Results: Nowadays the cystatinsuper family is known to comprise about 30 members.The members of cystatin family II –cystatin C, D, S, SA, SN, E/M and F, are found in body fluids.Their involvement in inflammatory processes, cancerogenesis and metastasis, bone remodeling and in other processes has been undoubtedly demonstrated.Cystatins S, SN and SA were found in submandibular and sublingual glands, while cystatin D was detected only in parotid glands.Salivary cystatinsS, SN, SAand D contribute to the maintenance of the oral health through the inhibition of endo- and exogen cysteine proteases, antimicrobial and antiviral protection and regulation of hard tooth tissue remodeling.

Conclusions: Studies of major significance attest the clinical utility of cystatins' assay (cystatin C) for the diagnosis of some diseases (for ex. renal failure). In addition, cystatin C, among other cystatins, decreases the formation of osteoclasts by interfering at a late stage of pre-osteoclast differentiation.Cystatin D is produced by the parotid gland and is secreted through blood serum to the whole body, similar to a hormone, and thus there are setnew research directions of cystatins as markers of diseases, including the ones causing oromaxillofacial pain,prosopalgia, and the monitoring of disease's evolution and of treatment efficiency.

Key words: Cysteine proteases, cystatins, salivary cystatins,disease marker

14. GENETIC HETEROGENEITY OF DEAFNESS AND ITS PRACTICAL IMPLICATIONS

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Introduction: The fundamental process involved in audition is controlled by hundreds of genes, 69 of which are known: 24 AD, 40 AR, 2XL, 3 mitochondrial. Mutant alleles of these genes may determine hereditary deafness. Loss of hearing is etiologically heterogeneous, 2/3 of childhood onset deafness being of a genetic cause. The prevalence of bilateral sensoneural deafness (≥40 dB) is 1:500 healthy new-borns. More than 50% of prelingual deafness is of genetic origin, 70% of which is nonsyndromic, from which 85% is autosomal-recessive. About 400 syndromes include deafness as a component of its phenotype. In Republic of Moldova, deafness hold 3rd place in the structure of disability, number of hearing impaired children being more than 2000.

Purpose and objectives: Analysis of molecular-genetic aspects of deafness and subsequent implications. Objectives were defined as: (1) systematic review of scientific literature regarding epidemiology, genetic heterogeneity, diagnosis and consequences of hereditary deafness; (2) analysis of deafness incidence in Republic of Moldova; (3) elucidating critically the practical implications regarding genotype variations of hereditary deafness.

Materials and methods: The study group was prospectively selected during 2013/14, from deaf patients at Republican Center of Audiology. Patients filled up a questionnaire at discharge. Additionally to cases (patients with hereditary deafness), were random selected an equal number of controls (patients with non-hereditary deafness).

Results: The study group consisted of 10 cases and 10 controls, with a mean age of 5±2,22 years, 65% females. Mean age of diagnosis was 2,3±1,49 years, being fit into maximal plasticity period of central auditory pathways. The count of sensoneural deafness in hereditary group represented 90%, with a single case of transmission hearing loss, bilateral in 90% of cases and 10% of controls, with a postlingual onset in 70%. Pedigree analysis of cases showed 90% AR transmission pattern, and 10% AD.

Conclusion: (1) The cause of deafness may be clinically suspected due to anamnesis and syndrome association, but substrate confirmation should be done with molecular-genetic tests. Although family history can help suspect the genetic cause of deafness, absence of hearing-loss anamnesis at family members does not mean that hearing impairment is not of genetic origin. (2) Recognition of genetic heterogeneity is important in clinical diagnosis, prognosis and genetic counseling regarding recurrence risk.

Keywords: hereditary deafness; connexin-26; sensoneural hearing-loss