Iranian J Publ Health, Vol. 37, No.4, 2008, pp.131-137

Case Report

First Report of New Oral Findings in a Case with Noonan Syndrome

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(Received 2 Jul 2008; accepted 13 Dec 2008)

Abstract

Noonan syndrome is an autosomal dominant disorder that is typically evident at birth. In many affected individuals, this syndrome is associated with cardiac defects and a distinctive facial appearance. The high frequency of cardiac disorder, ophthalmic, growth and orthopedic signs, associated with Noonan syndrome emphasizes the need for early diagnosis. This report aimed to present a 19 year old Iranian girl suffering from Noonan syndrome. In this case in addition to typical signs and symptoms reported for Noonan syndrome earlier, there are three other significant signs which have not been reported yet in any other cases. So they are supposed to be related to Noonan syndrome.

Key words: *Noonan syndrome, Taurodontism, Prominent rugae, Missing canine, Iran*

Introduction

Noonan syndrome was described in 1963 as a multisystem disorder characterized by short stature and low-set ears (1). Noonan syndrome is one of the most common autosomal dominant disorders seen in children with congenital heart disease (2-5).The disorder may be characterized by a wide spectrum of symptoms and physical features that vary greatly in range and severity. Affected individuals have distinctive facial features including hypertelorism, ptosis, flat nasal bridge, open bite, high arched palate, short neck, and often with chest, breast and spine deformity.

Noonan syndrome is difficult to be diagnosed by facial appearance alone. Until recently the diagnosis rested solely on clinical features. A mutation in the PTPN 11 gene has been reported in about 50% of individuals with Noonan syndrome (6). There are a number of syndromes which may be difficult to distinguish from Noonan syndrome such as Turner syndrome, Leopard syndrome and Shprintzen- Goldberg syndrome.

Noonan syndrome is relatively common with an estimated incidence of between 1 per 1000 and 1 per 2500 live birth (7). Many cases have been reported; however, none of them described the characteristic oral features of bilateral taurodontism, missing canine and prominent rugae.

Turner syndrome

A congenital heart defect occurs in approximately 30% of patients with Turner syndrome (TS) (8, 9). Hypertension is common in TS, even in the absence of cardiac or renal malformation. Between 10-30% of individuals with TS develop hypothyroidism generally associated with antithyroid antibodies. Hearing loss is common in girls with TS often with speech problems (10). Strabismus, amblyopia and ptosis are common in TS (11). The small and retrognathic mandible may contribute to malocclusion and other dental abnormalities. Short stature caused by growth retardation in these individuals is originated from the loss of action SHOX gene on the X chromosome (12-14).

Leopard syndrome

This rare syndrome shares many features similar to Noonan syndrome including autosomal dominant inheritance, similar facial dysmorphism and similar cardiac defects and skin manifestations reported in Noonan syndrome including pigmented nevi (15).

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Shprintzen-Goldberg syndrome

The characteristic facial appearance with more than two thirds of all individuals include having hypertelorism, down slanting palpebral fissures, a high-arched palate, developmental delay, micrognathia and apparently low-set and posteriorly rotated ears. Other common reported manifestations include hypertonia in at least the neonatal period; the most skeletal manifestations were arachnodactyle, pectus deformity, camptodactyly, scoliosis and joint hypermobility (15).

Case Report

Medical History

A 19 yr old girl was admitted to the Oral Medicine Department of Dental Faculty, Tehran University of Medical Sciences in Iran. She had not been diagnosed as having Noonan syndrome by ten and family history was negative for any other similar cases. The patient has passed high school courses; no obvious sign of mental retardation was detected. At age 6 and 12 she underwent surgical operations for spine deformity. The patient referred by chief complaint about gingival bleeding and crowding of teeth especially in lower jaw. By an overall examination, clinical features compatible with what is now called Noonan syndrome were found. Physical examination revealed a short stature (Fig. 1a), low-set rotated ears, loss of intertragic notch (Fig. 1b), short broad neck, chest deformity, lack of breast growth, low-set breast position especially in right side (Fig. 1c), Kyphoscoliosis (Fig.1d) and clinodactyly of right hand (Fig .2a,2b). Facially the patient exhibited hypertelorism, ptosis, strabismus, sporadic freckle and nevi especially on forehead (Fig.3a), protruded lower lip, small mandible, depressed chin, flat nasal bridge, tendency for exophthalmia (Fig.3b) associated with low-set hair line (Fig. 4). Based on physical examination and consultation with audiologist, the patient did not suffer from hearing loss. The result of her Echo and Electrocardiography taken by the cardiologist, showed no sign of heart disease. Complete blood count and the blood biochemistry test revealed just light hypo chrome microcytic anemia. The patient has authorized the authors for publishing her pictures in medical media.

Oral Findings

Oral examination revealed an anterior open bite, severe anterior gingival enlargement, dental crowding associated with diastema, tapered incisors (Fig.5a), narrow high-arched palate and prominent rugae (Fig. 5b). Study casts of the jaws were prepared for examination. Tapered incisors and malocclusion (Fig.6a), prominent rugae and high-arched palate (Fig.6b) were noticeable.

Radiographic Findings

On radiographic survey what we observed were a bilateral taurodontism on mandibular second molars, impaction of right maxillary canine and missing of left maxillary canine, and an idiopathic osteosclerosis between left maxillary first and second premolars area (Fig .7).

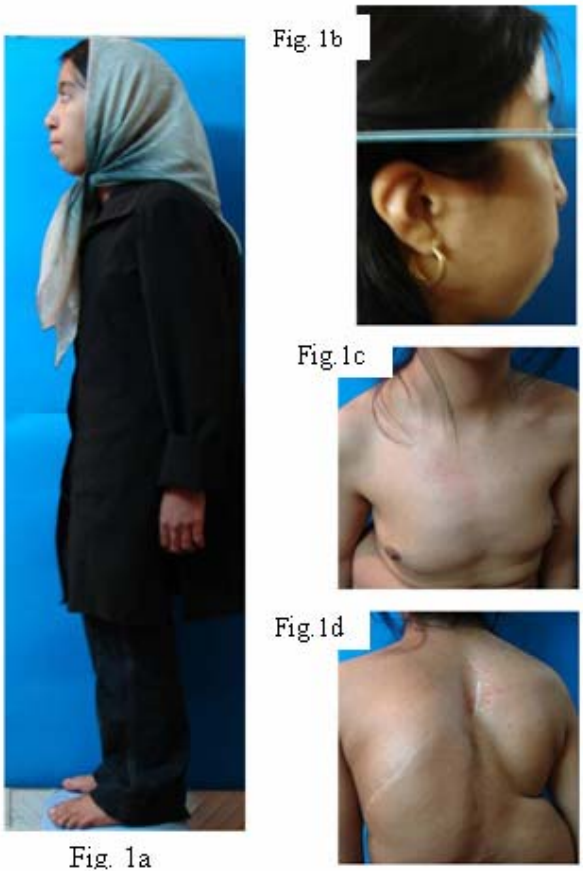

Fig. 1: Short stature (a), Low set ear (b), Lack of breast growth and Low-set breast position (c), Kyphoscoliosis (d)

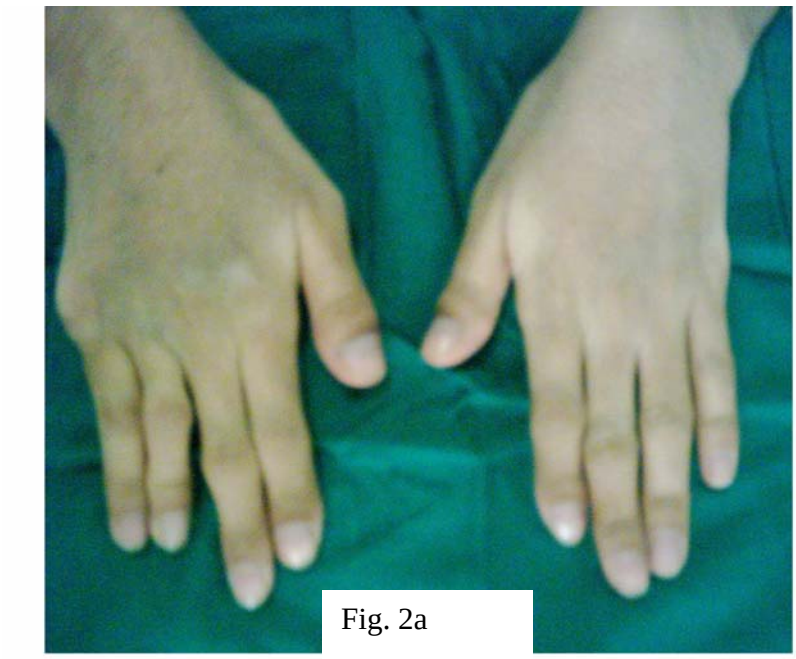

Fig. 2a

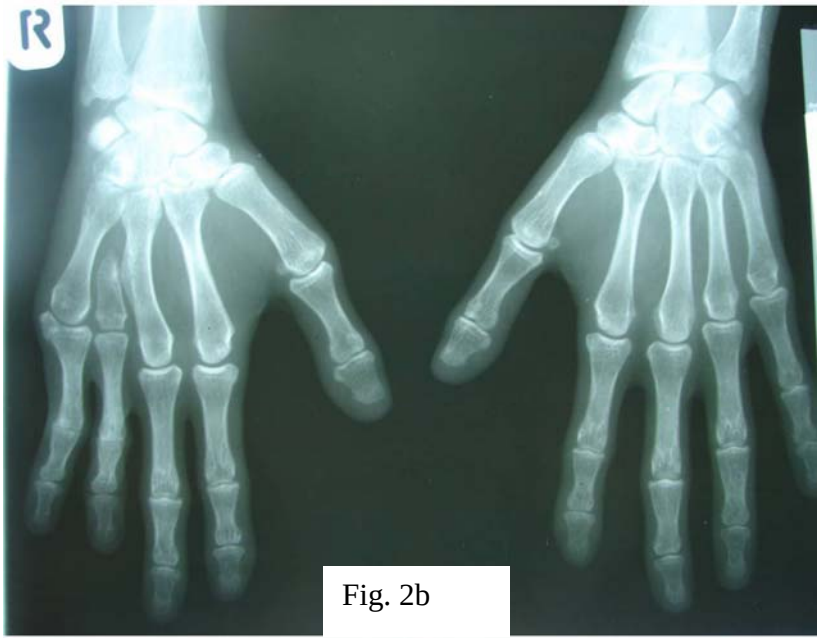

Fig. 2b

Fig .2: clinodactyly of right hand: clinical (a) and radiograph (b)
Fffffff Fig 2: Clinodactyly of right hand: Picture (a), Radiograph(b)

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Fffffff Fig. 2: Clinodactyly of right hand: Picture (a), Radiograph(b)
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Fig. 3: Sporadic freckle (a), protruded lower lip, Flat nasal bridge, Tendency for exophthalmia (b)

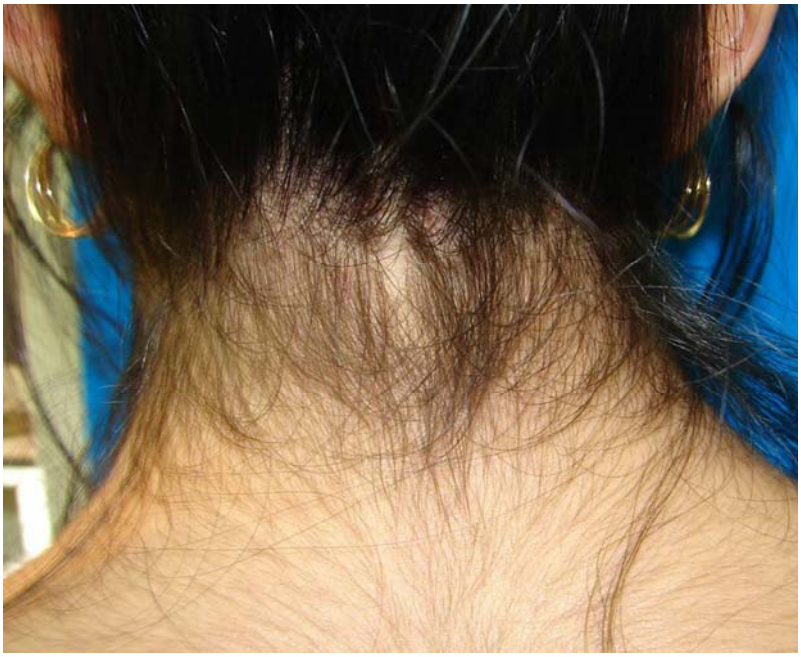

Fig. 4: Low-set hair line, Web neck

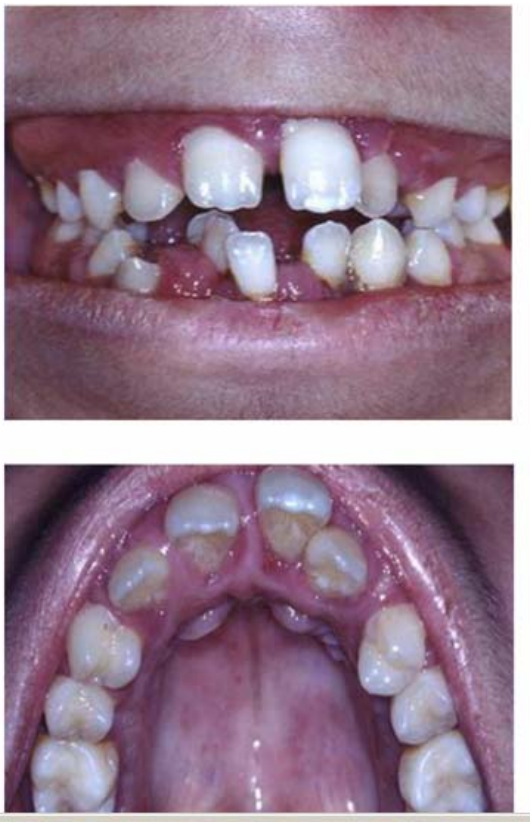

Fig .5: Anterior open bite, Teeth crowding, Gingival enlargement (a), Prominent rugae (b)

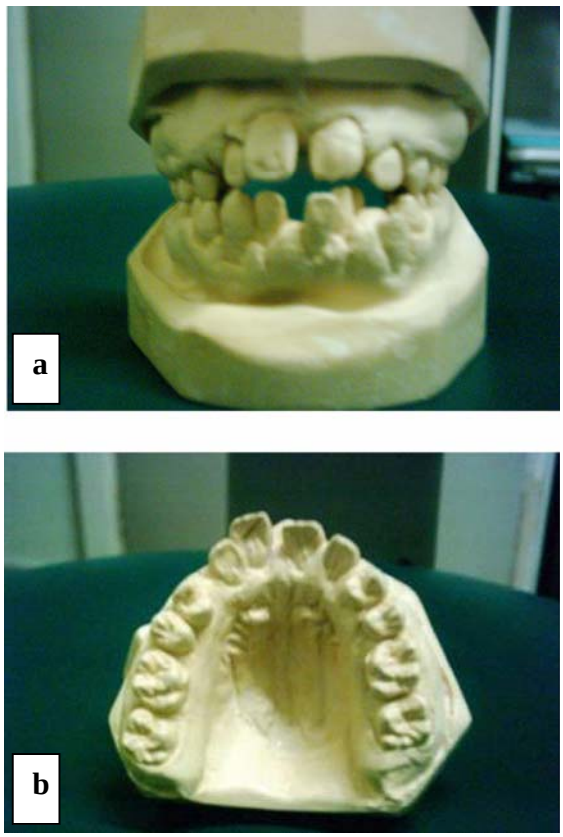

Fig. 6: Malocclusion (a), Prominent rugae (b)

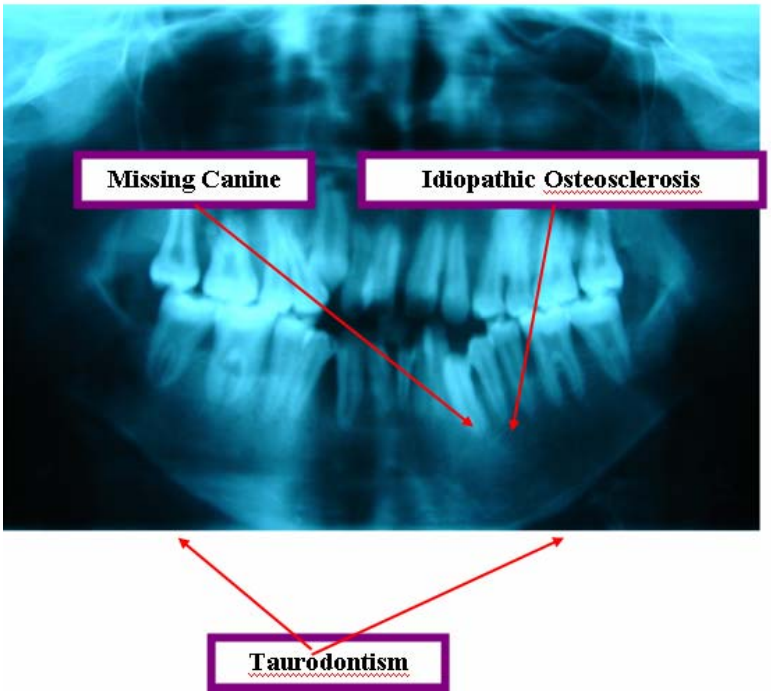

Fig .7: Radiographic view

Discussion

The high frequency of cardiac, ophthalmic, growth, orthopedic and oral manifestations associated with Noonan syndrome, emphasizes the need for early diagnosis. The present case was diagnosed as Noonan syndrome but the report describes some new oral manifestations including, bilateral taurodontism, missing canine and prominent rugae. There have been several other reports of oral findings such as anterior open bite, high palatal arch, macroglossia with this syndrome (16-17), however, none of them has reported these findings and according to the literature these abnormalities are very rare among normal cases. Therefore based on what we found in the present case we believe that they can be related to Noonan syndrome and can be considered as other suggestive oral features of Noonan syndrome. There are a number of syndromes which have similar feature to Noonan syndrome (8, 13), so it is sometimes difficult to distinguish them from each other. Turner syndrome, Shprintzen-Goldberg syndrome, and Leopard syndrome are a number of these disorders which cause difficulty in diagnosis of the disorders (Table 1).

Table 1: Comparison of signs of present case with Noonan syndrome and three comparable syndromes

Signs of disorder	present case	Noonan syndrome	Turner syndrome	Leopard syndrome	Shprintzen Goldberg syndrome
Clinical Features :					
Short stature	++	++	++	++	++
Web neck	++	++	++	+	
Growth retardation	++	++	+	++	+
Low hair line	++	+	++		
Low-set ears	++	++	+		+
Small mandible	++	++	+		+
Hypertelorism	++	++	+	++	
Strabismus	++	++	++		
Freckle and nevi	++	+		+	
Ptosis	++	++	++		
Kyphoscoliosis	++	+	++		+
Oral Findings :					
High-arched palate	++	+	+	+	+
Anterior open bite	++	++			
Crowding and spacing	++	++			
Bilateral prominent rugae	++				
Radiographic Findings:					
Taurodontism	++				
Missing of canine	++				
Idiopathic osteosclerosis	+				

Acknowledgements

The authors would like to acknowledge the collaboration of: Department of Oral Medicine, Dr Beytollahi, Dr Afshar, Dr Rohani & Mrs Amiri; Department of Removable Prosthetics, Dr Mir Mohammad Rezayie & Dr Mirfazaelian; Department of Fixed prosthetics, Dr Mahgoli, Dr Nikzad, Dr Nokar& Dr Hajmiragha; Department of Orthodontics, Dr Gerami, Dr Mojtahed Zadeh; Department of Oral Surgery, Dr Abbasi; Department of Periodontics, Dr Khorsand, Dr Kadkhoda, Dr Save. We also appreciate the kind support of personnel of Library and Audio-Visual Department, Faculty of Dentistry, Tehran University of Medical Sciences.

The authors declare that they have no conflict of interests.

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