Faculté Internationale de Médecine Dentaire
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AMELOGENESIS IMPERFECTA ASSOCIATED TO HEIMLER SYNDROME

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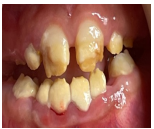
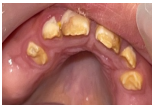
INTRODUCTION: Heimler syndrome (HS) is a rare disease of genetic origin. This syndrome was first described in 1991 by Heimler et al as the association of sensorineural hearing loss (SNHI), progressive macular dystrophy and IA without any cerebral or hepatic dysfunction. The prognosis is primarily auditory, since the severity of the deafness will limit the reception of speech and hinder the appropriation of the oral language, causing social repercussions. This initial disability will be aggravated by the gradual appearance of a second disability affecting sight and hindering sensory “compensation”.

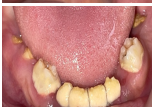
Case Presentation

SNHI

RETINAL DYSTROPHY







AMELOGENESIS IMPERFECTA

Discussion

The management of deafness is based on hearing aid devices and speech therapy. Retinal dystrophy (RD) most often occurs a little later and is variable depending on the patient. The description of the visual phenotype seems to range from cone-rod retinal dystrophy to macular dystrophy, including isolated cone dystrophy. Amelogenesis imperfecta (AI) is one of the most specific clinical signs of HS.

It most often affects the permanent secondary dentition, the posterior teeth (premolars and molars) being more severely affected than the anterior teeth (incisors). Its diagnosis and management require the intervention of a specialized dentist. The diagnosis of Heimler syndrome is confirmed by a genetic analysis prescribed by the clinical geneticist in view of the association of sensorineural hearing loss with retinal dystrophy. Pathogenic biallelic variations of PEX1 or PEX6 are responsible for almost 100% of HS cases but also for other forms of peroxisome biogenesis disorders (PBD).

Conclusion

The first clinical sign is congenital or early onset sensorineural hearing loss (before 3 years). This is most often bilateral congenital and detected in the maternity ward. The diagnosis and management of hearing disorders in young children must be carried out in a referral center for pediatric ENT. Diagnosis is based on age-appropriate objective and subjective audiometric testing.