

Shwachman-Diamond Syndrome Italian Research Grant 2016

Progetti selezionati

1. **Further insights into the leukocyte immunophenotype of SDS patients and extensive analysis of mTOR activation in leukocyte subsets**
Dr. Antonio Vella - Unità di Immunologia - Azienda Ospedaliera Universitaria Integrata Verona. [PDF](#)
2. **Role of notch pathway in Shwachman-Diamond Syndrome hematopoiesis**
Dr. Simone Cesaro - Direttore Unità Oncoematologia Pediatrica - Azienda Ospedaliera Universitaria Integrata Verona. [PDF](#)
3. **Preclinical evaluation of the effect of Ataluren in restoring the expression of mutated SBDS protein in SDS cells**
Dr. Marco Cipolli - Centro Fibrosi Cistica Ospedali Riuniti Ancona. [PDF](#)
4. **Role of the human niche in inducing and supporting myelodysplastic syndrome and/or leukemia evolution in Shwachman Diamond Syndrome patients**
Dr. Giovanna D'Amico - Centro Ricerca "M. Tettamanti" Clinica Pediatrica - Ospedale San Gerardo Monza. [PDF](#)
-> [Relazione intermedia](#)
5. **Genomic and clinical variations in Shwachman Diamond Syndrome: in silico and molecular analysis of data from exome sequencing in 16 patients**
Dr. Antonella Minelli - Dipartimento di Medicina Molecolare - Unità di Biologia Generale e Genetica - Università degli Studi Pavia. [PDF](#)
6. **Deletion of the long arm of chromosome 20 in bone marrow encompassing the EIF6 gene in Shwachman Diamond Syndrome: benign prognosis mechanism evaluated by cytogenetic, aCGH and expression arrays approaches**
Dr. Roberto Valli - Dipartimento di Medicina Clinica e Sperimentale Università dell'Insubria Varese. [PDF](#)