curriculum vitae

BIO AND EDUCATION

Education

- April 2008: PhD in Molecular Medicine, Università del Piemonte Orientale, Italy.
- April 2002: Master degree in Biological Sciences, cum laude, Università del Piemonte Orientale, Italy

Research Experience

- January 2006-December 2006: Visitor student in the laboratory of Biochemistry and Molecular Biology (Prof. S.R. Ellis), University of Louisville, Louisville, KY, USA
- December 2003-January 2011; February 2012 - October 2015: Research fellow in the laboratory of Genetic Pathology (Prof I. Dianzani), Università del Piemonte Orientale, Novara, Italy
- September 2003: Visitor student in the laboratory of Genomics and Physiology of Lactation (Prof M. Ollivier Bousquet), INRA, Jouy-en-Josas Cédex, France
- April 2003-November 2003: Research fellow in the laboratory of Molecular Pathology, (Prof C. Isidoro), Università del Piemonte Orientale.
- September 2000-April 2002: Undergraduate student in the laboratory of Human Anatomy (Dr F. Renò), Università del Piemonte Orientale.

UNIVERSITY CAREER

November 2015 – present:	Assistant Professor in Pathology, Department of Health Sciences, Università del Piemonte Orientale, Italy
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MAIN FIELDS OF INTEREST

1. Ribosomopathy
2. Diamond-Blackfan anemia
3. Mesothelioma

CURRENT ISSUES OF RESEARCH

1. Pathogenesis of Diamond-Blackfan anemia

This project aims to unravel the molecular bases of Diamond-Blackfan anemia, a rare erythroid aplasia caused by mutations in ribosomal protein genes.

2. Genetic factors involved in the development of pleural mesothelioma

This project aims to identify genetic factors that predispose to the development of asbestos-induced pleural mesothelioma.

CURRENT FUNDED PROJECTS

PROGRAMME	FUNDED PROJECT
Fondazione Europea per la DBA	Rescue of Diamond Blackfan Anemia haploinsufficiency by SINEUP molecules
Department of Health Sciences, Università del Piemonte Orientale	Role of the interaction between osteopontin and B7h in cancer
DBA Foundation USA	The problem of interpreting missense mutations of DBA genes: proposal for a new functional assay
Diamond Blackfan Anaemia UK	Improving the interpretation of genetic diagnosis of Diamond-Blackfan anaemia: a new functional assay to assess the pathogenicity of missense mutations

TOP FIVE PAPERS

1. Aspesi A, Betti M, Sculco M, Actis C, Olgasi C, Wlodarski MW, Vlachos A, Lipton JM, Ramenghi U, Santoro C, Follenzi A, Ellis SR, Dianzani I. A functional assay for the clinical annotation of genetic variants of uncertain significance in Diamond-Blackfan anemia. *Hum Mutat.* 2018 Aug;39(8):1102-1111. doi: 10.1002/humu.23551
2. Aspesi A, Monteleone V, Betti M, Actis C, Morleo G, Sculco M, Guarrera S, Wlodarski MW, Ramenghi U, Santoro C, Ellis SR, Loreni F, Follenzi A, Dianzani I. Lymphoblastoid cell lines from Diamond Blackfan anaemia patients exhibit a full ribosomal stress phenotype that is rescued by gene therapy. *Sci Rep.* 2017 Sep 20;7(1):12010. doi: 10.1038/s41598-017-12307-5.
3. Betti M, Aspesi A, Biasi A, Casalone E, Ferrante D, Ogliara P, Gironi LC, Giorgione R, Farinelli P, Grosso F, Libener R, Rosato S, Turchetti D, Maffè A, Casadio C, Ascoli V, Dianzani C, Colombo E, Piccolini E, Pavesi M, Miccoli S, Mirabelli D, Bracco C, Righi L, Boldorini R, Papotti M, Matullo G, Magnani C, Pasini B, Dianzani I. CDKN2A and BAP1 germline mutations predispose to melanoma and mesothelioma. *Cancer Lett.* 2016 Aug 10;378(2):120-30. doi: 10.1016/j.canlet.2016.05.011.
4. Aspesi A, Pavesi E, Robotti E, Crescitelli R, Boria I, Avondo F, Moniz H, Da Costa L, Mohandas N, Roncaglia P, Ramenghi U, Ronchi A, Gustincich S, Merlin S, Marengo E, Ellis SR, Follenzi A, Santoro C, Dianzani I. Dissecting the transcriptional phenotype of ribosomal protein deficiency: implications for Diamond-Blackfan Anemia. *Gene.* 2014 Jul 25;545(2):282-9. doi: 10.1016/j.gene.2014.04.077.
5. Flygare J, Aspesi A, Bailey JC, Miyake K, Caffrey JM, Karlsson S, Ellis SR. Human RPS19, the gene mutated in Diamond Blackfan anemia, encodes a ribosomal protein required for the maturation of 40S ribosomal subunits. *Blood.* 2007 Feb 1;109(3):980-6.

AWARDS

1. Winner ex-aequo of the best oral presentation award for young researchers, 2nd Meeting Sphingolipid Club, June 3-4, 2003, Sale Marasino (BS), Italy.