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”, 80138 Naples, Italy; mariarosaria.barillari@unicampania.it (M.R.B.); giuseppe.costa@unicampania.it (G.C.)
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


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