Craniofacial anomalies: from genetic mutations through development to disease and treatment

David Rice
Department of Orthodontics, University of Helsinki.
Consultant Orthodontist Helsinki University Central Hospital, Finland

In this lecture I will explain some of the dramatic recent advances that have been made in our understanding the aetiology of craniofacial anomalies. I will describe normal development of the craniofacial region including the lips, palate and bones emphasising the critical stages and the key regulatory mechanisms that can go wrong and result in a craniofacial deformity. Progress in unravelling the molecular aetiology of craniofacial anomalies is principally due to advances in human genetics and developmental biology in combination with clinical knowledge. However, although this has an immediate impact on patient comprehension and insight of their condition, to date it has done relatively little to affect treatment protocols.

Using craniosynostosis, cleft lip and palate, cherubism and Treacher Collins syndrome as examples, I will detail work from my own lab and others of how novel strategies are being developed which are based on our understanding of the molecular aetiology underlying these conditions and which are starting to bridge the gap between the lab bench and the patient. I will emphasise how important transgenic animals are as models of disease and show how they are being used to not only unravel the mechanisms of disease but also to test new treatments. Conversely, I will also describe the seemingly insurmountable limitations of correcting the root causes of developmental abnormalities that occur before birth.
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Current position
Professor of Orthodontics, Head of Department, University of Helsinki, Finland 2007～
Hon Visiting Professor, King’s College & Guy’s Hospital, University of London, UK 2008～2012

Previous professional appointments
BDS Royal London Hospital Medical College, London University 1987
FDS Royal College of Surgeons of England 1990
MSc Guy’s Hospital Medical School, University of London 1993
MOrth Royal College of Surgeons of England 1994
PhD Developmental Biology Program, University of Helsinki, Finland 1999
FDSOrth Royal College of Surgeons, Intercollegiate specialty examination 2000
PGCAP Postgraduate Certificate in Academic Practice, KILT, University of London 2005

Scientific awards and honours
WJB Houston Research Award, European Orthodontic Society 1994
Travelling Research Fellowship (One year), Wellcome Trust 1994
WJB Houston Research Scholarship, European Orthodontic Society 1995
Sulo Toivanen Best PhD Thesis Award, Finnish Cell and Developmental Biology Society 1999
Best Poster Award, European Symposium on Calcified Tissues 2000
Senior Clinician Scientist Award, Medical Research Council, UK 2001
Best Poster Award, Medical Research Society Fellows Meeting 2002
Honorary Fellowship in Dental Surgery, Royal College of Surgeons of Edinburgh 2007