Denouement and Discussion

Fanconi Anemia

Photographs from an intraoral examination show the uneven brown discoloration of the tongue (Figure 1) and brown gums with smooth surface texture and generalized inflammation (Figure 2); these alterations are related to melanin deposition and poor oral hygiene, respectively, and are consistent with Fanconi anemia.[1] Saleh and Stephen[2] describe a generalized black hyperpigmentation on the oral mucosa of a patient with Fanconi anemia. Other oral manifestations include periodontitis, supernumerary teeth, and congenital anodontia. The augmented predisposition to periodontal disease in patients with Fanconi anemia may be due not only to pancytopenia and defective detoxification of oxygen radicals, which are typical of the disorder itself, but also to medications administered during immunosuppressive treatment.[3]

A generalized tan pigmentation of the skin, micrognathia, microcephaly, the absence of the fifth finger,[4] growth retardation, and a congenital hip dislocation that required surgical treatment at age 1 year are all important features associated with this congenital disease.[5] Hematologic analysis revealed macrocytic anemia, and pancytopenia due to bone marrow failure was confirmed by biopsy. A positive diepoxybutane test result demonstrated an increment on the chromosome-breaking rate in lymphocytes. Fanconi anemia cells have an abnormal cell cycle, with an increased frequency of cells arrested at the G2 phase. Using flow cytometry for diepoxybutane test analysis, we determined that 53.5% of cells stopped at the G2 phase.[6] In addition to these characteristics, delayed eruption of permanent teeth was identified on a panoramic dental radiographic scan (Figure 3). The patient was successfully treated: her maxillary lateral, central incisors and her mandibular first left and right primary molars were extracted. A prosthetic removable appliance was placed on the upper anterior segment and lingual arch for mandibular space maintenance. The clinical findings and paraclinical analysis were consistent with Fanconi anemia.

Fanconi anemia is a rare autosomal recessive disorder, characterized by physical abnormalities, congenital malformation of the skeleton, bone marrow failure, and increased risk of malignancy. This malady affects both men and women, and members of all ethnic groups; however, it is more frequent in Ashkenazi Jews and the Afrikaans population of South Africa. Its frequency has been estimated at 1 in 350 000 births. The genes responsible for all of 15 Fanconi anemia complementation groups have been identified.[7] However, Kim et al[8] have reported that biallelic mutations in *SLX4* cause a new subtype of Fanconi anemia, Fanconi anemia-P.

Fanconi anemia is the most common genetic origin of aplastic anemia and one of the most usual genetic causes of hematologic malignancy. Differential diagnosis is established with other diseases (with cells derived from individuals with chromosomal breakage), such as Bloom syndrome, Nijmegen breakage syndrome, Seckel syndrome, neurofibromatosis 1, TAR syndrome (thrombocytopenia-absent radium syndrome), and non–Fanconi anemia–related VACTERL association. Although there are a few reports about oral manifestations of Fanconi anemia, other disorders can be present with it, such as Peutz-Jeghers syndrome and Laugier-Hunziker syndrome, and treatment with interferon and ribavirin is usually prescribed. Although these rare oral signs are seen in other diseases and are not pathognomonic, the hyperpigmentation of the gums and tongue is highly suggestive of Fanconi anemia.

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