

# Waardenburg syndrome in an infant of diabetic mother - A rare case report

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**Abstract:** This case has been reported because it is a rare autosomal dominant disorder of the neural crest development. A newborn male child born to a diabetic mother by full term normal vaginal delivery in our hospital presented with white forelock in midline frontal scalp, dystopia canthorum, broad nasal root and spina bifida occulta. On examination for hearing, startle response was present, no upper limb defects, cranio skeletal defects or intestinal disorders were evident. The child is diagnosed as a case of Waardenburg syndrome type 1 (WS1) fulfilling the criteria for diagnosis, however not having a typical autosomal dominant presentation which may have arisen due to a mutation.

**Keywords:** Waardenburg Syndrome; White Forelock; Dystopia Canthorum; Spina Bifida Occulta.

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## 1. INTRODUCTION

Waardenburg syndrome (neurocrestopathy) is a genetic disorder usually inherited as an autosomal dominant trait with incomplete penetrance and variable expressivity that alters neural crest development. The syndrome was first described by the Dutch ophthalmologist, P.J. Waardenburg in 1951, who noted the appearance of six distinctive features involving anatomical and pigmentary anomalies of eye, skin and hair as well as deafness across the family members.<sup>1</sup>

It includes displaced canthi, heterochromia of the irides, white forelock, broad nasal root, and deafness.<sup>2</sup> There are four types on the basis of array of clinical features.

## 2. CASE REPORT

We present a newborn male child born to a diabetic mother by full term normal vaginal delivery, product of non-consanguineous marriage. On examination we found that the infant had white forelock in midline frontal scalp, dystopia canthorum, broad nasal root and spina bifida occulta. Family history was unremarkable. No hypopigmented patches were noted. Ophthalmological examination demonstrated dystopia canthorum, however there was no evidence of iris heterochromia. Neurological examination revealed spina bifida occulta confirmed by ultrasound. On examination for hearing, startle response was present, suggesting normal hearing. No upper limb defects, cranio skeletal defects or intestinal disorders were evident. In our case two major and one associated feature was present viz. white forelock, dystopia canthorum and broad nasal root, hence supporting the diagnosis of Waardenburg syndrome type 1.



### **3. DISCUSSION**

The diagnostic criteria have been established by an international WS consortium .

#### **Major criteria**

- Hearing loss  
Sensorineural deafness
- Iris pigmentary anomalies  
Heterochromia irides or intense blue irides, hypopigmented ocular fundus.
- Hair hypopigmentation  
White forelock-poliosis or premature graying, hypopigmented eyebrows or eyelashes
- Dystopia canthorum
- 1<sup>st</sup> degree relative affected

#### **Minor criteria**

- Hypopigmented skin lesions
- Prominent nasal root
- Bushy eyebrows

#### **Associated criteria**

- Cleft/lip palate
- Spina bifida
- Musculoskeletal anomalies

Diagnosis is made by the presence of two major or one major plus two minor or one major plus a first degree relative with WS.<sup>3</sup>

WS 1 and WS 2 are autosomal dominant inherited in most cases. Mutations in PAX3 gene on chromosome 2q37 are seen in WS1 and WS 3.<sup>4,5</sup> MITF mapped on 3p12-p 14.1 are mutated in type 2 WS<sup>5</sup>. WS 4 is due to Sox 10 or endothelin-b receptor (EDNRB) gene mutations.<sup>6</sup>

Waardenburg syndrome also manifests at birth as localized areas of depigmented skin and hair. There are 4 types of Waardenburg syndrome.

The hallmark of WS 1 is the white forelock, which is seen in 20-60% of patients only. Some patients have areas of depigmented skin. Deafness occurs in 9-37%, heterochromia irides in 20 % and unibrow (synophrys) in 17-69% of those affected. Dystopia Canthorum i.e. telecanthus is seen in all patients of WS 1. WS 2 lacks dystopia canthorum and have higher incidence of deafness. In WS 3 patients have limb abnormalities. WS type 4 is associated with Hirshprung's disease.<sup>7</sup>

Our patient fulfills the diagnostic criteria of WS 1. However, in this case, history of similar illness or findings suggestive of Waardenburg syndrome was not present in the other family members. Different features of WS 1 in this neonate might have resulted from a new mutation. The diagnosis of WS is essentially clinical and not every case expresses all clinical manifestations of the complete WS.

#### **4. CONCLUSION**

Facial abnormalities and a white forelock are prominent features difficult to overlook during a routine neonatal examination. A careful medical history in patients with suspected Waardenburg syndrome is important for classification and identify this rare condition as well as its systemic complications associated with each subtype.

#### **REFERENCES**

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