

Annual Meeting on Rare Diseases

Cluj-Napoca 2019

Editors:

**Bogdan Chiş, Simona Bucerzan, Paula Grigorescu Sido,
Dan L. Dumitraşcu**

The authors have the responsibility for the content of these paper abstracts

Editor in Chief

Radu Ion Badea

Associate Chief Editor

Petru-Adrian Mircea

Senior Editor

Dan L. Dumitraşcu

Honorary Board Adviser

Monica Acalovschi

Associate Editors

Ioana Berindan Neagoe

Radu Nicolaie Oprean

Editorial Assistant

Monica Platon Luşor

Editorial Office

Mîndra Eugenia Badea

Mihaela Băciuţ

Cristian Bârsu

Simona Clichici

Horaţiu Colosi

Doiniţa Crişan

Ofelia Crişan

Diana Dudea

Sorin Marian Dudea

Daniela Fodor

Călin Lazăr

Felicia Loghin

Valentin Muntean

Andrada Seicean

Copy Editor

Ioana Robu

Website Manager

Daniel-Corneliu Leucuţa

Art and Graphic Design

Adrian Grecu

Desktop Publishing

Mihai Ioan Lazăr

Editorial Secretary

Dorina Sorcoi

MPR - Medicine and Pharmacy Reports

Supplement No. 2, Vol. 92, 2019

e-ISSN 2668-1250

CONTENTS

| | |
|--|-----|
| Foreword | S5 |
| Cardiac involvement in Fabry disease <i>Dan Radulescu, Liliana Radulescu, Bogdan Chis</i> | S7 |
| Diagnosis, treatment and outcome in patients with Gaucher disease from Romania. Contributions to knowing the disease <i>Paula Grigorescu-Sido, Cristina Drugan, Anca Zimmerman, R. A. Popp, Camelia Al-Khzouz, Ioana Nascu, Cecilia Lazea, Calin Lazar, Simona Bucerzan</i> | S7 |
| Pulmonary hypertension and valvular involvement in Gaucher disease patients <i>Cecilia Lazea, Simona Bucerzan, Anca Zimmermann, Camelia Al-Khzouz, Mirela Crisan, Ioana Nascu, Radu Popp, Paula Grigorescu-Sido</i> | S8 |
| Clinical, genetic characteristics and outcomes of Romanian patients with Gaucher disease diagnosed under the age of 18 <i>Ioana Nascu, Paula Grigorescu-Sido</i> | S9 |
| Neurological manifestations in Fabry disease <i>Adina Stan, Fior Dafin Muresanu</i> | S9 |
| Fabry disease - still underdiagnosed <i>Bogdan Chis, Dan Dumitrascu</i> | S10 |
| Clinical and paraclinical aspects in Pompe disease <i>Alina-Costina Luca, Andreea-Simona Holoc, Elena Braha</i> | S10 |
| Structural abnormalities of the heart in patients with genetic diseases and facial dysmorphism <i>Mirela Crişan, Eva Kiss, Cecilia Lazea, Ana Curt, Paula Grigorescu-Sido</i> | S11 |
| Integrated care – connecting medical care, social care and educational services <i>Zsuzsa Almási, Paula Neagu</i> | S11 |
| Diagnosis, treatment and outcome in patients with 21-hydroxylase and 11-β-hydroxylase deficiency (monocentric study, Cluj) <i>Simona Bucerzan, Paula Grigorescu-Sido, Camelia Al-Khzouz, Anca Zimmerman</i> | S12 |
| Rare forms of obesity in children <i>Camelia Al-Khzouz, Diana Miclea, Simona Bucerzan</i> | S13 |

”Medicine and Pharmacy Reports”

Editorial Office

Str. Moșilor, nr. 33
RO-400609 Cluj-Napoca, România
Tel/fax: +40-264-596086

E-mail

clujulmedical@umfcluj.ro

Site

www.medpharmareports.com

Indexed

PubMed
PubMed Central
SCOPUS
CAB Abstracts
EBSCO
Index Copernicus
getCited
JournalSeek
Open Access Directory
InfoBase
SCIPPIO

Publisher

Iuliu Hațieganu Medical
University Publishing House

MPR - Medicine and Pharmacy Reports

Supplement No. 2, Vol. 92, 2019

e-ISSN 2668-1250

| | |
|--|-----|
| Idiopathic pulmonary fibrosis – <i>Rara avis</i> in pneumology <i>Ana Florica Chiș, Milena Adina Man, Monica Pop</i> | S13 |
| Congenital hypothyroidism through thyroid aplasia diagnosed at 6 months of age. Why? <i>Carmen Culcitchi, Camelia Al-Khzouz, Carmen Asavoai</i> | S14 |
| Acute intermittent porphyria <i>Bogdan Chis, Daniela Fodor</i> | S15 |
| Eosinophilic esophagitis <i>Elvis Popovici, Teodora Surdea Blaga, Dan Dumitrascu</i> | S15 |
| Achalasia <i>Teodora Surdea-Blaga, Bogdan Chis, Liliana David, Dan L. Dumitrascu</i> | S16 |
| Spinal muscular atrophy, standards of care. The experience of the Clinical Hospital for Children Cluj-Napoca <i>Mihaela Vișan, Monica Mager, Diana Orza, Mihaela Dubau, Remus Babici, Laura Bodea, Nicoleta Daraban, Alexandra Maris, Mihai Militaru, Loredana Oana, Diana Păcurar Vlona, Sorin Man, Călin Lazăr, Cornel Aldea</i> | S16 |
| Establishing an etiological diagnosis in disorders of sex development <i>D. Miclea, C. Al-Khzouz, S. Bucerzan, A. Zimmermann, R. A. Popp, V. Cret, M. Farcas, M. Crisan, D. Ștefan, P. Grigorescu-Sido</i> | S17 |
| Genetic counselling in rare hereditary cancer <i>Andreea Cătană, Patriciu Achimaș Cadariu, Daniela Martin, Andrada Orodan, Radu Anghel Popp, Mariela Militaru</i> | S17 |
| Genetics of malignant melanoma in children <i>Eleonora Dronca, Andreea Cătană, Mihai Militaru, Radu Anghel Popp, Mariela Sanda Militaru</i> | S18 |
| Hypophosphatemic rickets – diagnostic and therapeutic aspects <i>Daniela Iacob, Andrei Corbu, Dan Cosma</i> | S19 |
| Congenital lactic acidosis due to a mitochondrial defect. Case report <i>Ligia Blaga, Romana Vulturar, Gabriela Abrudan, Bogdana Todea, Adriana Ciubotariu, Marta Muresan</i> | S19 |
| Lynch Syndrome <i>Roxana Flavia Ilieș, Felicia Maria Bogdan, Gabriela Morar Bolba, Diana Militaru, Andreea Cătană, Eleonora Dronca</i> | S20 |