Three-Dimensional Analysis of Rare Facial Clefts and Clinical Facial Appearance

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For everyone born with some type of orofacial cleft.

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PREFACE

The Hospital for Rehabilitation of Craniofacial Anomalies, University of São Paulo (HRCA/USP) is recognized by the World Health Organization as a "world reference" in craniofacial anomalies care (Freitas et al. 2012). Over 55 years have been dedicated to the multidisciplinary care of individuals with orofacial clefts, including cleft lip and palate, cleft palate, rare facial clefts, craniosynostosis, craniofacial midline anomalies with ocular hypotelorism and hypertelorism, anomalies of the first and second branchial arches, hearing impairment and other complex craniofacial anomalies. Rare facial clefts, also called Tessier clefts, may be isolated or associated with other craniofacial, skeletal or systemic abnormalities. It is not uncommon to observe a combination of multiple rare facial clefts in the same individual, classified as complex cases of rare facial clefts. Most of these cases require a long and costly rehabilitation process, which impacts the affected individual's social, neuropsychological and academic development. This book aims to contribute to the different specialities involved in rare facial cleft care by presenting a three-dimensional representation of craniofacial clefts of 0 to 14 and 30 in the Tessier classification using the presentation of clinical cases of individuals followed up at HRCA/USP. The description of anatomical variations illustrated by photographic images and three-dimensional images from computed tomography reconstruction allows the reader to understand better the craniofacial structures involved in each type of cleft and their phenotypic variations and associations with other rare facial clefts. Visualizing a relationship between tissue changes and bone changes is also possible. We, the authors, hope that a better knowledge of the morphology of rare facial clefts will allow the establishment of personalized and individualized therapeutic practices in high-complexity cases. Thus, recognising repetitive patterns can assist in a more accurate syndromic diagnosis. It will also assist the rehabilitation team in predicting the risks and benefits of therapeutic procedures.

—Freitas, José Alberto de Souza, Lucimara Teixeira das Neves, Ana Lúcia Pompéia Fraga de Almeida, Daniela Gamba Garib, Ivy Kiemle Trindade-Suedam, Renato Yassutaka Faria Yaedú, Rita de Cássia Moura Carvalho Lauris, Simone Soares, Thais Marchini Oliveira and João Henrique Nogueira Pinto. 2012. "Rehabilitative Treatment of Cleft Lip and Palate: Experience of the Hospital for Rehabilitation of

Craniofacial Anomalies/USP (HRAC/USP) - Part 1: Overall Aspects." *Journal of Applied Oral Science* 20 (1): 9–15.

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Affectionately, we thank all the patients and their family members who contributed to the preparation of this work by allowing the use of their images and examinations.

We would like to express our special and sincere thanks to all the employees and professionals of the Hospital for Rehabilitation of Craniofacial Anomalies, University of São Paulo (HRCA/USP), for their collaboration in clinical care and carrying out research projects. We also appreciate the collaboration of our friend Pricila Copedê Frascareli.

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INTRODUCTORY NOTE

Craniofacial clefts are uncommon congenital anomalies that affect facial bones and soft tissue structures, resulting in anatomical deficiencies (Tessier, 1976, David, Moore and Cooter, 1989). The exact frequency of rare craniofacial clefts is unknown. The estimated incidences of rare orofacial clefts have been reported as 1.43 to 4.85 per 100,000 live births (Kawamoto, 1976, Fijałkowska and Antoszewski, 2015).

Patients presenting some type of craniofacial cleft frequently need an extensive follow-up by a multidisciplinary team and different treatment protocols according to the type and amplitude of the cleft. The first author that recognized the three-dimensional complexity of cleft was Paul Tessier (1976), who established that a cleft in the soft tissue corresponds to a cleft in the bone structure. The Tessier classification (1976) consists of a numerical classification of the anatomical structures, considering the location of the cleft, taking the sagittal midline as a reference, both for soft and bone tissue. The orbit is the primary reference structure, and 15 regions are defined counter-clockwise. The clefts affecting the middle and upper craniofacial region are numbered from 0 to 14, and the mandibular cleft (lower face region) is 30 (Figure 1). Another classification was proposed for craniofacial clefts (van der Meulen, 1983). However, the Tessier classification is the most used. This study aims to morphologically evaluate the craniofacial structures involved in individuals with isolated or syndromic rare craniofacial clefts; to evaluate the association of bone and soft tissue clefts; and to contribute to the diagnosis, management and understanding of the pathogenetic mechanisms of these types of clefts. The computed tomography images of every patient were digitized and processed using the Mimics 17.0 software (Mimics, Materialize, Belgium). A craniofacial surgeon assisted the team in evaluating the computed tomography scans, and the orthodontists supervised the segmentation of craniofacial structures and the evaluation of craniofacial growth. This project was approved by the Research Ethics Committee (CEP-HRAC number 2.146.750/2017). The participants or legal guardians authorized all images and files from patients' photographs and computed tomography.

References

David, D. J., M. H. Moore, and R. D. Cooter. 1989. “Tessier Clefts Revisited with a Third Dimension.” Cleft Palate J 26 (3): 184–85.

Fijałkowska, M., and B. Antoszewski. 2015. “Rare Facial Clefts.” Pol Przegl Chir 87 (8): 389–94. https://doi.org/ 10.1515/pjs-2015-0077

Kawamoto, H. K. 1976. “The Kaleidoscopic World of Rare Craniofacial Clefts: Order out of Chaos (Tessier Classification).” Clin Plast Surg 3 (4): 529–72.

Meulen, J. C. van der, R. Mazzola, C. Vermey-Keers, M. Strieker, and B. Raphael. 1983. “A Morphogenetic Classification of Craniofacial Malformations.” Plastic and Reconstructive Surgery 71 (4): 560–72. https://doi.org/10.1097/00006534-198304000-00022.

Tessier, Paul. 1976. “Anatomical Classification of Facial, Cranio-Facial and Latero-Facial Clefts.” *Journal of Maxillofacial Surgery* 4: 69–92. https://doi.org/10.1016/s0301-0503(76)80013-6.

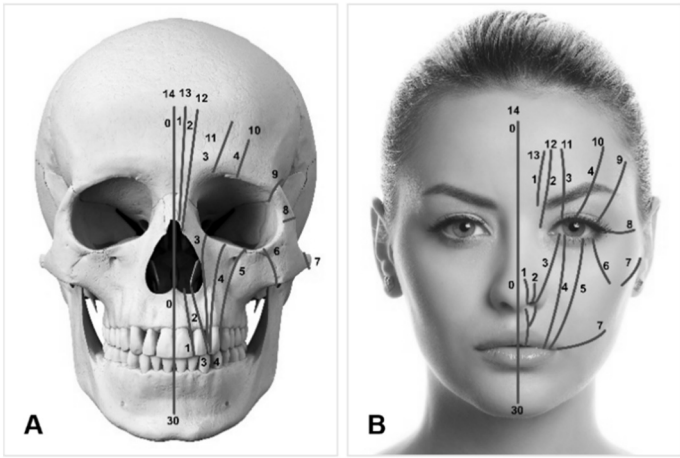


Figure 1 – Classification of the craniofacial clefts according to Tessier (1976). (A) Bone clefts numbered. (B) Soft tissue clefts numbered. Original artwork by the authors.

CHAPTER ONE

TESSIER CLEFT NUMBER 0: THE MEDIAN CLEFT LIP

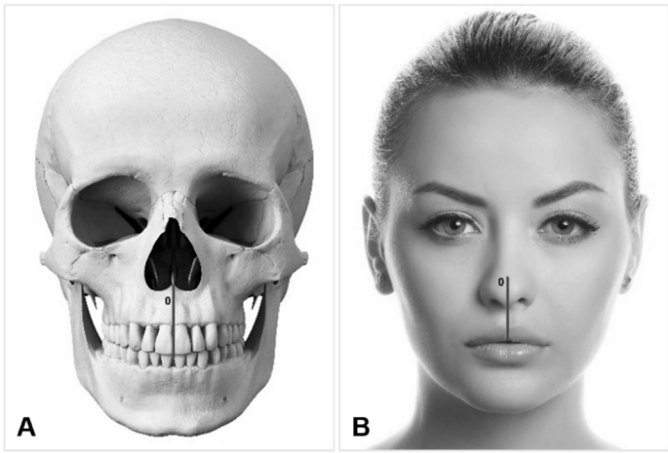


Figure 2 – Localization of Tessier cleft number 0. (A) Bone clefts numbered. (B) Soft tissue clefts numbered. Original artwork by the authors.

Tessier cleft number 0, also known as median cleft, is caused by the lack of fusion of the frontonasal and medial nasal processes. It can result in premaxilla agenesis (Figure 2). Cleft number 0 involves structures of the facial midline showing wide phenotypic variability and microforms. In some cases, the premaxilla may be partially affected. The cleft may present as a small notch in the upper lip, vermilion, or the alveolar arch (between the maxillary central incisors), with a bifid nose or a midline groove along the nasal dorsum. Tessier cleft number 0 can be associated with other midline anomalies, including hypotelorism or hypertelorism, holoprosencephaly, corpus callosum agenesis and frontonasal encephalocele. These combinations can be part of a known condition, such as holoprosencephaly (OMIM 236100), frontonasal dysplasia (OMIM 136760), orofacial digital syndrome (OMIM 311200), Pai syndrome

(155145), Sakoda complex (midline cleft, ethmoidal encephalocele, ocular anomaly, corpus callosum agenesis and cleft palate) (OMIM 610871), among others.

References

Baek, Seung-Hak, Yoon-Hee Park, Jee Hyeok Chung, Sukwha Kim, and Jin-Young Choi. 2018. "Orthodontic and Orthopedic Treatment for a Growing Patient with Tessier Number 0 Cleft." *Korean Journal of Orthodontics* 48 (2): 113–24. https://doi.org/10.4041/kjod.2018.48.2.113.

Cohen Jr MM. *Perspectives on the face*. Oxford University Press: New York; 2006

David, D. J., M. H. Moore, and R. D. Cooter. 1989. "Tessier Clefts Revisited with a Third Dimension." *Cleft Palate J* 26 (3): 184–85.

De Boutray, M., J. L. Beziat, J. Yachouh, M. Bigorre, A. Gleizal, and G. Captier. 2016. "Median Cleft of the Upper Lip: A New Classification to Guide Treatment Decisions." *J Craniomaxillofac Surg* 44 (6): 664–71.

Evans, Kelly N., Anne V. Hing, and Michael L. Cunningham. 2018. "Craniofacial Malformations." In *Avery's Diseases of the Newborn*, 1417-1437.e2. Elsevier.

Guruprasad, Y., and D. S. Chauhan. 2014. "Midline Nasal Dermoid Cyst with Tessier's 0 Cleft." *J Nat Sci Biol Med* 5 (2): 479–82.

Kolker, A. R., A. M. Sailon, J. G. Meara, and A. D. Holmes. 2015. "Midline Cleft Lip and Bifid Nose Deformity: Description, Classification and Treatment." *J Craniofac Surg* 26 (8): 2304–8.

Online Mendelian Inheritance in Man, OMIM®. McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University (Baltimore, MD). Accessed November 10, 2022. https://omim.org/.

Tessier, Paul. 1976. "Anatomical Classification of Facial, Cranio-Facial and Latero-Facial Clefts." *Journal of Maxillofacial Surgery* 4: 69–92. https://doi.org/10.1016/s0301-0503(76)80013-6.

Tessier cleft number 0

Case 1

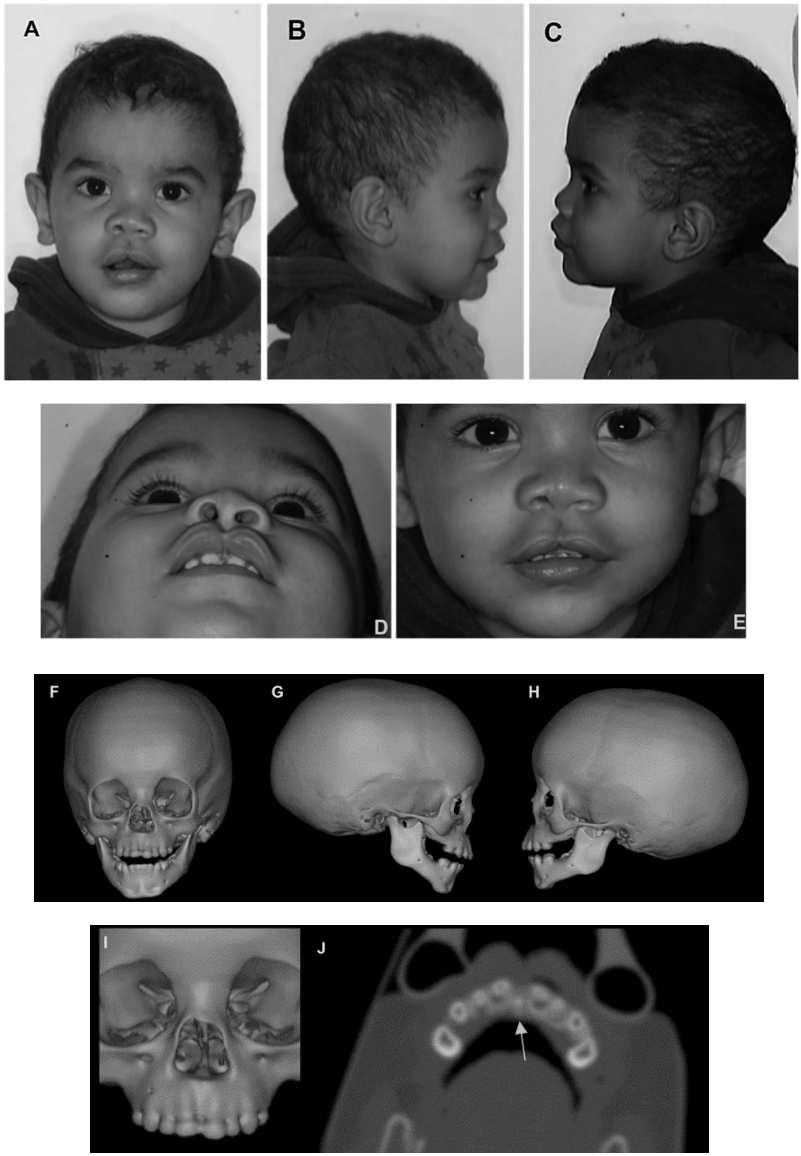
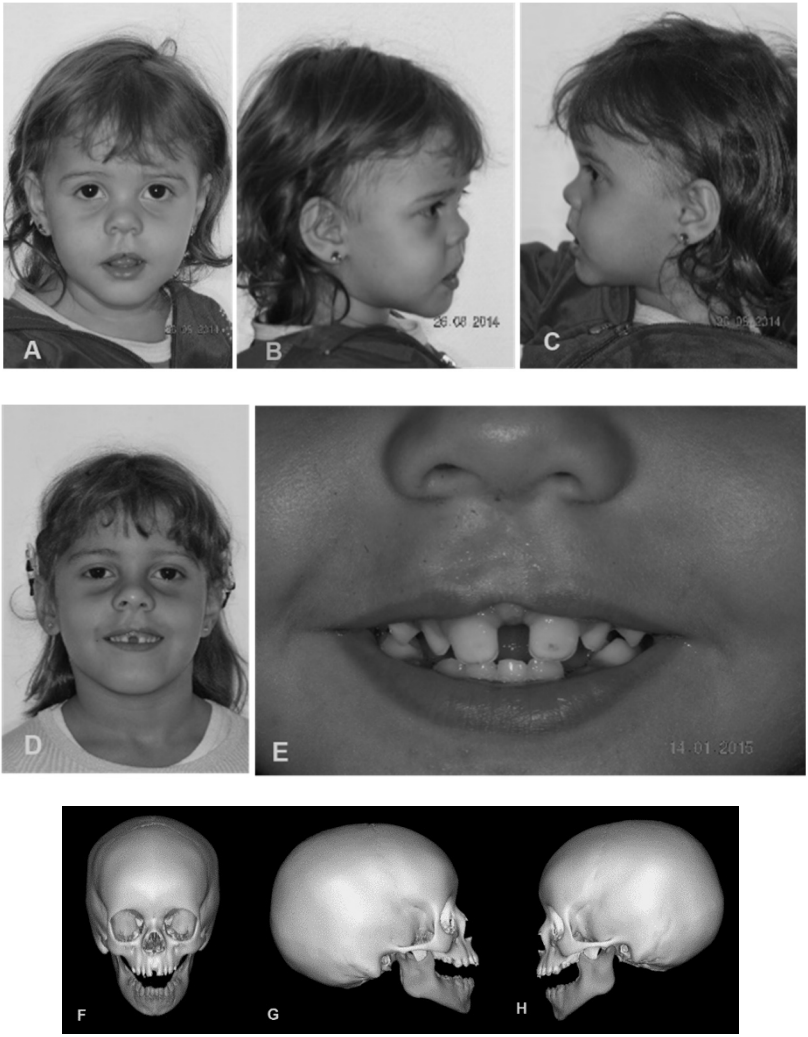


Figure 3 (previous page) – Clinical phenotype of a male patient with Tessier cleft number 0. A-E: Facial photographs at 12 months old. Note a mild midline cleft lip with separated vermillion and midline groove that extends until the philtrum. A wide nasal base with an asymmetrical nostril and a mild Tessier cleft number one (right side) on the nasal dorsum was presented with a furrow in the soft tissue. F-I: computed tomography 3D-reconstructions obtained at age 12 months (frontal, lateral and oblique view). I: observe a close mild alveolar cleft between the maxillary central incisors with a mild notch in the alveolar ridge. An asymmetrical nasal cavity with a mild notch in the left nasal bone was observed. J: computed tomography axial section showing a mild notch in the middle region of the maxillary alveolar ridge.

Case 2



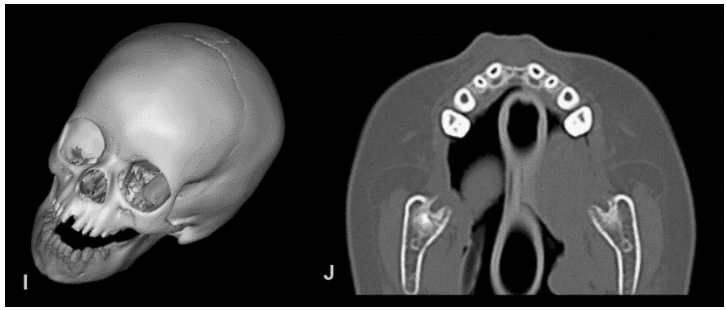
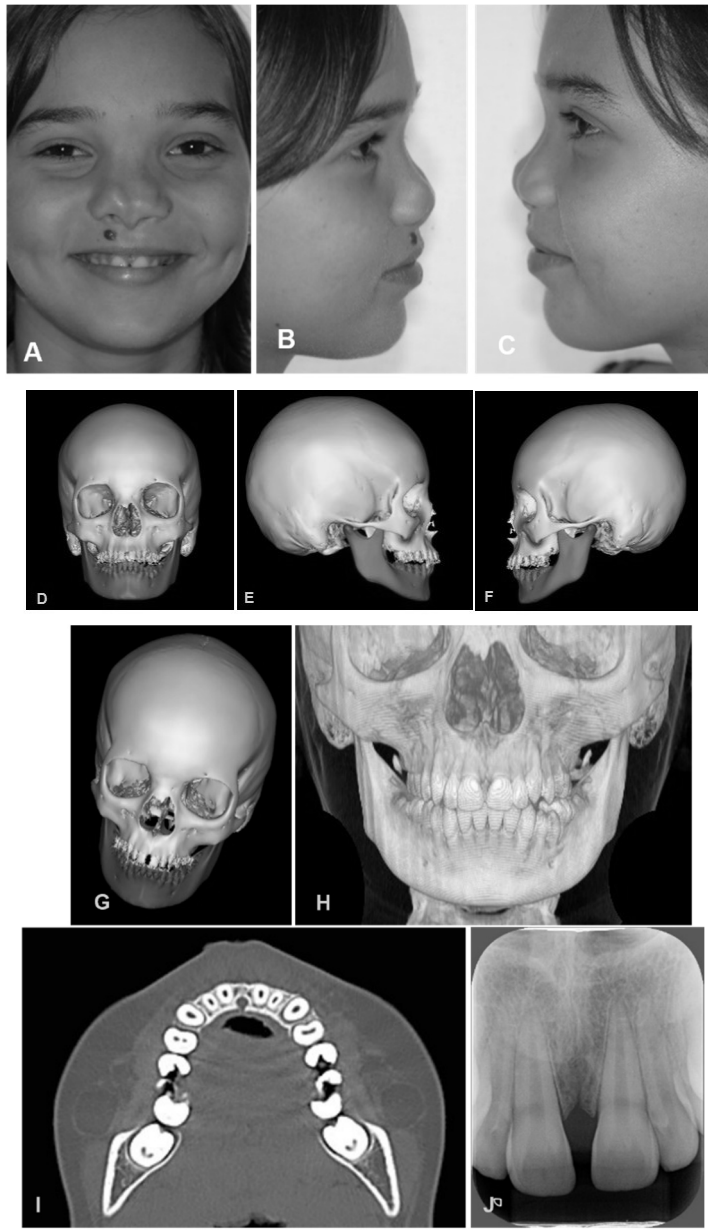


Figure 4 – Clinical phenotype of a female patient with Tessier cleft number 0. A-C: at one year and four months old. Observe the mild cleft at the centre of the upper lip extending as a wide groove in the philtrum. A wide nasal base and low nasal bridge were present. D-E: facial phenotype at age five showing a midline diastema between maxillary central incisors. F-I: computed tomography 3D-reconstructions obtained at two years old showing a mild midline groove in the premaxilla. J: computed tomography axial section showing a widened interdental bone septum between the maxillary central incisors.

Case 3



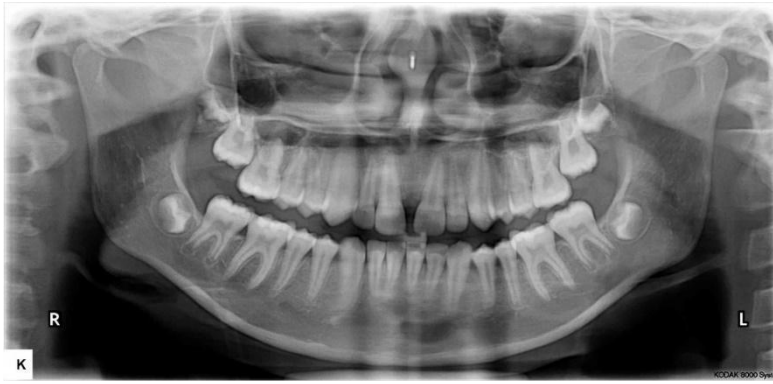
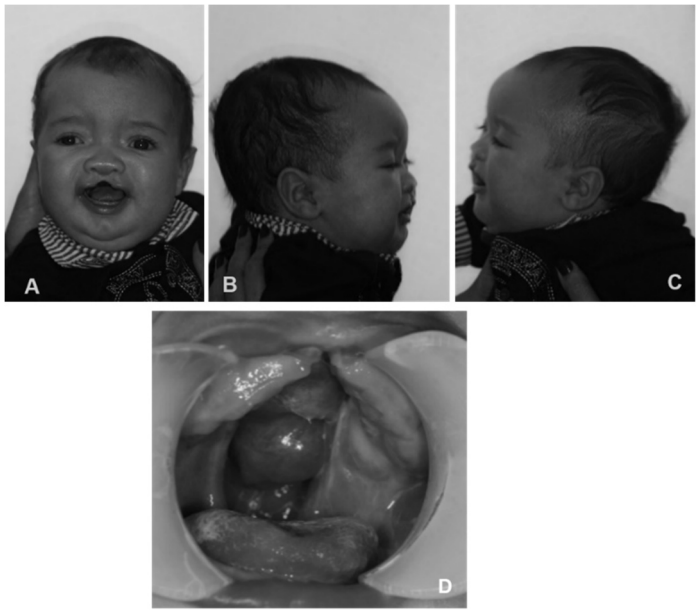
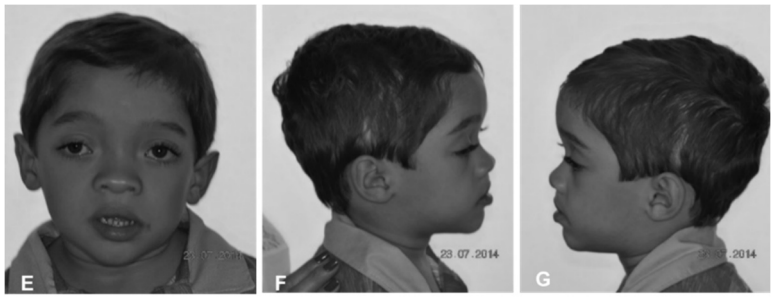
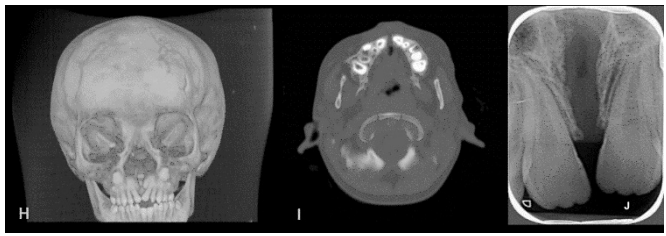


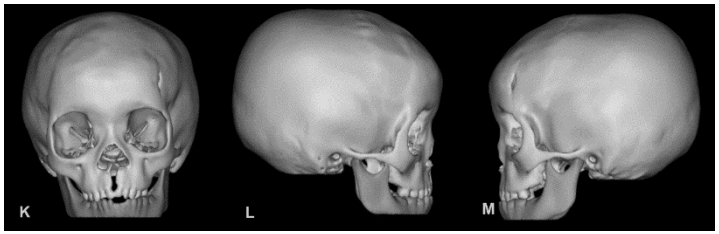
Figure 5 – Clinical phenotype of a ten-year-old female patient with Tessier cleft number 0. A-C: note the short nose with a wide and depressed nasal base, a large columella, a short philtrum and a nasal tip bifidity. D-G: computed tomography three-dimensional reconstruction obtained at ten years old. Observe a short and abnormal nasal bone and a midline diastema in the maxillary alveolar ridge. H-I: observe a small notch in the alveolar ridge between the central incisors. J: periapical radiography showing a V-shape midline bone defect in the alveolar ridge between the upper central incisors. K: panoramic radiograph at 11 years old showing alveolar defect between upper central incisors and absence of dental anomalies.

Case 4





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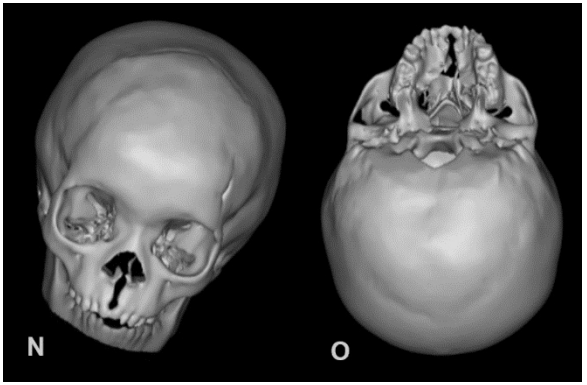
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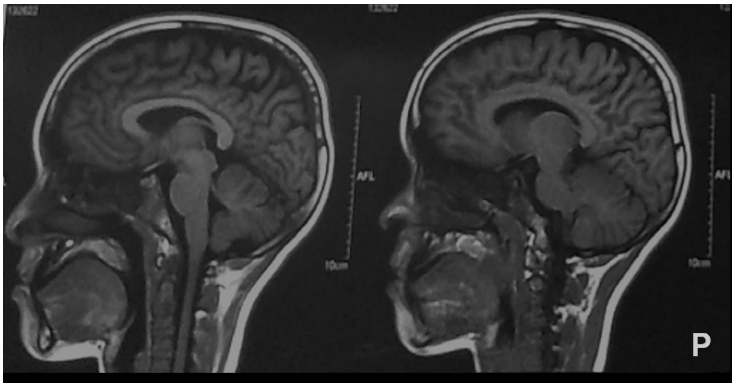
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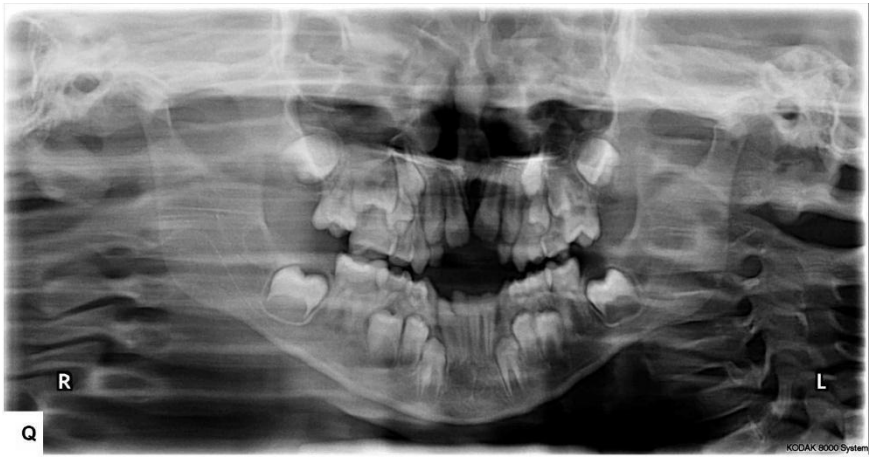
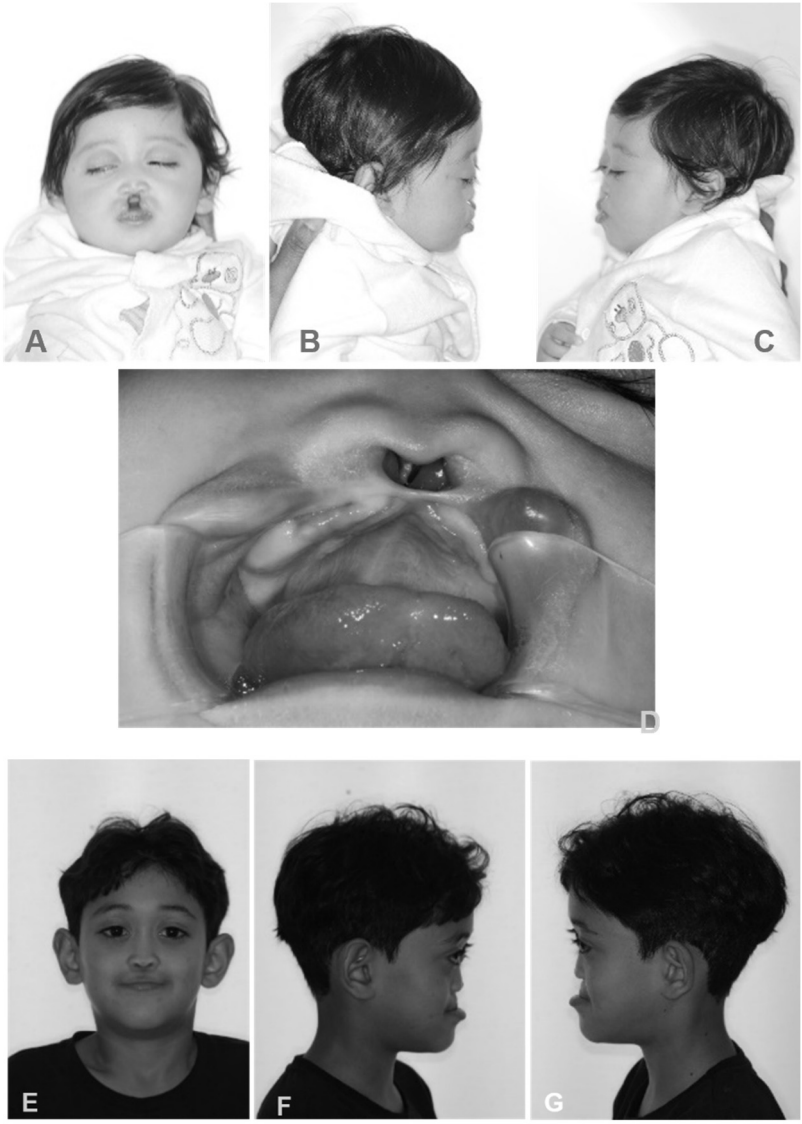


Figure 6 – Clinical phenotype of a male patient with Sakoda complex (OMIM 610871). A-D: at three months old, a short nose with a wide and depressed nasal base, large columella and midline cleft lip extending through vermillion and philtrum were observed. A midline cleft palate and basal cephalocele were present in the oral cavity. E-G: postoperative follow-up at five years old. Note the orbital hypertelorism and the wide nasal bridge. H: translucent computed tomography 3D reconstruction showing a maxillary midline cleft extending until the nasal cavity. I: computed tomography axial section showing a bone defect between the maxillary central incisors. J: periapical radiography showing a wide midline defect in the alveolar ridge between the maxillary central incisors. K-O: computed tomography 3D images obtained at five years old.

The midline cleft lip and palate were present. Observe the flat nasal bone, a vomer duplication, nasal septum deviation to the left, and (O) orthogonal occlusal view of the hard palate. P: magnetic resonance imaging at 12 years old showing brain structures. Q: panoramic radiograph at seven years old shows the alveolar defect between the upper central incisors and an absence of dental anomalies.

Case 5



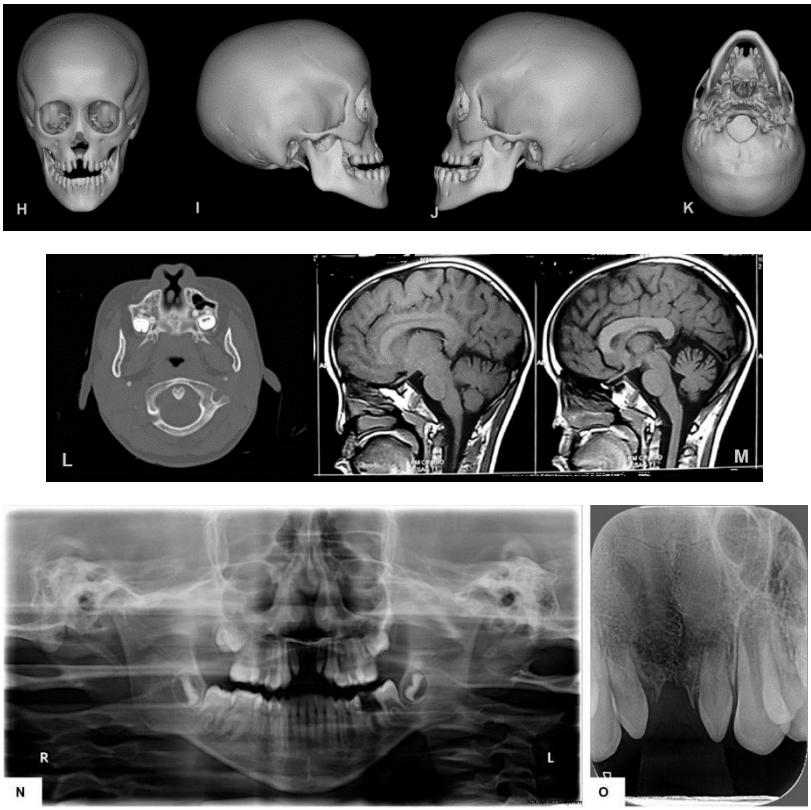


Figure 7 – Clinical phenotype of a male patient with holoprosencephaly-like. The facial phenotype is similar to holoprosencephaly except for the absence of an abnormal central nervous system. A-D: at three months old, note an ocular hypotelorism and a flat face with midface hypoplasia, depressed nasal base, absence of the nasal bridge, unique nasal cavity, absence of the columella and philtrum, a wide median cleft lip, and premaxilla agenesis with a V-shape defect in the central region of the maxillary alveolar arch. E-G: postoperative follow-up at nine years old. Observe the orbital hypertelorism and midface hypoplasia with a flat nasal bridge. H-J: computed tomography 3D-reconstructions obtained at nine years old. Note the lateral displacement of the nasal processes of the maxilla, the absence of the vomer and anterior nasal spine, and the absence of premaxilla with a wide midline cleft. Small lateral incisors were present in the mesial region of the maxillary processes. K: bottom view of CT 3D

reconstruction showing the median cleft restricted to the alveolar arch with a normal hard palate. L: computed tomography axial section showing the absence of the nasal septum. M: magnetic resonance imaging at nine years old shows midface hypoplasia, premaxilla agenesis and a normal central nervous system. N: panoramic radiograph at nine years old showing central maxillary defect with the upper central incisors agenesis. O: periapical radiography showing the maxillary defect and bilateral conoid tooth (lateral incisors maxillary).