PAEDIATRIC DERMATOLOGY

TWO CASES OF SHAH - WAARDENBURG IN COLOMBIA

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Background: Waardenburg syndrome is a rare association of pigmentary changes and sensorineural defects, affecting 1/32.400 - 1/50.000 newborns. Hirschsprung's disease is characterized by aganglionic megacolon, and presents in 1/5.000 newborns. Both entities belong to neurocristopathies, due to deficiencies of the neural crest cells migration in the embrionic period and can be found together as part of a syndrome known as Shah-Waardenburg Syndrome, or Waardenburg syndrome type 4 (WS4). There are only around 50 cases reported, and the probability of this association is of 4 in one million.

Observation: Two newborns presented with poliosis, extensive achromic macules, and hypochromic iris, without signs of real albinism; neurologic exam and brain CT were normal, and no complications were noted during gestation. Nonetheless, both were admitted to ER in the first week of life due to oral intolerance, greenish emesis and constipation, with subsequent diagnoses of Hirschsprung's disease (total aganglionosis: absence of ganglion cells in the submucous and intramural plexus). The first patient is currently 2 years old, and presents no other complications other than stature and weight deficit. Unfortunately the second patient got complicated after multiple surgical interventions and died at 23 days old.

Key message: The association of congenital poliosis, leukoderma, and blue iris, should raise the suspicion of syndromes like WS4, and prompt a complete clinical and imaging study as they might be the very first sign of internal involvement. Because of the risk of acute abdomen, early suspicion, and management define survival in these patients, and late complications to consider is the delay in height and weight, secondary to malnutrition that most of these patients suffer, so the multidisciplinary team must include nutritional support.