Prevention of cleft lip and palate and craniofacial anomalies

At this couple of minutes, another child with a cleft is born somewhere in the world. At the end of the day, the number will sum up to 700 (and about 230,000 per year). Many other children will have today their clefts operated. Their mothers will be relieved for a while and then afraid again when they will conceive another baby. Some of them will have a cleft. And this will go on and on and on… until we will change our gears and focus our interest, time, and funding to cleft prevention.

More, a number of individuals affected with a cleft will be increasing. We may not realize that this is happening also with our help, because we improved tremendously treatment outcomes and thus our patients live full live, having children - some of them will have a cleft.

Cleft lip and palate anomalies are the most common and the most severe congenital anomalies of the face and mouth and the second most common birth defects in general: one in every 560 newborns is affected with a cleft. If we focus only on individuals with nonsyndromic clefts, we can estimate that one individual with a cleft is among every 1,000 people. Thus, there are nearly 6.85 million individuals with a cleft living on this planet at present. Twenty years from now, with the projected population growth, there will be 1.4 million more of them. And in the year 2050, the cleft population will reach 9.28 million - 2.4 million more than today.

These facts provide us with very strong reasons for not only improving treatment of these serious anomalies, but also for developing efficient strategies for their prevention.

There is already enough scientific evidence suggesting that a significant proportion of cleft lip and palate anomalies are preventable.

In 1982, we first published in Lancet our study suggesting that nonsyndromic cleft lip and palate (NCLP) can be prevented by periconceptional supplementation with folic acid (FA). Our studies later showed 65% decrease in recurrences by mother's daily supplementation of multivitamins with 10mg of FA (Teratology 1995) and 27-50% decrease in occurrence when diet contained 400mcg of FA (Lancet 1995). Several studies that followed confirmed our results, but there were also other that did not support them. In our studies we found significant differences in genotype distributions for several "candidate/susceptibility" genes (MTHFR, RFC1, TGFβ3, BMP4, and others) among populations studied. Analysis of maternal diet revealed that four nutrients were associated with NCLP most often: low intake of folate, zinc, and B6 vitamin, and high intake of vitamin A.

We are concluding that our studies combined with studies of others show enough scientific evidence that a significant proportion of NCLP can be prevented. However, different genes are creating susceptibility for NCLP, and different environmental factors triggering them, exist in specific populations: “ONE SIZE DOES NOT FIT ALL”. Therefore, prevention approach has to address differences in genetic and environmental factors that exist among different populations.

Biography

Tolarova graduated from Charles University School of Medicine in Prague and received MD degree at the age of 23 years and is Board Certified in Pediatrics and Medical Genetics. She completed her PhD at the age of 28 years and her DSc at the age of 44 years – both at the Czechoslovak Academy of Sciences and Charles University School of Medicine in Prague. Dr. Tolarova has devoted over 45 years of her professional carrier to treatment, research, and prevention of cleft lip and palate and craniofacial anomalies. She has over 450 scientific publications and presentations, has been the keynote and featured speaker at numerous scientific meetings, and has held over 20 visiting professorships worldwide and recently helped to establish Indo-Pacific Cleft Prevention Program “ANAMAYA” in Chennai, India. She is Professor of Orthodontics and Executive Director of the Pacific Craniofacial Team and Cleft Prevention Program, University of the Pacific, Dugoni School of Dentistry in San Francisco, Director of Genetic Research and Prevention of Rotaplast Intl. Inc., and Trustee of International Cleft Lip and Palate Foundation.