



Research of Rare and Orphan Diseases: RE(ACT), International Congress on Research of Rare and Orphan Diseases, Basel, February/March 2012

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Special Topic Issue: Molecular Syndromology 2012, Vol. 3, No. 5 The medical community does not see rare diseases as a field of priority. In fact, poor characterization of the natural causes, pathologies and low numbers of cases make diagnosis difficult, often resulting in a real ordeal for patients and their families. This special issue of Molecular Syndromology presents six selected contributions of the First International Congress on Research of Rare and Orphan Diseases which brought together courageous researchers working on rare and orphan diseases. It shows the growing research interest in rare and orphan diseases, the new experimental approaches, such as next generation sequencing and therapeutic possibilities. Topics outlined in this issue are:

- Current debates on the emerging technical, medical and ethical issues
- Potential optimum use of the available technology
- Molecular genetics of Charcot-Marie-Tooth diseases
- Amelogenesis imperfecta – a clinically and genetically heterogeneous group of inherited defects of enamel formation
- Smith-Lemli-Opitz syndrome – an autosomal recessive disorder characterized by multiple congenital abnormalities and mental retardation
- Strategies for therapeutic suppression of nonsense mutations in inherited metabolic diseases

Furthermore, the involvement of patient organizations in the development of orphan drugs for rare diseases is emphasized. This volume is essential reading for all researchers in the fields of human genetics and syndromology and everyone interested in the study of rare diseases.

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