

Miscellaneous Abstracts

Oral Presentation

The Prevalence of Hearing Disorders among the 3-6 Years Old Children of Kindergartens of Yazd City

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Objectives: A hearing – impaired is defined as abnormal or reduced function in hearing resulting from auditory disorder. The goal of any preschool and schools screening program should be to accurately identify those children whose hearing has impaired resulting from either conductive and / or sensory – neural pathology.

Methods and Subjects: This research was carried out in cross – sectional descriptive study on 577 children (299 girls and 278 boys) among 3-6 years old children of kindergartens of the Yazd city from September 2005 to January 2006. The otoscopy examination, pure-tone screening and impedance were conducted after completion the parent awareness form of the hearing loss existence.

Findings: In this study, there were 12.6% abnormal conditions of external ear canal, 34.2% abnormal tympanic membrane, 35.9% abnormal tympanograms and 13.4% hearing loss.

Conclusion: With respect to high prevalence of need to medical care and the negative effects of middle ear disorders in learning of preschool children, and also due to the importance of early identification and intervention of hearing loss in aural rehabilitation programs, a warning and teaching the people about the effects of hearing disorders and it's prevention and identification are very important.

Key Words: Hearing screening; Hearing disorders; Universal Hearing Screening

Oral Presentation

Non Seeing Baby

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There are some babies who can not see, or at least it seems that they can not see. Mostly these are the babies who have some systemic problems and mainly problems involving central nervous system. Both parents and the physician think that it is a non seeing

baby which many times it is the baby who can see but not reacts to visual stimuli. On exam they don't have good fixation or following and no positive finding in their visual pathway and this is the point that should be considered by pediatrician when refers the baby who is suspicious to have some ocular findings.

Key Words: Non seeing baby; Blindness; Children

Oral Presentation

Enhancing Patient Safety in Pediatric Practice: Learning from Malpractice Claims in France

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Objectives: To examine pediatric malpractice claims and identify common characteristics likely to result in malpractice in children.

Methods and Subjects: First, we did a retrospective and descriptive analysis of all pediatric malpractice claims involving children aged 1 month to 18 years, in which the defendant was coded as pediatrician or *general practitioners*, reported to GAMM during a five-year period (2003-2007). Then, a comparison of these results with those from the United States was performed.

Findings: From 2003 to 2007, there were 10673 claims reported to GAMM. Pediatricians accounted for 1.2 % of these claims. The average annual incidence of malpractice claims was 1.1/100 pediatricians. Malpractice claims were more frequent (41%) with more severe outcomes in children younger than 2 years of age (52% deaths or major injuries). Meningitis (n=14) and dehydration (n=13) were the leading causes of claims, with highest mortality rates (93% and 92% respectively). The most common alleged misadventures were diagnostic related error (47%), and medication error (13%). Malignancy was the first medical condition incorrectly diagnosed (14%).

Conclusions: Pediatric malpractice claims are less frequent in France than in the United States, but they share many similarities with those in the United States. These data would enhance the knowledge of high-risk areas in pediatric care that could be targeted to reduce the risk of medical malpractices and to improve patient safety.

Key Words: Patient Safety; Claims; Malpractice

Poster Presentation

Prevalence of Habitual Snoring in Children of Elementary School in Khorramabad in 2006-2007**Azam Mohsenzadeh, MD**

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Objectives: A number of different epidemiologic studies suggest that about 10% of children are habitually snore. It was once thought that snoring alone in both children and adults was relatively benign. However, in addition to being a relative risk factor for the later development of OSA, there may be a link between snoring, with or without associated breathing problems, and poor school performance. This study is performed to evaluate the prevalence of habitual snoring in children of elementary school 7-12 years old in Khorramabad in 2006-2007.

Methods and Subjects: In this cross sectional study, 364 students were selected randomized in both sexes male and female with equal numbers. Information was collected with questionnaire named TUCASA.

Findings: Out of 364 students 73 students had habitual snoring with prevalence of 20/3%. 22/3% was boys and 18/3% girls. Statistical tests showed that between teeth grinding and snoring, sleep apnea and snoring, open mouth breathing and snoring, there is significant association. (p-value <0.001).

Conclusion: Habitual snoring is a significant problem for children and may be associated with diurnal symptoms. Recommended, increase parents information about necessity of medication and its effect on children cognition.

Key Words: Prevalence; Habitual snoring; Elementary school children

Poster Presentation

Pierre Robin Malformation and Outcome**Hossin-Ali Nazemian, MD**

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Pierre Robin Sequence (PRS) is a condition that usually present with severe problem at birth despite of benign nature. It is also known as Pierre Robin Syndrome or Pierre Robin Malformation, that is a congenital condition of facial abnormalities in humans. It is the name given to the following birth defects if they appear together: small lower jaw (micrognathia), posterior displacement or retraction of the tongue (glossoptosis), breathing problems, horsehoe-shaped (U-shaped) cleft palate may or may not be present. In addition most of patient may

experience the following problems: feeding problems in infancy, ear infections and hearing defects. Children affected with PRS usually reach full development and size. However, it has been found internationally that the child is often slightly below average size, raising concerns of incomplete development due to chronic hypoxia related to upper airway obstruction as well as lack of nutrition due to early feeding difficulties. However, the general prognosis is quite good once the initial breathing and feeding difficulties are overcome in infancy. Most PRS babies grow to lead a healthy and normal adult life. This syndrome, which often undermines several organs and systems, is treated with a multidisciplinary approach that involves several specialists. We present a case of Pierre Robin syndrome who treated under observe with appropriate position (prone), nasal oxygen therapy and feeding via nasogastric tube (NG). Decision making is according to the special patients condition and some patients need to emergency procedure like tracheostomy. Alternatively distraction osteogenesis is a good technique to correct micrognathia. In severe Pierre Robin sequence cases, temporary tongue-lip traction was a good adjunct to distraction osteogenesis because this Method avoided tracheostomy.

Key Words: Neonatal Respiratory Distress; Pierre Robin malformation

Oral Presentation

Clinical Manifestations of Meniere's Disease in Children**Bahram Malakooti, MD**

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Meniere's disease is rarely found in children, despite its frequency in adults and literature regarding it is very scarce. The diagnosis of Meniere in childhood is difficult, owing to a number of age-related factors. Here we report eight cases of Meniere's disease in children less than 8 years old who referred to our clinic during the past year. Patient's histories include intermittent vertigo attacks and cochlear findings, but neurologic examinations were normal. Complete behavioral and electrophysiology-auditory evaluations were made. Compared with hearing loss, vestibular symptoms such as vertigo regardless of their disturbance are often overlooked in pediatric patients. For children with vestibular problems, a test battery approach is necessary to ensure quality patient care. The existence of newborn hearing screening, one of the major achievements of medicine during the past 50 years, has led to the widespread recognition that all forms of hearing

impairments may not be expressed at birth and that hearing loss has many different causes which all need to be diagnosed early so that therapeutic/rehabilitation interventions be started as soon as possible. More issues will be discussed.

Key Words: Meniere's disease; Hearing loss; Neurological examinations; Children

Poster Presentation

Fatherhood Experiences During the Child's First Year: A Qualitative Study

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Objectives: Societal and economic shifts have expanded the roles that fathers play in their families. The importance of fathers' participation for development of the child and the well-being of the family is recognized from earlier research. The aim of the study was to explore experiences of the first year as father.

Methods and Subjects: In this study, the qualitative phenomenological approach was used by several in depth, semistructured interviews with 15 fathers that were selected by purposive sampling during in summer and spring 2009 in Tehran city which their first child born. Coding and analysis were done using content analysis to identify themes. The rigor of present study was based on transferability and credibility.

Findings: Essential themes emerged as the core categories: Being confident as a father and as a partner, increased responsibility for childcare, joy and fun in the relationship with the infant, spend as much as possible time with the child and his wife, found meaning in life and a deep feeling of togetherness, struggling with the limited time available for oneself, occasionally being fatigued, alternating between work and home, also became confused and they experienced chaos in their life and conflict between several aspects of equal value in life for example work, hobbies, friends, and family, became concerned on future of child, Fathers also expressed feelings of sadness at not having had any sexual relations after the birth.

Conclusion: First time fathers experienced a changing life, which included becoming a father, alternating between work and home, changing relationship towards partner and developing relationship towards their child. Although becoming a father was a fantastic experience, knowledge about first time fathers' experiences during infancy may increased support to both fathers and child for child care from midwives and child health nurses.

Key Words: Fatherhood experiences; Child's first year; Qualitative study

Oral Presentation

The Effects of Mouth Breathing on the Vital Organs of Children

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Breathing through the correct respiratory passage (which is nose not mouth) is a vital issue, and the effective factors changing this passage shall be carefully studied, which has not attracted attentions until yet. Indeed, only the effects of this change in the respiratory passage, which can be easily addressed, are studied, and whereas the complications are appeared at advanced ages, therefore this disorder is tried to be cured using medications, and auxiliary equipments such as oxygen making machine during night's sleep. However, in recent years various scholars of different countries are researching the effects and complications of the change in breathing passage. I as a dentist, who has studied more than 33 years the reasons of the respiratory disorders of children and the related complications, believe as follows:

If respiratory passage is divided into two parts of upper frontal and lower posterior, it can be concluded that:

Different researches conducted on the ill effects on vital organs of body indicate that tightness of upper jaw of children cause undesirable effects and various complications in the following organs: In oral cavity, it causes tooth decay, early losing teeth; premature touch of teeth and corrosion of condyles; jaw dislocation; pain in the muscles of face, headache, pain in the muscles of neck and backache; inflammation and infection of tonsils and adenoid; In the general system, it causes rhinitis, sinusitis, otitis media, pharyngitis, bronchitis, Asthma, hypoventilation, hypoxia, pulmonary hypertension, bed-wetting, OSA, misbalance of hormones, fatness, behavioral disorder, ageusia, Anosmia. The decrease of oxygen in by 40% causes hypoxia, which enhances the resistance of pulmonary vessels. If the factors causing hypoxemia increases, the probability of the contraction of pulmonary vessels and hypertension will increases too. Any increase in pulmonary resistance increases heart load and expands the right ventricle of heart. The obstruction of nose may lead to pulmonary expansion. The obstruction of air passage way chronically increases the pressure of pulmonary arteries, in such a case, the systolic and diastolic pressure will be more than normal degree. Hypoxemia decreases the oxygen in the arterial blood (PO₂), and increases the amount of CO₂ in the blood of artery (Pco₂) by oral breathing.

Oral breathing causes acidosis. According to our research conducted 14 years ago, the pressure of

blood P02 after orthodontic therapies increased up to 39%, and the pressure of Pco2 decreased up to 39%. Moreover, it is pertinent to mention that the colleagues shall not prescribe medications, which causes acidosis of blood, to the patients breathing orally. Due to acidosis, oral breathing influences the central nervous system negatively and weakens its function. It has improper effects on sympathetic and parasympathetic systems, e.g. it influences phrenic nerve and prevents the diaphragm to pace sufficiently, and that influences breathe inhalation and exhalation. Oral breathing has negative effects on brain vessels, decreases intelligent quotient, influences kidneys and their functions negatively. It weakens the sense of hearing of children up to 60%, causes otitis and infection of ears, inflation of mastoid, meningitis, and encephalitis. o It decreases the secretion of endocrine glands, whose malfunction influences all organs negatively and changes the metabolism and prevents physical growth. o Oral breathing and inhalation or air has negative effects on digestive system and cause gastritis. One of the other breathing disorders which may increase is apnea. Hypoxia has other effects such as, sleepiness, indifference, and delay in reacting, restlessness, irritability, mental and intellectual disorder, work disability. In a few Words, all above-mentioned factors have been observed and studied by me, and they can be presented in brief to our colleagues as orthodontists, in order to show the problems of oral breathing and provide them with some solutions, which are effective to remedy several parts of these disorders.

Key Words: Mouth Breathing; Oral breathing; Sleepiness

Poster Presentation

Cued Speech Can Improve Communication Abilities of Hearing-Impaired Children

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Cued Speech is a system of communication used with and among deaf or hard of hearing people. It makes traditionally spoken languages accessible by using a small number of handshapes (representing consonants) in different locations near the mouth (representing vowels), as a supplement to lipreading. It is now used with people with a variety of language, speech, communication and learning needs. This study was designed to investigate the effect of Cued Speech on the communication abilities of hearing-impaired children. Subjects were 8 profoundly hearing-impaired children at the age of 9-12 years

they were taught Cued Speech for five months in an everyday 45-60 minutes sessions program and matched with 8 hearing impaired children as the control group. Both control and experimental groups were presented with Auditory Perception test (APT-HI) as pretest, post-test and follow-up. Mixed Repeated Measurement was used to analyze the Results. Cued Speech substantially improved performance of the experimental group but there was no significantly important difference between pre-test, post-test and following scores of the control group). Results support the use of Persian Cued Speech for developing auditory perception abilities of the hearing-impaired children.

Key Words: Cued speech; Deaf; Communication abilities

Poster Presentation

Management of Oral and Dental Complications in Children Receiving Head and Neck Radiotherapy

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Management of oral & dental complications in children receiving head and neck radiotherapy Radiotherapy of the head and neck can cause numerous side effects such as mucositis, sialadenitis, xerostomia, dental caries, infections, trismus, osteonecrosis, abnormal curvature or root shortening in teeth, hypodontia, microdontia and hypocalcification. The child's age at the beginning of treatment, type, intensity and frequency of radiotherapy are important factors in the genesis of oral and dental abnormalities. It is necessary for the parents and the patients to be informed about acute and late effects of treatment. Radiotherapy can be divided into three time periods: before, during and after therapy. The involvement of dental professionals with radiation-oncology team can reduce the risk of oral and dental complications. Assessment of oral and dental status and treatment of pre-existing diseases prior to radiation therapy are critical. During and after radiation therapy, dental professionals must manage the patient by considering individual needs and presence of complications. The scope of this article is to review the oral & dental complications of head and neck radiation therapy in children; it also emphasizes the role of the dentist in the multi disciplinary team.

Key Words: Head and neck radiotherapy; Children; Oral complications

Oral Presentation

Eye Movement Desensitization and Reprocessing

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Posttraumatic stress disorder (PTSD) is characterized by a set of symptoms such as re-experiencing symptoms, distressing recollections, persistent avoidance, and hyperarousal in response to exposure to one or more traumatic events. Many children and adolescents are exposed to physical or sexual abuse, domestic violence, motor vehicle accidents, severe medical illnesses, or natural or human created disasters, leading to full-blown PTSD in some, and at least some PTSD symptoms in many others. PTSD occurs frequently in children and adolescents with up to 6 percent of youth meeting criteria for this diagnosis at some point. Developmental factors strongly influence the manifestations of symptoms. For example, re-experiencing of a traumatic event is often observed through play, recurrent nightmares without recall of the traumatic events, and behaviors that reenact the traumatic situation, along with agitation, fear, or disorganization. Childhood-PTSD is also associated with increased rates of other anxiety disorders in addition to depressive episodes, substance use disorders, and attentional difficulties. For many children and adolescents with milder forms of PTSD, symptoms may persist for 1 to 2 years after which they diminish and attenuate. In more severe circumstances, however, PTSD syndromes persist for many years or decades in children and adolescents, with spontaneous remission in only a portion of them. The prognosis of untreated PTSD has become an issue of growing concern for researchers and clinicians who have documented a variety of serious co-morbidities and psychobiological abnormalities associated with PTSD. Children and adolescents with histories of physical and sexual abuse have been found to exhibit higher rates of depression and suicidality themselves and in their offspring as well. This highlights the importance of early recognition and treatment of PTSD among youth that may significantly improve the long-term outcome for them. This article presents a variant of trauma-focused cognitive-behavior therapy for PTSD is called eye movement desensitization and reprocessing (EMDR) in which an exposure and cognitive reprocessing interventions are paired with directed eye movements.

Key Words: EMDR; PTSD; Child and Adolescent

Poster Presentation

Traditional Practices Affecting Maternal Care: A Qualitative Study from Iran

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Objectives: Health care is affected by people's culture, so identification of it is necessary for changing the health behaviors of people. The aim of this study was to discover the traditions of people in Gorgan concerning maternity care.

Methods and Subjects: A qualitative approach was applied. Data were collected through individual deep interviews with key elderly people from Gorgan. Open ended questions were about the traditions of labor, bearing, postpartum nutrition of the mother, umbilical cord, about the newborn, mother and newborn garments and breast ache. The data were coded and classified.

Findings: The Results indicate some identified traditions were useful and some were harmful or had unknown effects. The practices can be divided as psychological or physical support. Here are a few examples: "To keep the parturient safe; Putting an onion close to the bed as a symbol of having an attendant. To make the birthing process easier: Giving the parturient's comb to a special clergy (Seyyed) and asking him to bless it. Bringing a piece of green cloth from a Seyyed for the parturient and wearing it around the wrist or abdomen. Asking the parturient's mother-in-law to throw a little of her saliva in a glass of water and giving it to the parturient to drink or putting Panjeh-Maryam plant in water for the parturient to drink."

Conclusion: The traditions can influence the health care behavior of the people and they should be identified and analyzed for "change" of health related behaviors

Key Words: Tradition; Maternity; Newborn; Health care

Poster Presentation

Strategy of Treatment of Ciliary Dyskinesia

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Primary ciliary dyskinesia (PCD), also known as immotile ciliary syndrome or Kartagener Syndrome (KS), is a rare autosomal recessive genetic disorder caused by a defect in the action of the tiny hairs (cilia) lining the respiratory tract. Specifically, it is a defect in a gene coding for left-right dynein (Ird), a key structural protein in cilia. The diagnosis may be

difficult because symptoms are not specific and the tests identifying the abnormal ciliary beat pattern, frequency and structure are available only in a few specialized centres. Pulmonary symptoms include excessive phlegm, cough and recurrent infections. Most importantly, the prognosis of the respiratory disease seems to be much more favourable in PCD with stabilization of the disease with early, adequate and aggressive management.

Respiratory Monitoring: The airways become infected and inflamed because of excessive secretions which accumulate in the airways due to impaired mucociliary transport. Regular cough swab cultures in young or non-expectorating children, or sputum cultures in productive patients, are recommended. Fiber-optic bronchoscopy may be indicated in non-expectorating patients, who do not respond to conventional antibiotics, or in patients with persistent atelectasis. Pulse oximetry should be performed at every visit. Chest X-rays are insensitive and are recommended only during a respiratory exacerbation that is not responding to standard therapy. The diagnosis of bronchiectasis is best made by high-resolution computed tomography (CT) scan.

Medical Treatment: Early therapeutic intervention results in better symptom control and slowing of deterioration in lung function. Bronchial hyperresponsiveness may be observed in patients with chronic suppurative lung disease. The prescription of bronchodilators should thus be on an individual basis, after the demonstration of a significant bronchodilator effect. Neutrophilic airway inflammation and elevated levels of interleukin (IL)-8 have been observed in children with PCD. rhDNase Recombinant humanDNase (rhDNase) reduces sputum viscosity by degradation of extracellular DNA which may translate into an improvement in clinical symptoms and lung function. Airway clearance by combinations of chest physiotherapy and physical exercise, together with antibiotics, is standard treatment for patients with PCD. Physical activity should be an integral part of the management of patients with PCD. PCD patients should receive all immunizations, including pneumococcal and yearly influenza A. Contact with infected individuals should be avoided. If the diagnosis is made in the first months of life, prophylaxis against respiratory syncytial virus by means of palivizumab during the first winter may be justified. Any exposure to respiratory irritants, such as tobacco smoke, should be avoided. Any associated respiratory risk factor, such as malnutrition or gastro-oesophageal reflux, should be treated. Finally, surgical removal of localized irreversible bronchiectasis may be proposed in selected cases. Monitoring and treatment of the lower airways is not evidence based in PCD and derives largely from CF protocols. This aggressive therapeutic attitude has proven to be beneficial in maintaining a relative good and stable lung function in patients, particularly those diagnosed at an early

stage of their disease, before the presence of irreversible lung damage. Antibiotics and chest physiotherapy are the cornerstones of the respiratory management, while waiting for new adjunct therapies such as anti-inflammatory and mucoactive agents.

Key Words: Immotile ciliary syndrome; Primary ciliary dyskinesia; Recombinant humanDNase

Poster Presentation

Ski sickness in Adolescents Skiers

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Objective: There is shortage of knowledge about medical problems in adolescent skiers. This study aimed to determine frequency of medical signs and symptoms during or after ski among adolescent skiers.

Methods and Subjects: This cross sectional study was performed in 12 consecutive weekends in the winter of 2005 at Dizin ski pist in Iran. All adolescent skiers (<16years) who had been entered the pist during this time period were enrolled into the study. A questionnaire including demographics, duration of transport to the pist, wearing glasses and contact lenses for medical and non medical applications, duration and frequency of skiing and development of signs and symptoms of ski sickness during or after skiing was filled for each participant. In addition to determination of frequency of medical problems, association of mentioned variable with medical problems was investigated.

Findings: Of 162 skiers, 111 subjects (68.5%) were male. The mean age of the participants was 14.7±2.1 years. Of them, 127 skiers wore glasses or lens during skiing. Visual disorders including myopia, hyperopia and astigmatism were found in 24 skiers (14.8%). Our study showed that the frequency of main ski-related signs and symptoms varies from 0 to 10.5%. There was significant association between occurrence of signs and symptoms and presence of visual disorder (P=0.01).

Conclusions: Our results showed that development of ski-related signs and symptoms is relatively frequent. These sign and symptoms have association with minor ophthalmologic problems such as myopia or astigmatism.

Key Words: Ski sickness; Visual disorder; Vestibular

Oral Presentation

Prenatal Diagnosis of Non-classical Hyperphenylalaninemia (non-PKU HPA); Mutation in PCBD1 Gene

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Neonatal screening of phenylketonuria (PKU) is in effect in several provinces in Iran. PKU is inherited in autosomal recessive manner and Iran is regarded as high incidence area in the world. Affected child has one or several of the following symptoms, mental retardation, convulsions, disturbance of tone and posture, drowsiness, irritability, abnormal movements, recurrent hyperthermia without infections, hypersalivation, and swallowing difficulties. Families already having at least one affected child are at risk of having another one. Prenatal diagnosis can now be offered for at risk families. PKU prenatal diagnosis is complicated by another hyperphenylalaninemia (HPA) disorder called BH4. Hyperphenylalaninemia (HPA) may also result from the impaired synthesis or recycling of tetrahydrobiopterin (BH4), the cofactor in the phenylalanine, tyrosine, and tryptophan hydroxylation reactions GTPCH, PTPS, DHPR or PCD Non-PKU HPA may result by deficiencies of one of the following genes, GTPCH, PTPS, QDPR or PCBD1. Our center is one of the centers recognized for performing prenatal diagnosis for PKU. In one of our patients, after sequencing all coding regions of PAH gene, no mutation was found. We then designed primers for the genes responsible for BH4 phenotype named above. The affected child showed homozygosity for the cd105TAG>CAG(Ter>Gln) mutation in PCBD1 gene. Prenatal diagnosis was performed for the fetus. Prenatal diagnosis and carrier detection now can be performed for all HPA patients by applying complete gene sequencings of the above genes in our center.

Key Words: Phenylketonuria; PKU; PCBD1 gene

Poster Presentation

Cytogenetic Study of Sexual Ambiguity in Intersexual Children in Fars Providence

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Objective: Intersexuality is a condition in which genitalia and/or secondary sexual characteristics determined as neither exclusively male nor female, or with combine features of the male and female sexes. Between 0.1% and 0.2% of live births are ambiguous

enough to become the subject of specialist medical attention. There are several genetic and/or endocrine causes of sexual ambiguity. One of the first and most important steps to detect that is cytogenetic study of patients. Here we analysis the karyotype pattern of the intersexual children referred to ACECR genetic lab.

Methods and Subjects: Twenty intersexual children, from newborn to 15 year-old age, were referred to genetic lab of Iranian Academic Center for Education, Culture & Research (ACECR), Fars Province Branch during 2001- 2008. They were socially 17 males and 3 socially females with problems in their internal or external genitalia such as small testis, small penis, absence of uterus, ovarian, prostate or testis , ambiguous genitalia etc. karyotype of these patients were analysis by G-banding technique.

Findings: Out of 20 patients recruited in this study, 1 case was male pseudohermaphrodit (46,XY) and 1 case female pseudohermaphrodit (46,XX). 7 cases showed 46,XX/46,XY karyomerism karyotype include 5 socially men and 2 socially women. In 11 patients, karyotype were in conformity with their social sex (46,XY males).

Conclusion: Results show that abnormal karyotype accounts for approximately half of the sexual ambiguity and the rest relate to other genetic or endocrine issues which are not detected by common karyotype methods. Recognition of the true explanation for the reason of the problem will relief the family and helps them and physicians to treatment or corrective surgery, in addition to decide about later pregnancy. However applying other advance genetic methods in order to find the actual reason of the intersexuality of these patients is more useful and reasonable.

Key Words: Intersexuality; Sexual ambiguity; Karyotype; Genetic

Poster Presentation

Necessity of More Attention to Karyotype Analysis in Children Suspected to Autosomal Chromosome Abnormalities

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Objective: Chromosomal abnormalities affect at least 7.5% of all conceptions. Most of these abnormalities are spontaneously aborted and the frequency in live births is 0.6%. Unfortunately very few reports about chromosome analysis in Iran exist. So we performed a study to provide information about results of karyotyping in one of the cytogenetic lab in south of Iran.

Methods and Subjects: Karyotypes of 4151 individuals referred to genetic lab of Iranian Academic Center for Education, Culture and Research (ACECR), Fars province branch were analyzed for suspected chromosome abnormalities from 2001 to 2009.

Findings: Among 701 patients with known reason of referring to this lab 180(25.6%) were suspected to autosomal chromosome abnormalities and the others were suspected to sex chromosome abnormalities. Of the 4151 cases, chromosome aberrations were identified in 230 (5.64%). 108 (47%) autosomal alterations and 122(53%) sexual abnormalities were found. Among the 108 autosomal abnormal karyotypes, 103 (95.37%) were of numerical abnormalities and the remaining 5 (4.63%) had structural abnormalities. Among the autosomal abnormal karyotypes, the most frequent (92.57) was Down's syndrome. Other numerical abnormalities found were trisomy 18, trisomy 13 and triploidy. Among patients with Down's syndrome, free trisomy 21 constituted 74% of the cases and mosaicism made up 26% of them. The mean age of the patients with autosomal aberration was 4.25 years old and male to female ratio (sex ratio, sr) for them was 1.30.

Conclusion: The frequency of patients referred to this lab suspected to autosomal chromosome abnormalities and also percent of determined autosomal abnormal karyotypes were less than what we expected in base of previous studies. Less referral of patients with clear phenotypes of Down's syndrome, short length of life in children with autosomal chromosome abnormalities and lack of attending to the importance of cytogenetic analysis for such disorders can explain the reason of this situation. According to importance of chromosomal diagnosis in providing of information about the recurrence risk for future siblings, more attention to karyotype analysis in children suspected to autosomal chromosome abnormalities is necessary.

Key Words: Karyotype; Autosome; Chromosomal Abnormalities

Poster Presentation

Mucopolysaccharidosis Type III B in a Dizygotic Twin

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The mucopolysaccharidoses are a clinically and genetically heterogenous group of lysosomal storage diseases caused by defects in different enzymes for mucopolysaccharide degradation (glycosaminoglycans). Except for mucopolysaccharide storage disease type II (Hunter), all are transmitted by autosomal recessive inheritance. Clinical feature of these metabolic disorders result from accumulation of

mucopolysaccharides in various organs. In aspect of clinical manifestations & enzyme defect they have been classified. This case report is about a dizygotic twin, a boy and a girl, of a second cousin couple with short stature, scoliosis and characteristic bony changes (dysostosis multiplex), progressive and profound mental retardation, coarse facies and hepatosplenomegaly in both of them & also umbilical hernia in girl. Both of them have conductive hearing loss. Urinary mucopolysaccharide was positive in them, biochemical analysis shows heparan sulfate. Therefore we requested evaluation of heparan sulfamidase, alpha - N- acetyl hexosaminidase & N-acetyl glucosamine 6 sulfatase activity in their leukocytes that showed very low activity of alpha-N - acetyl hexosaminidase (glucosaminidase) and confirmed the diagnosis of Sanfilippo (MPS type III) B. Their parents attempt to have a healthy offspring which is now possible with prenatal diagnosis of fetus by CVS (choriovillosus sampling) and enzyme assay.

Key Words: Mucopolysaccharidosis Type III B; Heparan Sulfate; Alpha N Acetyl Hexosaminidase; Sanfilippo B

Poster Presentation

Management of Oral and Dental Complications in Children Receiving Head and Neck Radiotherapy

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Management of oral & dental complications in children receiving head and neck radiotherapy. Radiotherapy of the head and neck can cause numerous side effects such as mucositis, sialadenitis, xerostomia, dental caries, infections, trismus, osteonecrosis, abnormal curvature or root shortening in teeth, hypodontia, microdontia and hypocalcification. The child's age at the beginning of treatment, type, intensity and frequency of radiotherapy are important factors in the genesis of oral and dental abnormalities. It is necessary for the parents and the patients to be informed about acute and late effects of treatment. Radiotherapy can be divided into three time periods: before, during and after therapy. The involvement of dental professionals with radiation-oncology team can reduce the risk of oral and dental complications. Assessment of oral and dental status and treatment of pre-existing diseases prior to radiation therapy are critical. During and after radiation therapy, dental professionals must manage the patient by considering individual needs and presence of complications. The scope of this article is to review the oral & dental

complications of head and neck radiation therapy in children; it also emphasizes the role of the dentist in the multi disciplinary team.

Key Words: Head and neck radiotherapy; Children; Oral complications

Poster Presentation

Consanguinity Marriage and Neonates, Congenital Malformation

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Objective: Congenital malformation (CM) will begin to emerge as one of the major childhood health problems. Inheritance patterns of CM include : single gene, mutation, polygenic and multifactorial condition and chromosomal abnormalities. An accurate diagnosis of CM is of utmost importance in being able to treat the condition and anticipate any future health concerns that may arise .The aim of this study was to determine and compare frequency of CM in parental consanguinity.

Methods and Subjects: In a descriptive-observational cross-sectional study , all births of whole maternity hospitals in Yazd from October 2005 to June 2006 evaluated from view of prevalence of congenital malformation, types, patterns and parental consanguinity.

Finding: The overall prevalence of CM was 2.8% (136/4800) .Two point seven percent (2/7%) of live births and 12.6% of stillbirths had CM. Overall, musculoskeletal (0.83%), central nervous system (0.47%) and genital system (0.37%) were accounted as the most common types of CM. Parental consanguinity was seen in 30/6% of all births with CM ,among whom first cousin marriages occurred more frequently. Frequency CM in parental consanguinity and not related parents were 2.9% (43/1469) and 2.8% (93/3331) respectively (P=0.2) Therefore, no statistical difference was found between CM and parental consanguinity.

Conclusion: However, parental consanguinity is known to increase the risk of birth defects in offsprings, especially with autosomal recessive disorders, but other conditions like prematurity , teratogens exposure , infection of mother and environmental factors must be concerned and excluded before assigning an inherited cause.

Key Words: Congenital malformation; Neonate; Parental consanguinity

Oral Presentation

Amelogenesis Imperfeta

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Amelogenesis imperfeta refers to a group of inherited tooth disorders characterized by abnormal enamel formation. The predominant clinical features of patients are enamel hypoplasia in which enamel is normal but thin, hypomineralization including hypomaturation and hypocalcification and/or combined phenotype. So far four genes have been documented that associated with AI: ENAM, KLK4, MMP20 and AMLEX genes. We performed molecular genetic studies on 10 Iranian families with different models of inheritances. Of course most of our patients were autosomal ressesive model of inheritance in this study, two genes including ENAM and MMP20 which account for majority of AI and autosomally inherited were chosen for mutation detection by SSCP and DNA sequencing. Our result from SSCP revealed genetic alteration of interin 5 of MMP20 gene. We found genetic changes in two patients with autosomal ressesive model of inheritances, they were both had mutation in intron 5 of MMP20 gene. Interestingly these mutations have not been yet reported. Therefore for the first time we demonstrated that other parts of these genes are pron to mutations in genes involved in tooth development. We did not detest any mutation in intron 9 and exon 10 of ENAM gene. In order to identify the type of mutation samples will be subjected for DNA sequencing . the data presented here is in agreement with the previous studies that suggested these genes associated with tooth disorder. Taken together these findings support MMP20 as a disease gene and opened a new window on the molecular mechanism of AI disease and to the function of the amelogenin protein in enamel formation. This study can help researches who are interested to work in molecular genetic and so develop the knowledge of this disease.

Key Words: Amelogenesis imperfeta; Gene; Enamel hypoplasia