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Poster Session P18 – Syndromes and Genetics/Oral Medicine and Pathology

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Poster Session P18/Syndromes and Genetics/ Oral Medicine and Pathology

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A novel *DSPP* mutation (p.V18D) causing dentinogenesis imperfecta type II

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Introduction: Dentinogenesis imperfecta (DGI) is a hereditary disease with abnormal dentin formation. We had a chance to examine a Japanese family with autosomal dominant DGI type II. We performed mutation analysis of the *DSPP* responsible for DGI, and found a novel mutation. Furthermore, we demonstrated the histological findings of DGI-II affected teeth, which were characterized in the molecular pathogenesis background.
Materials and methods: Genomic DNA isolated from PBMCs from family members, as well as that from control individuals, were analysed. Coding exons of the *DSPP*, together with their flanking introns, were PCR-amplified and then directly sequenced. Non-carious primary incisors from proband and an age-matched normal control were sectioned with and without H–E staining. These samples were examined by conventional light microscopy.
Result: The proband's primary tooth crowns were uniformly amber brown, and enamel on the occlusal surfaces was often dislodged, exposing the soft dysplastic dentin, which led to rapid attrition. The analysis revealed a mutation (c.53T > A, p.V18D, g1192T > A) involving the first nucleotide within the *DSPP* gene of exon 3. Histological examination showed the dentin tubules in DGI specimens from the proband were wide and the number of tubules reduced compared to that of the normal control counterpart. Unregulated or poorly orientated dentin tubules were also observed.
Conclusion: We have identified the novel mutation (p.V18D) of the *DSPP* in a Japanese family with DGI-II. DSPP V18 residue is highly conserved among other mammalian species. Though the unique histological feature was not demonstrated in the present case with the novel mutation (p.V18D) of the *DSPP*, we believe these results would greatly help us to understand the pathogenesis of DGI.

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KBG syndrome – clinical features and specific dental findings

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Introduction: KBG syndrome was first reported by Herrmann *et al.* in 1975. This investigation sought to distinguish the most frequent manifestations of KBG syndrome and present specific dental findings in a 9-year-old Chinese boy.
Materials and methods: PUBMED search using the keyword KBG syndrome yielded 20 articles. Due to disparities in the quality of evaluations in the reports any physical feature that was not discussed was assumed to be absent. Five cases had to be excluded from the final analysis due to inadequate information.
Results: Of the 54 cases including the present case, 36 (66.6%) were males, 25 (46.2%) had at least one affected first-degree-relative and parent-to-child transmission was documented in 12 instances. Mental retardation or global developmental delay was reported in 42 (77.7%) cases and 46 (85.1%) exhibited shortness of stature. Oral features were evident in 53 (98.1%) cases; macrodontia was present in 49 (96%) documented cases; the present case was the only report to exhibit hyperdontia and a talon cusp. The other clinical features observed were craniofacial anomalies (90.7%) followed by abnormalities of the nose (88.8%), hands (88.8%), mouth (74%), eyes (70.3%), eyebrows (68.5%), philtrum (61.1%), ears (59.2%), low hairline and lower extremity.
Conclusion: The cardinal features of KBG syndrome include facial dysmorphism, short stature, skeletal anomalies and mild developmental delay. Of the oral findings macrodontia of the maxillary central incisors occurred in majority of the cases. This is the first report of KBG syndrome in a Chinese subject and also the only report to exhibit hyperdontia and a talon cusp.

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The clinical study on a Chinese family with amelogenesis imperfecta

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Introduction: Amelogenesis imperfecta (AI) represents a group of inherited disorders which are clinically heterogeneous and exhibit tooth enamel defects in the absence of systemic manifestations. The purposes of our study were to realize the clinical and genetic features of a Chinese family with Amelogenesis imperfecta (AI).
Patients and methods: The AI family was checked by the genealogy surveying way, and conditions have been analysed from a genetics perspective and a genealogy was constructed. The permission of our study was obtained from Peking University Health Science Centre of Ethical Committee.
Results: There were 4 generations in the pedigrees, AI patients were present in each generation, and the men and women were affected equally.
Conclusion: The findings proved that the characteristic of the pedigrees was dominant inheritance. According to Witkop's classification (1971), the patients' clinical phenotypes in the family were diagnosed as hypomaturation AI.

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A survey of oral biopsies from paediatric patients at the University hospital

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Introduction: It is important for paediatric dentists to identify various oral lesions in children.
Materials and methods: We examined 331 biopsy records of children aged 0–19 years collected over a 29-year period (1979–2008). All specimens were taken at the Paediatric Dentistry Clinic and sent to the Department of Oral Pathology at Niigata University Medical and Dental Hospital. The lesions were classified into 4 categories: inflammatory/reactive lesions, tumour/tumour-like lesions, cystic lesions and other anomalies. Malignant tumours were referred to the Oral Surgery Department and therefore, not included.
Results: There were 150 specimens from males and 181 from females (male: female ratio = 1: 1.2). A high prevalence was found in each of the 8-, 9- and 10-year-old groups. Inflammatory/reactive lesions formed the largest group of biopsy specimens (58.0%), followed by tumour/tumour-like lesions (19.9%), other anomalies (12.4%) and cystic lesions (9.7%). Of the 192 inflammatory/reactive lesions, pericoronal myxofibrous hyperplasia (Yonemochi *et al*., J Oral Pathol. Med 1998; 27: 441–452) was the most prevalent lesion (45 cases) followed by mucous extravasation cyst (44 cases) and fibrous hyperplasia (40 cases). Of the 61 tumour/tumour-like lesions, odontoma formed the largest group (45 cases) followed by ameloblastic fibroma (10 cases). Of the 32 cystic lesions, dentigerous cyst was the most prevalent (11 cases) followed by radicular cyst (6 cases). Dental anomalies like supernumerary teeth, hypoplastic teeth and natal teeth were included as other anomalies.
Conclusion: Paediatric oral lesions differ from those of adults; therefore pathological diagnoses of the oral specimens are necessary in paediatric dentistry.

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Investigation of the correlation between intestinal parasitic infections and bruxism among preschool children

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Introduction: Given the fact that bruxism is a prevalent oral habit among children and a potential destructor of oral tissues, the present study aims to investigate the relationship between intestinal parasitic infections and bruxism among kindergarten children.
Materials and methods: Questionnaires were administered among parents of kindergarten children in Isfahan to select 50 children identified by their parents to be in the habit of bruxism and 50 without the habit as control. Informed consent was obtained prior to the investigation. Parents were delivered sampling instruments with proper instructions to collect samples from both groups for parasitological tests. The diagnostic parasitological tests involved the direct stool smear, formol-ether concentration, and Scotch tape tests. Comparisons for the prevalence of intestinal parasitic infections among case group were performed using the Chi-square test.
Results: Parasitic infections were observed among 19% (22% cases & 16% controls) of the total study population. A statistically significant relationship was observed between infection with pathogenic parasites and bruxism ($P < 0.05$). Also, the prevalence of Enterobius vermicularis (16% in the study group and 4% in the control group) indicated a significant relationship between infection with this parasite and the development of bruxism habits ($P = 0.046$). Finally, a significant relationship was revealed between parents with a history of bruxism and frequency of bruxism among their children ($P < 0.05$).
Conclusion: Our findings suggest that pathogenic parasites may serve as the cause for the initiation of bruxism habits among children.

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Dentilisin involvement in coaggregation between *Treponema denticola* and *Tannerella forsythia*

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Introduction: Trep*onema denticola, Tannerella forsythia* and *Porphyromonas gingivalis* are frequently coisolated from periodontal lesions, and this combination of microorganisms has become known as the 'red complex'. Coaggregation plays a key role in the formation of biofilm by these microorganisms. Although several adhesion factors have been identified for *T. denticola*, the ligand by which it achieves coaggregation remains to be determined. The purpose of this study was to clarify the component involved in coaggregation between *T. denticola* and *T. forsythia*.
Materials and methods: Coaggregation between *T. denticola* and *T. forsythia* was evaluated by measuring OD_{660} using a spectrophotometer or visual turbidimetric method. *T. forsythia* ATCC43037 and *T. denticola* ATCC35405, ATCC33520, ATCC33521, K1 (dentilisin-deficient mutant from ATCC35405), KpSano7 (dentilisin-deficient mutant from ATCC33520) and DMSP3 (major outer sheath protein-deficient mutant from ATCC35405) were used in this study.
Results: *T. denticola* coaggregated with T. forsythia within 30–60 min. Coaggregation was inhibited by heat treatment of *T. denticola*, whereas that of T. forsythia was not. Disaccharides such as sucrose, maltose and lactose reduced coaggregation by approximately 50%. Magnitude of coaggregation differed among T. denticola strains in accordance with their dentilisin activities. T. *denticola* K1 and KpSano7 did not coaggregate with *T. forsythia*, whereas *T. denticola* DMSP3 did. Serine protease inhibitor PMSF treatment of *T. denticola* ATCC35405 abolished its activity, but did not affect its coaggregation reaction with *T. forsythia*.
Conclusion: The results indicate that dentilisin is involved in coagregation between *T. denticola* and *T. forsythia*.

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Gingival expression of SOD and NOS mRNA in NOS1 knockout mice

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Introduction: Active oxygen and free radicals play an important metabolic role in cells and tissues. However, little is known about

the morphological dynamics of their related enzymes in gingiva. We investigated gingival mRNA expression of active oxygen-scavenging and free radical-synthesizing enzymes in normal and neuronal nitric oxide synthase (NOS1) knockout (KO) mice to elucidate the mutual dynamics of these enzymes in NOS1 deficiency.

Materials and methods: Five-week-old normal and NOS1 KO mice were used. mRNA expression in their gingiva was investigated by in situ hybridization. Animals were maintained and used in accordance with recommendations in the Guide for Care and use of Laboratory Animals, prepared by the institute of laboratory Animal Resource, and guidelines for treatment of experimental animals in Tokyo Dental College.

Results: In normal mice, NOS1, manganese superoxide dismutase (Mn-SOD) and endothelial nitric oxide synthase (NOS3) mRNAs were positive in the lamina propria mucosae, although the signal of NOS3 mRNA was weak. Mn-SOD and NOS3 mRNAs were also positive or weakly positive immediately below the horny layer. Additionally, a positive signal for NOS1 mRNA was observed in the epithelial basal layer. However, inducible nitric oxide synthase (NOS2) mRNA was negative in all tissues. In NOS1 KO mice, no expression of NOS1 mRNA was observed, as expected. However, NOS2 and NOS3 mRNAs were strongly expressed in the basal cell layer in KO mice. Furthermore, NOS3 mRNA was also expressed in the lamina propria layer.

Conclusion: Taken together, the results suggest that NOS2 and NOS3 compensate for NOS1 in the gingiva under NOS1-deficient conditions.

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Can mesiodentes be resorbed?

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Introduction: Mesiodens is the most common supernumerary tooth with a prevalence of 1–3%. Eight to 23% of the mesiodentes erupt spontaneously, while the rest are impacted, about half of them are in an inverted position. Absolute indications for removal are eruption disturbances of the permanent incisor or hindrance for orthodontic treatment. This means that a large number of mesiodentes are not removed and may cause problems later on. However, there have been case reports indicating that the impacted mesiodens will be resorbed over time. If this is generally true the treatment planning for mesiodentes may be possible to change. The aim with this study is to investigate the radiographical changes and the frequency of resorptions in mesiodentes left in situ.

Materials and methods: About 100 individuals where mesiodentes have been left in situ will be radiographically followed. The material is collected from three areas in Sweden; Stockholm, Gothenburg and Jönköping. The study has been approved by an ethical committee.

Preliminary results and conclusion: In the so far collected material there are a number of cases showing different levels of resorption over time. The study and the analyses of the material will be finished during the spring and detailed results will be ready to be presented in München June 2009.

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Clinic and subgingival bacteria research on aggressive periodontitis and chronic periodontitis

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Introduction: To investigate prevalence of aggressive periodontitis (AP) and chronic periodontitis (CP) in middle school students at Huangpu District in Shanghai, and to study on their clinical characteristics and subgingival bacteria.

Materials and methods: Of 9548 students aged 11–19 years, 73 subjects were diagnosed having juvenile periodontitis in 2002. Recently, 64 patients were further consultation. Base on the 1999 new classification of periodontal disease, they were classified anew by clinical examination and radiographic examination. Samples of 18 patients were taken to analyze subginginval bacteria including *Actinobacillus actinomycetemcomitans* (Aa), G- obligatory anaerobic black bacteria and Fusobacteria.

Results: Ten subjects (15.6%) of 64 subjects were diagnosed as AP; 54 subjects (84.4%) of 64 subjects were presented with light and moderate CP. In longitudinal comparison, community periodontal index of treatment of AP patients had significant difference ($P < 0.01$). In the CP patients, there was a significantly positive correlation between CPITN, SBI and Cl-S, respectively ($P < 0.01$). Of 128 separate species 185 (66.4%) were facultative anaerobes. 143 (33.6%) were obligatory anaerobes in AP patients, of 133 separate species, 65 (48.9%) were facultative anaerobes, 68 (51.1%) were obligatory anaerobes in CP patients ($P < 0.01$).There was a significant difference in the number of patients positive for Aa ($P < 0.05$).

Conclusion: There is difference in aetiology and pathology between AP and CP. Due to use of different classifications and diagnostic criteria of the disease, epidemiological investigation before may overestimate the prevalence of juvenile periodontitis (AP, new classification). AP may be closely related to Aa.

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Some salivary parameters of children with and without black stain

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Introduction: Black stain is an aesthetic problem and has significant effect on the personality and self confidence. The prevalence of the black stain in a group of Turkish children is 18.5% that is higher than other populations. There are very few biochemical studies about black staining in the literature (Reid & Beeley 1977, 1976 and Surdacka 1989). So the purpose of the present study was to compare some salivary parameters of children with and without black stain.

Materials and methods: Totally 52 randomly selected children (boys 27, girls 25) ages between 5–13 years were divided into two groups: Study group ($n = 27$) had black stain, control group ($n = 25$) had no staining. Whole saliva samples were collected from children without any stimulation and buffering capacity, flow rate, pH, total calcium and total protein levels were determined and statistical analysis was performed using the Statistical Program of Social Science (SPSS 16) for Windows. Statistical significance was considered at the 5% level.

Results: No significant differences were found between groups in any parameters ($P > 0.05$).

Conclusion: Since the prevalence of the black stain in a group of Turkish children was high, other factors that may be related to black stain need to be investigated.

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IMUDON for treating atopic cheilitis in children

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Introduction: This was to study the course of atopic cheilitis and optimize cheilitis treatment using IMUDON (INN – mixture of bacteria lysates) having immunomodulatory properties and corrective indices of local immunity.
Materials and methods: Forty four children aged 8–15 years were selected for the study. All of them had obvious manifestations of atopic neurodermitis combined with atopic cheilitis: skin edema round the mouth, infiltration and peeling of the red border of the lips, radial banding, papular spots in the mouth corners. The children felt pain while eating and constant itch of lips. They were divided into two groups: 20 were treated traditionally (Bepanthen and methiluracil ointments), 24 received a prescription to take IMUDON (6–8 pills per day during 3–4 weeks).
Results: The condition of the children treated traditionally improved after three to four days but after ointment withdrawal manifestations appeared again. Those treated with IMUDON had significant improvements only in six to fourteen days. At the end of the treatment manifestations of atopic cheilitis in three children completely disappeared, significant improvements were found in nineteen persons and two children had no effect. There was an increase of concentration of lysozyme and segmental immunoglobulin in saliva. And it is just the point facilitating healing of fissures in the mouth corners and removing oral mucosa inflammation. All the children had a good drug response and liked the IMUDON flavour which is important while treating children.
Conclusion: IMUDON increases a local immunity index that leads to disease regress. 92% of the children had favorable clinical results.

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Clinic and laboratory aspects of herpetic stomatitis severity course in children

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Introduction: Aim of this study was to investigate the local immunity in the oral cavity in children with acute herpetic stomatitis (AHS).
Materials and methods: 100 children diagnosed with different stages of AHS were selected for this study. In mild cases the number of the lesions in the oral cavity was ≤ 5, in the moderate cases up to 10–15. Samples of children's from saliva were collected at the very beginning of the disease. The activity of lysozyme and secretory immunoglobulin A concentration were investigated by immunoferment analysis. Microbiocenosis structure of *Staphylococcus aureus, Streptococcus pyogenes* groupe A, *Staphylococcus epiderm*, *Candida albicans* were also studied.
Results: SgA and lysozyme concentration correlate with the severity and nature of AHS. The microbiocenosis structure in the oral cavity has cut down twice relative to concentration of bacteria typical of this biotope. A three-fold increase of the number of pathogens (*Staphylococcus aureus* and *Streptococcus pyogenes* group A) was observed. Microflora dissemination atypical of children's oral cavity has risen in 1.5 times.
Conclusion: Local immunocorrectors make diverse correction of microbiological and immune shifts that normalizes microbiocenisis and shortens a disease period.

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Vermilion border and tongue at children with diseases of the alimentary tract

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Introduction: The aim of the present work was the study of peculiarities and general regularities of clinical displays of glossitis and cheilitis in patients with pathology of digestive system.
Patients and methods: Fifty three children, in the age 10–17 years, with the diagnosis are clinically surveyed: ulcerative colitis, Crohn disease-15 (Group A), chronic mucous colitis-20 (Group B), chronic gastritis, gastroduodenitis-18 (Group C). The group of comparison was made by 35 children without pathology (Group D).
Results: Changes were discovered on the vermilion border at 43 examined patients. Edema and dryness of the lips, angular and commissural cheilitis were revealed. Small rifts were discovered on vermilion border. All children had changes in papillae of the tongue, showing hypertrophy filiform papillae, mainly in medial and distal parts of back of the tongue. The hypertrophy fungiform papillae occur on periphery of tongue and on its tip. Thick white, yellowish, grey plaque was situated on a back of the tongue - at 53 patients. Ribbed tongue, furred tongue and geographical tongue were revealed. The tongue plaque index (Yaegaki K., 1998) was 2.8 in Group A; 2.2 in Group B; 2.4 in Group C; 1.3 in Group D respectively. Index WTC (Winkel Tongue Coating, E. Winkel, 1998) was 9.9 in Group A; 7.6 in Group B; 9.3 in Group C; 3.8 in Group D respectively.
Conclusion: Diseases of various departments of digestive system at all children reveal on a mucous membrane dorsal surface of tongue and in vermilion border of the lips.

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Buco–dental health in children with HIV

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Introduction: 270 HIV(+) children are living in Chile. Caries, plaque, gingivitis and orofacial injuries are described as major oral problems. The objective of this study was to investigate the dental and mucosal characteristics in a group of HIV(+) children and to compare them with healthy controls.
Materials and methods: Twenty three HIV(+) children aged between 2 and 14 years were examined in the Hospital de Niños Roberto del Río. The controls consisted of 60 healthy children without HIV infection. The children were classified and examined according to the WHO standard. Orofacial lesions were examined and diagnosed according to 'Classification and Diagnosis in HIV Infected Children', (Ramos-Gomez and coworkers). Statistical analysis was performed with Software Systat (version 11). Mann–Whitney–U-Test, the Kruskal–Wallis-Test and Chi-Square-Test were used with significance level set to $P < 0.05$.
Results: Regarding caries experience no significant differences were found between both groups (HIV: DMFT = 3.5, dmft = 5.9; Controls: DMFT = 2.54, dmft = 4.3). Index of

hard deposits amounted to 0.1 for HIV children and 0.053 for the control group. DDE in HIV children with permanent teeth amounted to 52, for primary teeth to 49. Orofacial lesions were only observed in children with HIV.
Conclusion: In this study, the children with HIV had a similar caries experience compared to healthy children. However the DDE and OHI-S were significantly larger.

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Dental health of children with cleft lip and palate

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Introduction: Cleft lip and palate compound about 30% from all human congenital defects. Birth rate of children with cleft lip and palate varies from 1:500 to 1:1000 of neonates. During last 100 years the frequency of this defect has increased in 3 times. This was to study dental health of children with cleft and lip palate.
Materials and methods: 98 children with cleft and lip palate at the age from 1 to 17 were examined according to the criteria of WHO.
Results: Caries prevalence was 70% in primary occlusion (33 children), 92% in mixed occlusion (34 children) and 98% in permanent occlusion (31 children). Caries intensity was determined as follows: dmft = 4.3; dmft/DMFT = 8.9; DMFT = 9.5 pro tanto. Enamel hypoplasia (local and general) was detected in 8% of children. Malocclusion had 100% of children. Gingivitis with different severity had 85% of patients. Oral hygiene level was good in 27% of cases, satisfactory level was detected in 41% of children and poor oral hygiene had 32% of patients. 70% of children were never trained how to treat their teeth.
Conclusions: (i) Dental health care for children with cleft lip and palate must be priority and it should be improved because of high level of dental diseases prevalence and intensity in these children. (ii) It is necessary to activate the realization of preventive and health educational programs in dispensary centres for children with cleft and lip palate.

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Taste perception evaluation in a hundred healthy children sample

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Introduction: The aim of this study was to carry out a standardized assessment of taste in 100 healthy children sample.
Materials and methods: 100 healthy children (aged between 4 and 12) referred to the Paediatric Department of Dental Clinic in Brescia were enrolled. Children were divided into two groups (A and B) including 50 patients each. The study was carried out using a taste perception test, composed in two phases: the first one was the identification of the stimulus (bitter, sour, salty, sweet, water) and the second one was the valuation of stimulus intensity. Stimulus consisted in 16 sterile solutions containing sapid substances in different concentrations and 1 solution containing water as placebo, at the temperature of 24°C; stimulus intensity was evaluated through an analogical scale from 0 to 10 (where 0 corresponds to a neutral stimulus = water, and 10 to the maximum intensity). The data were collected and, through statistical analysis, mean and standard deviation of threshold values and intensity, in four administrations, for each taste and for placebo, were obtained. Validation of taste test was obtained comparing the results of group A and group B, collected from different operators.
Results: Threshold and intensity values were similar, with a no statistical significant difference ($P > 0.05$) for all the four sapid solutions and for placebo.
Conclusions: The identification of normal taste parameters in healthy children will allow comparing, in a following phase, dysgeusia conditions in childhood.

