0,001). The aberrations chromosomal of a type registered authentically above for children in locale STNR (0,47), as contrasted to by group of children of Aral sea region (0,30) ($p < 0,05$). Children of locale Aral sea region more often had aberrations chromatide of a type (1,60). The stable aberrations, such as translocations and deletions registered for children of Aral sea region (0,08) and for children of locale STNR (0,07). Thus, for children ecological unfavorable regions with effect of the radiation and chemical toxic factors the heightened frequency of aberration chromosomes is detected, which one are one of the first indicators of unfavorable effect of the factors of an environment on an organism.

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OROFACIAL CLEFTS AND ASSOCIATED CONGENITAL ANOMALIES: THE 18 YEARS EXPERIENCE OF A MULTIDISCIPLINARY GROUP IN A TERTIARY HOSPITAL IN PORTUGAL

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Background: Orofacial clefts are among the most common birth defects, arising in about 1.7 per 1000 newborns. This heterogeneous group of disorders can occur as an isolated condition or associated with other congenital anomalies or syndromes.

Aim: To evaluate associated malformations in a population of orofacial cleft patients.

Methods: Retrospective study of the medical records of patients that attended the Cleft Lip and Palate Multidisciplinary Group at Hospital S. João, Porto-Portugal, from January 1992 through December 2009. Patients were divided in four groups: cleft lip (CL), cleft lip and palate (CL/P), isolated cleft palate (CP) and atypical cleft (AC). Further categorization included sex, affected relatives, associated congenital anomalies and syndromes.

Results: Of the 340 patients included, 57.6% were males; 38.5% had CP, 37.4% had CL/P, 22.9% had CL and 1.2% had AC. Nineteen percent had known affected relatives. Associated congenital anomalies were found in 52.6% of patients, 43% of which had

recognizable syndromes. Pierre-Robin ($n=34$) and 22q11.2 microdeletion syndrome ($n=12$) were the most common. Orofacial region was the most common site of associated anomalies, followed by the cardiovascular and central nervous systems. Different groups of anomalies arose equally in both genders, except for orofacial region that affected more females ($p < 0.05$) and genitourinary that affected more males ($p < 0.05$). The majority of anomalies with or without recognizable syndromes occurred in CP patients (65.6%; CI 95% [57.3; 73.9%]).

Conclusion: Due to the increased risk of associated malformations in children with clefts further diagnostic investigation might be important, especially regarding those affecting cardiovascular and nervous systems.

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CYSTINURIA OVER TWO GENERATIONS

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Cystinuria is an autosomal recessive disorder that is characterized by an impaired tubular transport of cystine and dibasic amino acids, resulting in cystine urolithiasis. The transport of these amino acids is mediated by the rBAT/b⁰AT transporter, the subunits of which are encoded by the genes *SLC3A1*, located on chromosome 2p16.3-21, and *SLC7A9*, located on chromosome 19q12-13.1. In this work we present a family in which cystinuria was manifested in two generation.

The index case is a female, aged 9 years when she was diagnosed with a kidney stone and underwent surgery. Nitroprusside test was positive and urinary excretion of cystine was high (127.5 mM/M creatinine, normal 0.6-20.0 mM/M creatinine). The family screening revealed that two siblings (sister and brother) had positive nitroprusside test and increased urinary excretion of cystine. The mother had history of surgery for nephrolithiasis, had positive nitroprusside test and increased urinary excretion of cystine. The mutational analysis of the *SLC3A1* gene revealed that the index case and the mother were compound heterozygote (T216M/M467T), while the female and the male siblings were homozygote for M467T mutation. The father was heterozygote for M467T mutation. The ethnic origin of the patients is Egyptian. To the best of our knowledge this is the first report of cystinuria occurring over two generations.