Mini review

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Developing Clinical Genetics Diagnostic Skills: Van Der Hoeve-Waardenburg Syndrome

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ABSTRACT

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There are many rare clinical syndromes and dysmorphic syndromes including genetic syndromes, and it is really difficult for many clinicians to equip themselves with adequate professional knowledge that help them to make an early diagnosis for many of the rare syndromes they may encounter [1-5]. We have previously published our pioneering huge and unique experiences with clinical genetics and dysmorphology in a plethora of publications. We have previously described a large number of rare disorders in Iraq [6-14], and we have also helped doctors in the diagnosis and publication of rare conditions observed in other countries [15]. The aim of this papers is to help practicing physicians in developing the diagnostic skills in the field of clinical syndromes by briefly reviewing a rare syndrome that have not been reported in Iraq, but it is associated with unique clinical characteristics that allow doctors who see the syndrome for the first time, capable of making an early diagnosis rather easily, by knowing few information about the syndrome. Van der Hoeve-Waardenburg syndrome is a rare genetic associated with unique clinical manifestations including a white forelock (Figure 1) or premature grayness of hair, heterochromia iridum (Figure 2) or sectoral heterochromia iridum (multiple colors in one eye), hypertelorism (Figure 2) or telecanthus which is also called dystopia canthorum (Figure 1), and congenital sensorineural hearing impairment [16,17].

Figure 1: A sketch of a patient with a white forelock in the front-centre of the head and telecanthus (Increased distance between the medial canthi, the inner corners of the eyelids associated with reduced visibility of the nasal portion of the sclera and shortening of the palpebral fissures in the horizontal direction (blepharophimosis).

Figure 2: A sketch of a patient with heterochromia iridum, differently colored eyes and hypertelorism (Increased distance between the whole eyes; lateral displacement of the eyes, eyelids, and orbits).

Figure 3: Jan van der Hoeve (April, 13, 1878-April, 26, 1952), a Dutch ophthalmologist who is best known for describing the concept of the phakomatoses, which is also called neuro-cutaneous syndromes.

Hammerschlag (1907) [18], and Urbantschitsch (1910) [19] reported the occurrence of heterochromia iridum in association with partial albinism and hearing loss, but without suggesting the possibility of a distinct clinical entity. In 1916, Jan van der Hoeve (Figure 3) reported a pair of 14 year-old monozygotic twin girls with congenital sensorineural hearing loss and dystopia canthorum [16]. In 1926, German physician Irmgard Mende reported a family of four generations from Berlin in which five children had congenital sensorineural hearing loss , dystopia canthorum (blepharophimosis which resulting from lateral displacement of the inner canthi), and leucism (Partial loss of pigmentation of the skin and hair) manifested as white hairlock or poliosis of the eyebrows [20]. Leonardi (1931) reported the association of heterochromia iridum and dystopia canthorum in a 4-year old boy (Figure 4). The mother of the boy milder condition [21]. John (1934) reported a nine year-old girl with total heterochromia, a partial sector-like heterochromia in the right eye of her younger sister, and an aunt with very light blue eyes; three members of a 3-generation family with dystopia canthorum [22]. In 1947, Swiss ophthamologist David Klein (1908-1993) reported a patient with a variant of Van der Hoeve-Waardenburg syndrome associated with bilateral deafness, pigmentation deficiencies, characteristic facial features and severe malformation of the arms [23].

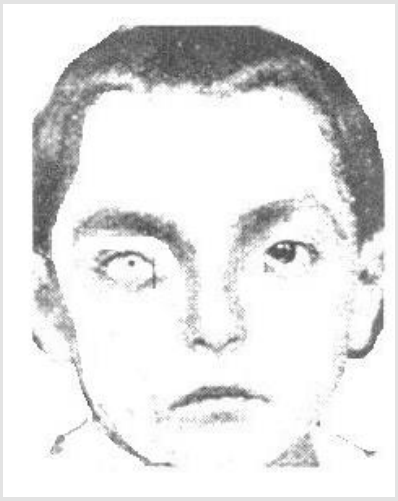

Figure 4: A sketch of the boy reported by Leonardi (1931) who had heterochromia iridum and dystopia canthorum.

Figure 5: Petrus Johannes Waardenburg (1886-1979), Dutch ophthalmologist and geneticist.

In1951, Petrus Johannes Waardenburg (Figure 5) studied 840 patients with hearing loss from five Dutch institutes for the deafness and found 12 cases of the syndrome. Waardenburg confidently emphasized the emergence of a new syndrome, and described it as including [17]:

1. Dystopia canthorum (Telecanthus)/broad, high nasal root (Hypertelorism).
2. Partial or total heterochromia iridum.
3. Congenital deafness or partial (unilateral) deafness.
4. Circumscribed albinism of the frontal head hair (white forelock).

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