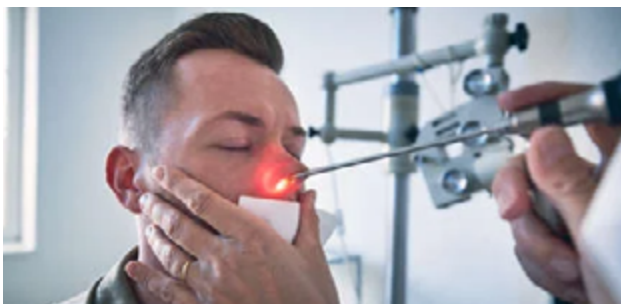
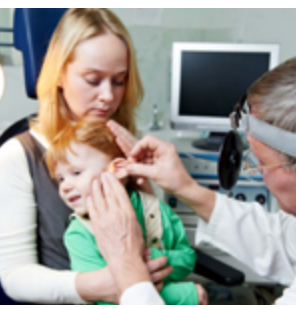
Keynote Forum
May 15, 2023

ENT 2023

10th International Conference on

Otolarngology: ENT Surgery

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Chiara De Luca

University of L'Aquila, Italy

Craniosynostosis is a feature of CHD7-related CHARGE Syndrome

CHARGE syndrome is a rare genetic disorder characterized by the presence of multiple malformations involving the craniofacial district, in particular eyes, ears, heart, and genitourinary tract in association with delayed growth and development. Its phenotypic spectrum is highly variable. The syndrome is caused in most cases by heterozygous variants in CHD7 and, more rarely, SEMA3E. Craniofacial alterations involving eyes, ears, nose, and mouth as well as the upper airways are typical of this condition, however, to date, craniosynostosis is not considered part of the clinical spectrum. We report a newborn affected by CHARGE syndrome confirmed by the identification of the de novo heterozygous c.6157C>T, p(Arg2053*) CHD7 variant, presenting with bi-coronal craniosynostosis and found two additional subjects in the literature with different craniosynostoses and distinct CHD7 alterations. We suggest the inclusion of CHD7-related CHARGE syndrome within the group of rare causes of syndromic craniosynostoses.

Recent Publication

1. Fortugno P, Monetta R, Cinquina V, Rigon C, Boaretto F, De Luca C, Zoppi N, Di Leandro L, De Domenico E, Di Daniele A, Ippoliti R, Angelucci F, Di Cesare E, De Paulis R, Salviati L, Colombi M, Brancati F, Ritelli M. Truncating variants in the penultimate exon of TGFBR1 escaping nonsense-mediated mRNA decay cause Loeys-Dietz syndrome. Eur J Hum Genet. 2023 Jan 4. doi: 10.1038/s41431-022-01279-4. Epub ahead of print. PMID: 36599937.

2. Maria Asif, Emrah Kaygusuz, Marwan Shinawi, Anna Nickelsen, Tzung-Chien Hsieh, Prerana Wagle, Birgit Budde, Jennifer Hochscherf, Uzma Abdullah, Stefan Höning, Christian Nienberg, Dirk Lindenblatt, Angelika A. Noegel, Janine Altmüller, Holger Thiele, Susanne Motameny, Nicole Fleischer, Idan Segal, Lynn Pais, Sigrid Tinschert, Nadra G. Samra, Juliann M. Savatt, Natasha L. Rudy, Chiara De Luca, Paola Fortugno, Susan M. White, Peter Krawitz, Anna C.E. Hurst, Karsten Niefind, Joachim Jose, Francesco Brancati, Peter Nürnberg, Muhammad Sajid Hussain. De novo variants of CSNK2B cause a new intellectual disability-craniodigital syndrome by disrupting the canonical Wnt signaling pathway. Human Genetics and Genomics Advances, 2022, 100111, ISSN 2666-2477, doi.org/10.1016/j.xhgg.2022.100111.

3. Fortugno P, Monetta R, Belli M, Botti E, Angelucci F, Palmerini MG, Annarita NS, De Luca C, Ceccarini M, Salvatore M, Bianchi L, Macioce P, Teson M, Ricci F, Network IUD, Macchiarelli G, Didona B, Costanzo A, Castiglia D, Brancati F. RIPK4 regulates cell-cell adhesion in epidermal development and homeostasis. Hum Mol Genet. 2022 Feb 26:ddac046. doi: 10.1093/hmg/ddac046. Epub ahead of pri`nt. PMID: 35220430.

Biography

Chiara De Luca is a Medical Geneticist currently attending her last year's Ph.D. course at the University of L'Aquila (Italy) on the genetic diagnosis of rare diseases. She has worked as a senior assistant at the Department of Human Genetics at KU Leuven, Belgium for six months. She works on the definition of the genetic bases of human genetic disorders, and she has 11 publications in the field.

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Jann Louie Tan

Vicente Sotto Memorial Medical Center, Philippines

A rare case of Tessier number 7 unilateral cleft: Macrostomia, left with accessory tragus, bilateral presenting in a nine-year-old Filipino female patient: A case report

Tessier Number 7 Cleft also known as Transverse Facial Cleft or Congenital Macrostomia is a rare congenital anomaly with an incidence of 1 in 60,000 births to 1 in 300,000 live births. It is caused by the migration failure of the mesodermal process after the fusing of the embryonic grooves between the maxillary and mandibular prominences of the first branchial arch during the fourth and fifth week of the embryonic life. Presentation of this condition varies from a slight widening of oral commissure to a cleft extending to the tragus of the ear and hemifacial microsomia. Macrostomia presents both aesthetic and functional problems, such as oral incompetence and difficulties in facial expression, mastication, and speech. These are the factors that should be considered during reconstruction. However, psychosocial factor is also an important aspect in dealing with these types of defects. Surgery is the treatment of choice, wherein the goal is the proper repair of the orbitalis oris muscle for restoration of oral function, forming a symmetric and aesthetically pleasing oral commissure with a minimally visible scar.

In this report, we present a 9-year-old female with intermittent drooling due to a facial deformity. She was diagnosed with Congenital Macrostomia and Bilateral Accessory Tragus and eventually underwent repair via Vermillion Return Flap Technique. There are multiple types of repairs for this condition, however, other techniques tend to have visible scars due to the pattern and a sharp oral commissure. In this case, the choice of this technique is mainly to avoid the complications mentioned above. Post-operatively, the patient had a good oral sphincter function, facial and lip movements were preserved, was able to grin/smile, and was aesthetically pleasing to both the scar and the oral commissure. It was also reported that the patient had renewed self-confidence, especially in her school performance.

Biography

Jann Louie Tan has completed his medical school at the University of the Visayas – Gullas College of Medicine took his Post Graduate Internship at Chong Hua Hospital in Cebu and is currently a Senior ORL-HNS Resident in Vicente Sotto Memorial Medical Center in Cebu, Philippines. He was able to present this case during the 11th International Conference of Pan Asia Academy of Facial Plastic and Reconstructive Surgery in Singapore last February 22, 2023.

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Daniel Erick T Amparado

Vicente Sotto Memorial Medical Center, Philippines

The use of a deltopectoral flap in an entire lower lip defect: A case report

The majority of the malignant neoplasms (>90%) of the oral cavity are squamous cell carcinoma, which predominates in the lower lip. With surgery being the mainstay treatment; The goals for resection and reconstruction of a lip carcinoma include adequate cosmesis and function, such as maintaining oral competence. There are various reconstructive options for lip carcinomas and they are dependent on the size of the lesion. Options range from direct closure of smaller lesions to commonly using flaps for larger defects. Local flaps, such as the Bernard-Weber's procedure have been commonly used for lesions spanning more than 2/3rds of the lip length. With large lesions approaching the full span of the lower lip, there has been a challenge in meeting the desired functional and cosmetic goals for reconstruction. Regional pedicled flaps have been used commonly in head and neck cancers to improve safe and functional outcomes. There has been a great success in using the Deltopectoral Flap in reconstructing defects of the lower thirds of the face and oral cavity. A medially based deltopectoral flap is preferable in head and neck skin defects requiring reconstruction because it is less bulky, produces less donor site impairments and improved blood supply compared with random flaps. Also, the color and texture of the skin paddle are very suitable for facial reconstruction. Another advantage is that a large area of well-vascularized skin is readily available and can be used without delay in most cases.

In the report, we present a case of a 63-year-old man who presented with an entire lower lip squamous cell carcinoma. He underwent Wide excision coverage of the defect via Deltopectoral flap, right with a satisfactory outcome. There was noted restoration of oral competence and aesthetic outcome.

Biography

Daniel Erick T. Amparado got his medical degree from the Cebu Institute of Medicine and took his Post Graduate Internship at Perpetual Succour Hospital, where he was awarded the most outstanding Post Graduate Intern. He is currently the Chief Resident in the Department of ORL-HNS at Vicente Sotto Memorial Medical Center. He was able to present this case during the Academy of Head and Neck Oncology Virtual Interhospital Grand Rounds last November 19, 2022.

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