proceedings

MDPI

Extended Abstract

Odontostomatological Findings in Heimler Syndrome: A Case Report †

Antonio Romano [1], Maria Rosaria Barillari [2], Carlo Lajolo [3], Fedora della Vella [4], Giuseppe Costa [2], Alberta Lucchese [1], Rosario Serpico [1], Francesca Simonelli [1] and Maria Contaldo [1,*]

[1] Multidisciplinary Department of Medical-Surgical and Odontostomatological Specialties, University of Campania "L. Vanvitelli", 80138 Naples, Italy; antonio.romano4@unicampania.it (A.R.); alberta.lucchese@unicampania.it (A.L.); rosario.serpico@unicampania.it (R.S.); francesca.simonelli@unicampania.it (F.S.)

[2] Department of Mental and Physical Health and Preventive Medicine, University of L. Vanvitelli, 80138 Naples, Italy; mariarosaria.barillari@unicampania.it (M.R.B.); giuseppe.costa@unicampania.it (G.C.)

[3] Head and Neck Department, Fondazione Policlinico Universitario A. Gemelli-IRCCS, School of Dentistry, Università Cattolica del Sacro Cuore, 00168 Rome, Italy; clajolo@gmail.com

[4] Interdisciplinary Department of Medicine, University "Aldo Moro" of Bari, 70121 Bari, Italy; dellavellaf@gmail.com

* Correspondence: maria.contaldo@gmail.com or maria.contaldo@unicampania.it; Tel.: +39-3204876058

† Presented at the XV National and III International Congress of the Italian Society of Oral Pathology and Medicine (SIPMO), Bari, Italy, 17–19 October 2019.

Published: 12 December 2019

Heimler syndrome (HS) is rare autosomal-recessive disorder caused by mutations of peroxin genes, *PEX1* and *PEX6*. Defects in peroxin genes alter peroxisome assembly and its metabolic pathways, essential for the metabolism of fatty acids, ether lipids, polyamines and amino acids, thus supporting the peroxisome biogenesis disorders (PBDs): a variety of severe conditions, among which HS is the mildest form. Heimler et al. first described HS in 1991 [1], and, since then, scientific literature has reported less than 12 families and less than 20 cases. HS is generally characterized by pre-lingual hearing loss, nail abnormalities, ocular involvements and dental anomalies [2]. Here we report a case of a 9-year-old female, whose genetic analyses revealed to be affected by HS and who underwent multi-disciplinary examinations to define her complete clinical features. She referred at the Eye clinic of the University of Campania "L. Vanvitelli" for ophthalmological evaluations. At the beginning, the presence of sensorineural hearing loss and early onset atypical Retinitis Pigmentosa, oriented toward the clinical diagnosis of Usher syndrome (US), another autosomal-recessive disorder, typically characterized by partial/total hearing loss and worsening vision loss. Hence, molecular genetic tests were performed to refine the diagnosis. After sequencing more than 2000 genes, it results two pathogenic variants of PEX1, thus excluding US and orienting toward HS. At this point, a multi-disciplinary team approached the patient to identify the further HS features. In addition to ophthalmological and audiological findings, clinical and instrumental phoniatric and neuropsychological examinations revealed deficit in language skills and moderate intellectual disability; dermatologists confirmed the presence of leukonychia. Odontostomatological evaluation was performed by two oral pathologists expert in pediatric dentistry. After excluding facial disharmonies at the extraoral examination, the intraoral one revealed a mixed dentition, as expecting according to age [3]. The upper central incisors displayed white spots on the vestibular surfaces and the first permanent molars cusps were yellowish and hypoplastic. The X-ray orthopantomography revealed no agenesis and a pale reduction of the enamel density of canines and premolars, whose cusps appeared slightly hypoplastic. The patient reported mouth breathing, probably responsible for the anterior open bite and the high-arched palate. Oral mucosae were healthy and normochromic. Tongue and teeth were covered by a visible layer of dental plaque, which has been removed by

professional oral hygiene. To date the patient is under multidisciplinary follow-up for her various affections, included the dental ones. It is notably the unanimity of the literature in reporting dental defects in HS. Which differs among the reports is the ascription of these defects under the diction of "amelogenesis imperfecta" given by some authors, despite the lack of genetic tests supporting defective genes involved in amelogenesis. Hence, we retain the condition here presented closer to the molar incisor hypomineralization (MIH), whose causes are still not clearly defined [4].

References

1. Heimler, A.; Fox, J.E.; Hershey, J.E.; Crespi, P. Sensorineural hearing loss, enamel hypoplasia, and nail abnormalities in sibs. *Am. J. Med. Genet*. **1991**, *39*, 192–195.
2. Contaldo, M.; Di Stasio, D.; Santoro, R.; Laino, L.; Perillo, L.; Petruzzi, M.; Lauritano, D.; Serpico, R.; Lucchese, A. Non-invasive in vivo visualization of enamel defects by reflectance confocal microscopy (RCM). *Odontology* **2015**, *103*, 177–184, doi:10.1007/s10266-014-0155-4.
3. Gentile, E.; Di Stasio, D.; Santoro, R.; Contaldo, M.; Salerno, C.; Serpico, R.; Lucchese, A. In vivo microstructural analysis of enamel in permanent and deciduous teeth. *Ultrastruct. Pathol.* **2015**, *39*, 131–134, doi:10.3109/01913123.2014.960544.
4. Contaldo, M.; Serpico, R.; Lucchese, A. In vivo imaging of enamel by reflectance confocal microscopy (RCM): Non-invasive analysis of dental surface. *Odontology* **2014**, *102*, 325–329, doi:10.1007/s10266-013-0110-9.