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Poster Session P16 – Syndromes and Genetics

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Poster Session P16/Syndromes and Genetics

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Zinsser-Engman-Cole syndrome: a case report

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Introduction: Zinsser-Engman-Cole (ZEC), (OMIM #305000) syndrome is a rare, often fatal, inherited disease. The main clinical features of this multi-system bone marrow failure disease are mucocutaneous, haematological anomalies and an increased predisposition to cancer opportunistic infections and other abnormalities. The orodental manifestations described in this syndrome are leukoplakia, early tooth loss and dental anomalies.

We report one case of this syndrome to illustrate the orodental features.

Clinical management: We present the case of twin boys, 15 years old, affected with ZEC. One of the two boys died after serious septic cutaneous complication (probable leprosy). The orodental findings observed in the case reported here are multiple caries and periodontal disease with resorption of alveolar bone and premature tooth loss. The other clinical signs are skin pigmentation, nail dystrophy, sparse hair, leukoplakia in the mouth and testicular atrophy. The treatment consisted of extraction of damaged teeth under antibiotic treatment with conservative treatment of the other teeth and achievement of total prosthesis of the upper maxillary and partial prosthesis of mandible.

Regular monitoring has been established to maintain good hygiene for the remaining teeth, and to prevent leukoplakia.

Conclusion: ZEC syndrome also known as Dyskeratosis congenita is a very polymorphous disease where five responsible genes have been identified (DKC1, hTERC, hTERT, TINF2, NOP10) encoding components of the telomeres and telomerases assuming critical functions in aging and cancer. It is important to correctly diagnose ZEC as this can have major implication for the clinical management of the patient.

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Ellis-van Creveld syndrome (case report)

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Introduction: Chondroectodermal dysplasia (Ellis-van Creveld syndrome) is an autosomal recessive syndrome that was initially reported by Ellis and van Creveld (EVC) in 1940. A higher prevalence has been reported among certain populations such as the Amish and the Arabs of the Gaza strip. Today, the syndrome has been described in other population such as our community in Saudiarabia.

Clinical management: A 5-year-old girl presented from Al-Taheel Rehab Center in Riyadh city for examination. Anamnesis showed that the parents belong to the same family and that the sister of the patient had died at the age of 6 months suffering from Tetralogy of Fallot. Clinical examination of our patient revealed Polydactyly, short stature and hairy skin. Oral examination exhibited general caries involvement, conical and peg shaped teeth and a yellowish lump on the left side of the tongue. Under general anaesthesia all destroyed teeth were extracted and the remaining teeth were restored with composite restorations. Surgical excision of the lump on the tongue was performed. Histological examination showed it is fibrous tissue. Follow up examinations are performed every 6 months in the Pediatric Cardiology Clinic.

Conclusion: Genetic counseling is recommended for prospective parents with any family history of (EVC) syndrome. It provides parents with the knowledge to make intelligent, informed decisions regarding possible pregnancy and its outcome. Children with EVCS can live their life normally not needing to be hospitalized unless they have other problem such as heart disease.

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Floating–Harbor syndrome: orofacial manifestations and dental management

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Introduction: Floating-Harbor syndrome is a rare genetic disorder initially described in 1973. Approximately 30 cases have been reported in literature up to the present. A 5-year-old female diagnosed with Floating-Harbor syndrome was referred to the Department of Paediatric Dentistry, Leeds, for dental management. The patient had the major features of the syndrome: short stature, speech delay and a characteristic face. On physical examination patient's height was 91 cm and weight 15 Kg. Learning difficulties were mentioned by parents. Triangular face with prominent nose, deep-set eyes, long eyelashes, short smooth filtrum, thin upper lip and short neck were evident. Extra-oral examination revealed class III skeletal pattern. On intraoral examination caries, extensive surface loss of all incisors, hypomineralised canines and primary molars, as well as malocclusion were diagnosed. Panoral radiographic examination revealed delayed dental age. Delayed bone age and celiac disease are common manifestations of the syndrome, however, only the former was evident in our case.

Clinical management: Dental treatment under general anaesthesia included pulpotomies and stainless steel crowns, extraction of primary teeth with poor long-term prognosis, fissure sealants on non carious teeth and smoothening of sharp edges of anterior teeth. On clinical and radiographic examination 5 months post-operatively, no caries and furcational pathology were present, all restorations were intact and patient maintained very good oral hygiene.

Conclusion: Very few cases of Floating-Harbor syndrome have been reported. The aim of this case presentation is to emphasize the craniofacial characteristics and to give insight into the general features involved in this multi-system condition.

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Rothmund–Thomson syndrome: a case report

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Introduction: Rothmund-Thomson syndrome (RTS) is an autosomal recessive disorder characterized by developmental skeletal abnormalities and poikilodermatous skin changes. The relationship between RTS and immune function impairment is described in the literature. In this case report an association with myelodysplastic syndromes is reported.
Clinical management: In this case report, the dental and periodontal features of RTS in a 6-year-old female are presented. In addition, periodontal bone topography and maxillofacial structures were assessed by an Ultra Cone Beam CT Scanner. Skeletal structures were found normal. Maxillary pre-molars were found to be congenitally missing.Untreated caries lesions were detected in all primary teeth. The gingiva was very fragile and gingival bleeding occured spontaneously. The patient did not allow the restoration of her teeth under local anaesthesia and besides due to the present medical status general anaesthesia was contraindicated.
Conclusion: All the findings indicate that preventive approaches are important and not only the dentists but also haematologists must focus on oral hygiene of patients with RTS before the dental and periodontal problems have occured.

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A case report of Pallister-Killian syndrome (PKS): new dental findings

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Introduction: Pallister-Killian syndrome (PKS) is a sporadic condition with autosomic tetrasomy of chromosome 12 p, resulting in a mosaic distribution of the supernumerary isochromosome. We report a case of PKS affecting an 11-year-old girl with 12 p mosaicism (karyotype 46, XX). Diagnosis was confirmed at 2 years of age. She presented with alopecia, dysmorphic facies, diaphragmatic hernia, PDA and a dilated aorta root. Extra-oral findings included facial asymmetry, low implantation of the ears and sparse eyebrows. The dental findings included delayed eruption, macrodont maxillary right central incisor (or double tooth) with congenital absence of the lateral incisor in both the primary and permanent dentitions. There was an anterior single tooth crossbite, spacing, and a posterior cross-bite.
Clinical management: The discrepancy in size between the maxillary permanent right and left central incisors presented a treatment dilemma, because reduction of right central incisor was impractical, so the left central incisor was built up to improve the aesthetics of the anterior region. All of the invasive procedures were performed under prophylactic antibiotic cover according to the advice of her paediatrician. Due to her delayed dental development, orthodontic treatment has not yet been carried out.
Conclusion: In PKS dental anomalies, such as macrodontia, may be present. The management of this anomaly should be cognisant of the systemic problems and yet able to provide an aesthetic outcome.

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Unusual dental findings in a girl with Russell–Silver syndrome

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Introduction: Russell–Silver syndrome (RSS) is a congenital disorder of unknown aetiology characterised by pre-natal and post-natal growth retardation, short stature, limb asymmetry, facial dysmorphism and oral abnormalities. This case report describes the general features, and in particular the dental findings and their significance, in a Chinese girl with RSS who has been followed up at our dental hospital over the past eighteen years.
Clinical management: This patient was referred to our hospital at 13 months of age because of the presence of an unrepaired cleft palate and failure to thrive. Characteristically, her body weight and height were consistently below the 3rd percentile, with her head circumference on the 25th percentile. She also exhibited mental retardation and bilateral hearing loss. Examination revealed frontal bossing with triangular facies and a cleft of the palate. Furthermore, she presented with down-turned corners of the mouth and small maxillary and mandibular dimensions. Following closure of the palate, her dental development was reviewed and found to be delayed in both the primary and permanent dentitions. Severe crowding and tilting of the teeth were subsequently noted in addition to a double tooth in the primary dentition. Panoramic radiographs have revealed two congenitally missing pre-molars, impacted mandibular first permanent molar, a macrodontic mandibular incisor and some teeth with bulbous crowns and short, fused roots.
Conclusion: Few reports describe unusual dental findings in patients with RSS. Nevertheless, the presence of dental anomalies, especially short roots along with intellectual impairment can limit treatment options, especially orthodontic therapy.

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Ketone utilization disorder and hypodontia

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Introduction: Ketone Utilization Disorder is a deficiency in mitochondrial acetoacetyl-CoA thiolase. Children with lack of this enzyme are unable to break down proteins, resulting toxic build-up in body tissues. There are only 50 cases reported at the literature but there is no evidence for oro-dental findings.
Clinical management: In this study a 10-year-old male patient with the Ketone Utilization Disorder was examined. Dental examination showed normal intraoral soft tissues but there was delayed eruption of anterior mandibular permanent teeth. Also deep caries lesions in the primary molar teeth were detected. In the radiological evaluation, congenital missing of five permanent teeth indicating hypodontia was observed. There was no family history of delayed eruption or hypodontia. Calcium deficiency according to special diet of the patient may explain the caries prone structure of the primary teeth. Dental restorations were completed and it is planned to monitor the patient until the end of the craniofacial growth, when it will be again evaluated and forwarded for the final oral rehabilitation. Dental examination is an important parameter which must be taken into consideration in the future case reports.
Conclusion: Congenital missing of permanent teeth is an important dental observation in this case as no previous publications reported

the association of hypodontia and this deficiency. Further investigation would probably be required to show the relationship of oro-dental findings and Ketone Utilization Disorder.

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Levy-Hollister syndrome – case report

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Introduction: The extremely rare Levy-Hollister syndrome is an autosomal dominant disorder with multiple dysfunction of renal functions and abnormalities in orofacial region.
Clinical management: A Two year-old boy was referred to the *Department* of Paediatric Dent., University Hosp., Hradec Kralove because of recurrent mucous lesions. Based on clinical and ultrasonic examination these orofacial signs were found: unilateral congenital absence of the lacrimal duct, bilateral congenital absence of parotid and submandibular salivary glands resulting in severe dryness of oral mucosa and abnormally shaped primary teeth. Subsequent paediatric and radiographic examination revealed the diagnosis- Levy-Hollister syndrome. Therapy was focused to the substitution of saliva and primary prevention of dental caries.
Conclusion: Early recognition of this disorder and continuous dental care should reduce the long-term morbidity in affecting patients.

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Ectodermal dysplasia – a case report

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Introduction: Ectodermal Dysplasia is a group of closely related conditions involving at least two of the ectoderms (hair follicles, sweat glands, teeth, nails, salivary glands). Oral findings are important and usually multiple abnormalities affect both function and aesthetics. This plays a major role in the psychological development of the child as the condition manifests itself at an early age.
Clinical management: A patient with ectodermal dysplasia requires counseling as well as extensive dental treatments. Here a case report of an 8 year old girl who reported to the dental clinic with complaints of dryness of mouth is presented. Medical history revealed that the child had intolerance to heat, no sweating and lack of lacrimation. The mother had early loss of primary and permanent teeth. For the child a history of extraction of all primary teeth due to rampant caries one year before was elicited. Dental examination revealed absence of all primary teeth. The maxillary and mandibular central & lateral incisors and first molar teeth were present and the orthopantomograph revealed the presence of the remaining permanent tooth buds. A removable partial denture was given to maintain function and aesthetics until the eruption of the permanent teeth. regular follow up and review has been done.
Conclusion: Rehabilitation of the patient with a removable partial denture which was aimed at restoring function provided a big psychological boost to the patient. This emphasizes the need for early diagnosis of oligodontia due to ectodermal dysplasia and its treatment in young children.

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Airway characteristics implications in Pierre Robin Sequence on intubation for dental treatment

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Introduction: Pierre Robin Sequence (PRS) is a congenital disorder including: cleft palate, micrognatia, glossoptosis and airway obstruction. The oral cavity is smaller than usual, muscle development may be poor and the airway itself may be narrow, resulting in breathing difficulties and feeding problems.
Clinical management: A 5-year-old girl was diagnosed as having PRS and severe Early Childhood Caries. Her medical history included: surgical closure of Cleft Palate, Failure to Thrive, Asthma, Strabismus and Mental Retardation. Due to her medical and dental conditions, dental treatment was performed under General Anaesthesia. To avoid hypoxia during induction, a flexible fiberoptic bronchoscope device was used for the intubation. Vital signs were normal throughout the procedure. The recovery was uneventful and the child was released 3 h after the dental treatment was completed.
Conclusion: This case demonstrates the important problems associated with PRS during General Anaesthesia. The use of a flexible fiberoptic bronchoscope device for intubation is mandatory for airway security.

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Management of anodontia in hypohidrotic ectodermal dysplasia

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Introduction: Ectodermal Dysplasia (ED) is a rare inherited disorder in which two or more ectodermally derived structures either fail to develop or are abnormal in their development. Approximately 150 variations exist of which around three-quarters exhibit oral anomalies including abnormalities in eruption patterns, alveolar ridge volume, salivary secretions and most frequently tooth number (hypodontia or anodontia). Hypohidrotic Ectodermal Dysplasia (HED) or Christ-Siemens-Touraine syndrome is the most common form of ED with a reported incidence of 1/10,000 to 1/100,000 births. Affected individuals are characterised by hypohidrosis, hypotrichosis and oro-facial abnormalities.
Clinical management: Two male patients (Child A: 2-years-old; Child B: 4-years-old) with confirmed HED were referred to the Department of Paediatric Dentistry. In each case the parents were concerned regarding the lack of development of the primary dentition and its impact on their child. On extra-oral examination, both children had light-coloured scanty hair, frontal bossing and dry skin consistent with HED. Intraoral examination revealed an absent primary dentition and subsequent radiographs confirmed anodontia in both cases. Following careful case evaluation, each child was provided with a set of complete removable maxillary and mandibular dentures (Child A at 3-years-old; Child B at 4-years-old). Both adapted well to denture wear and their parents perceived an improvement in their overall well-being. At subsequent reviews (Child A over 5 years; Child B over 1 year) both continue to progress well with replacement dentures provided for Child A due to growth.

Conclusion: Appropriate dental intervention in children with HED can help facilitate well-being.

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Phenotype and treatment of amelogenesis imperfecta with unerupted and resorbed permanent teeth

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Introduction: Amelogenesis Imperfecta (AI), the earliest dental syndrome recognized in human ancestors, is a collection of diverse inherited disorders, featuring enamel defects in the absence of significant nondental symptoms. AI phenotypes vary and are categorized as hypoplastic, hypocalcified and hypomaturation types. The different conditions are caused by mutations in a variety of genes that are critical for enamel formation. The aim of this presentation is to describe the phenotype of a rare case of AI and the dental treatment provided.
Clinical management: A.D., a 16-year-old Bedouin girl was referred for dental treatment to the Pediatric Dental Clinic at Barzilai Medical Center, Ashkelon. Her major complaint was brown discolorations of the permanent teeth and missing teeth, a condition that affected some other members of her family. Based on clinical and radiographic oral examination and the family pedigree, she was diagnosed as suffering from autosomal recessive hypoplastic Amelogenesis Imperfecta combined with eruption defects and crown resorption of permanent teeth. Due to the extensive and complex dental treatment, the dental work was performed under general anaesthesia. The treatment included aesthetic composite restorations for all anterior teeth, surgical exposure of unerupted canine, pre-molars and molar and coverage of all permanent pre-molars and molars with SSC. A 6-month follow-up showed improved Quality of Life.
Conclusion: This rare case of AI demonstrates the phenotype variability of this AI disorder and the complex approach needed for restorative treatment.

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Dental management for patient with incontinentia pigmenti: a case report

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Introduction: Incontinentia pigmenti (IP), also known as Bloch-Sulzberger syndrome, is a rare, X-linked, dominantly inherited disorder of skin pigmentation that is often associated with ocular, dental and central nervous system abnormalities. This report describes a patient with incontinentia pigmenti who exhibited multiple missing teeth in both arches. The dental anomalies and treatment procedure are presented.
Clinical management: A girl with incontinentia pigmenti presented with the chief complaint of only two upper incisors being erupted by the age of one year and eight months. Clinical exam found that she had pigmentation over her body including the extremities and trunk. One of her eyes was blind. Intraoral exam found that only two maxillary peg-shaped incisors had erupted. After thorough palpation of her ridges, more tooth buds were suspected to exist. Therefore, it was planned to supply the child have dentures made after all of the expected primary teeth had erupted. Two years after the first appointment, 8 more teeth had erupted, the 4 canines being peg-shaped. Dental treatment under general anaesthesia was planned due to her poor cooperation. Full mouth rehabilitation was performed; the pegged incisors restored using composite resin. Maxillary and mandibular dentures were fabricated and delivered. After fluoride application, periodic follow up has been done every 3 months. After 18 months of follow-up, the dentures are functioning well and so far, no more primary teeth have appeared.
Conclusion: Patients with IP need interdisciplinary coordination; thereby resulting in better treatment and improved quality of life. Routine follow-up and consulting ophthalmologists, dentists, and neurologists are essential.

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Uncommon oral findings in Ellis-van Creveld syndrome: a case report

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Introduction: Ellis-van Creveld syndrome (EVC) is an autosomal recessive disorder characterized by short ribs, polydactyly, growth retardation and ectodermal and heart defects. Common dental findings of EVC are abnormal fraenal attachments, malformed teeth, supernumerary teeth, hypodontia, eruption disturbances and enamel hypoplasia. In the present case, uncommon dental findings of the syndrome including talon cusp, taurodontism, double tooth and root dilaceration are presented.
Clinical management: A 3.5-year-old girl with EVC was referred to the paediatric dentistry clinic. Intraoral examination revealed constricted palate and shallow upper and lower sulci. There were multiple abnormal upper and lower fraenal attachments along with a broad upper labial fraenum. A talon cusp was detected on carious maxillary primary left central incisor. The mandibular primary right molars and mandibular primary left first molar were also carious. The teeth were generally malformed. In the radiographic examination missing maxillary and mandibular primary lateral incisors were detected. The mandibular primary left central incisor was horizontally impacted since its root was dilacerated. There was an unerupted permanent double tooth located in the right maxillary anterior region. Taurodontism of the mandibular primary first and second molars was also detected radiographically. The carious teeth of the patient were restored with a poly-acid modified resin composite (Dyract eXtra, Dentsply DeTrey, Konstanz, Germany). The patient has been under control for 8 months to observe any changes in the mouth.
Conclusion: Although EVC has typical oral mucosal and dental anomalies, the presented uncommon oral findings may help to establish a proper diagnosis of the syndrome.

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Infantile Malignant Osteopetrosis: dental findings and management in 2 siblings

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Introduction: Infantile Malignant Osteopetrosis in children is a rare autosomal recessive genetic disorder characterized by impaired osteoclastic functions. The estimated prevalence is 1 in 300,000 births. Osteoclastic equilibrium in these patients is altered, where bone apposition continues without normally balanced resorption. Patients present with short stature, skeletal malformations, predispositions to fractures, haematopoiesis dysfunctions, and cranial nerve compressions. They are also at risk of developing osteomyelitis of the jaws. Allogenic bone marrow transplantations have been proposed as a viable treatment option.
Clinical management: The oral findings of 2 sisters are presented. Only the older sibling had undergone bone marrow transplantation. Similarities in clinical features observed included delayed

tooth eruption and exfoliation, hypodontia, enamel hypoplasia, and tendency for caries and periodontal disease. Radiographic examination revealed bone sclerosis leading to failure of eruption, impacted and ankylosed teeth; periodontal membrane defects; abnormal tooth shape and missing molar roots; atypical pulp morphology; and disturbances in enamel mineralization. Carious teeth were restored and options for replacement of missing teeth were explored. Strong preventive regimes and frequent recalls were implemented.

Conclusion: The development of the dentition is severely disturbed in both siblings with osteopetrosis. Despite having undergone bone marrow transplantation, the older sibling did not appear to present with osteoclastic normalization. Oral presentations of the older sibling did not markedly differ from that of the younger sibling. There was no 'catch up' dental development, therefore normal tooth eruption and development was not reestablished. Management is aimed at preservation of existing dentition and rehabilitation of function.

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Dental management of a young girl diagnosed with neurofibromatosis

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Introduction: Neurofibromatosis or von Recklinghausen disease is an autosomal dominant condition, first described by von Recklinghausen in 1882. It is a rare condition occurring once every 2000 or 3300 births. The disease is a gene mutation, where the anomalies affect mesoderm and neuroectoderm. It commonly presents with vast numbers of neurofibromas and patches of skin hyperpigmentation. The aim of this presentation is to present the combined therapeutic dental treatment in a patient diagnosed with neurofibromatosis. A 15 year-old Caucasian girl, diagnosed with neurofibromatosis presented at the graduate clinic of the paediatric dentistry department. Extra-oral examination revealed no evident skin lesions, while intraoral and radiographic examination revealed multiple carious lesions, partial anodontia, retention of primary teeth and taurodontism.

Clinical management: The dental management was performed in two phases and with the cooperation of the prosthodontic department. The first stage involved restorative care of all carious lesions and the second phase involved the prosthetic rehabilitation of the missing teeth. Most of the primary and permanent teeth, regardless of their mobility, were retained and a partial upper denture was constructed. The treatment was satisfactory and in the 6 and 12-month post-operative follow-ups the patient reported no further problems or symptoms. The treatment performed to date is provisional until growth is completed.

Conclusion: This was an intermediate phase in order to temporarily improve aesthetics and restore function, in terms of occlusion. Once growth is completed a more interfering treatment plan will be plausible.

