Genomic medicine for the rare diseases patients – a Romanian experience

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Background: Genomic studies has developed in the last decade and is now routine for clinical diagnosis. Although the price of molecular genetic tests has gradually decreased, the burden of costs is considerable even for highly developed European countries. Romania is a developing country where even though appropriate laboratory equipment now exists, thank to funding from research grants in some genetic centres, the cost for consumables needs to be well planned. We aimed to present an approach for prioritizing the allocation of molecular genetics tests at national level.

Methods: The norms for allocating resources need to account for clinical usefulness of a test(treatment/prevention strategies); and treating people equally. With limited resources, the degree of an individual's need for medical intervention may be the most important criterion. Nonetheless, it is imperative to offer genetic tests non-discriminatively to all people in need, regardless of their economical/social/ethnic status

Results: The *six* Regional Centres for Medical Genetics (*Timis, Bihor, Dolj, Iasi, Bucuresti, Cluj),* together with the *Ministry of Health, Romanian Society of Medical Genetics Romanian and National Alliance for Rare Diseases* (RoNARD) are developing a national strategy for allocating resources, based on epidemiology of genetic conditions in Romania. In accordance with needs, healthcare expertize and existing infrastructure, prioritization criteria are being developed to optimize patient access to genetic tests. Coordination between regional centres of medical genetics is crucial for improved efficiency. Guidelines and improved education for clinical geneticists may be key factors to influence good cost/efficiency for diagnosis approach in rare diseases. *The Regional Centers for Medical Genetics* together with the *NoRo Center for Rare Diseases Zalau* are part of the European Reference Network on Rare Congenital Malformations and Rare Intellectual Disability (ERN ITHACA) that offers support from expert Centres in Europe, for optimized patient care.

Conclusion: Further work is needed to determine the criteria necessary in decision making, in order to respect equality but also clinical need within a limited budget.

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