

RARE GENETIC DISEASES IN TIMIS REGIONAL CENTRE OF MEDICAL GENETICS, ROMANIA.

Maria Puiu^{1,2}, Adela Chirita Emandi^{1,2}, Iulia Jurca Simina^{1,2}, Alexandra Mihailescu^{1,2}, Nicoleta Andreescu^{1,2}, Dorica Dan³

1. Emergency Hospital for Children "Louis Turcanu" Timisoara Romania
2. Center of Genomic Medicine, University of Medicine and Pharmacy "Victor Babes" Timisoara, Romania
3. National Alliance for Rare Diseases Romania

Introduction:

Despite outstanding advances in policies, technology and bioinformatics, the burden of genetic rare diseases is spread worldwide, raising specific issues due to their rarity.

We aim to evaluate the diagnostic approach strategy in Timis Regional Centre of Medical Genetics (RCMGT-part of ERN ITHACA), in collaboration with the Centre for Genomic Medicine in the University of Medicine and Pharmacy "Victor Babes" Timisoara.



Material and method:

Patients addressing the RCMGT receive

- comprehensive clinical assessment
- different genetic testing, in accordance to their phenotype.

Diagnostics yields were calculated for each type of test performed.



Results: 1432 unique patients

- evaluated January 2015-January 2020,
- 882 males and 550 females,
- 83% from 4 surrounding counties and 17% from other counties.
- age 1-7 years (55,6%),
- median age 10.12 years (range 0–78).
- Most frequent disease suspicions were micro-deletion/duplication syndromes, conditions with intellectual disability, and multiple developmental syndromes.
- Diagnostic yield was:
 - karyotype-26.4%,
 - SNP array (molecular karyotype)-20.5%,
 - Next Generation Sequencing: Cardio panel-38,4% and "Clinical exome"-39.3%.
- RCMGT was successful to reach a diagnosis (sometimes using more than one type of test/per patient)
- longer turnaround time due to limited human and financial resources.
- Genetic counselling offered screening and prevention possibility and early treatment.

Conclusion: Rare diseases have become a health priority worldwide and in each community, aligning policies to improve strategies in the coordination of care, diagnosis and access to treatment in patients.



UNIUNEA EUROPEANĂ



GUVERNUL ROMÂNIEI



Instrumente Structurale
2007 - 2013

PROGRAMUL OPERAȚIONAL SECȚIONAL "CREȘTEREA COMPETITIVITĂȚII ECONOMICE"
-AXA PRIORITARĂ 2 -co-finanțat prin Fondul European de Dezvoltare Regională-
„Investiții pentru viitorul dumneavoastră”

Centrul de Medicină Genomică v2 Universitatea de Medicină și Farmacie "Victor Babeș" Timișoara
ID: 1854; SMIS:487449 Nr.contract: 677/09.04.2015