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Clinical Genetics and Dysmorphology: Our Extraordinary Experiences

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1. Abstract

Background: We have previously described our extensive experiences with clinical genetic and dysmorphology in a plethora of publications. In a previous study, we reported 43 children with uncommon, rare and very rare genetic and hereditary disorders whom were observed during three-year period (2016-2018). In that series, very rare genetic, the case number 104 of Sanjad-Sakati-Richardson-Kirk syndrome in the world, the case number 130 of Townes Brocks syndrome and the case number 170 of Coffin Siris syndrome in the world. The aim of this paper is to describe our 2019 extraordinary new experiences with genetic disorders and dysmorphic syndromes.

Materials and methods: During the year 2019, 37 patients (25 males and 12 female) with genetic disorders and dysmorphic syndromes and were studied at the Children Teaching Hospital of Baghdad Medical City. Their ages ranged from 2 days to sixteen years.

Results: Eight patients had Down syndrome (6 boys and 2 girls), 5 patients had Cornelia De Lang syndrome (4 boys and one girl), 2 male patients had Fragile X-syndrome, 2 male patients with Prader Willi syndrome, two patients had Noonan syndrome (a boy and girl), 2 brothers had Goldberg Shprintzen syndrome. Ten patients each had Facioscapulohumeral muscular dystrophy, Virchow Seckel Syndrome, Mowat Wilson syndrome, Toriello-Carey Syndrome, Ruprecht Majewski-Bosma syndrome, congenital myotonic muscular dystrophy (Congenital dystrophia myotonica), extended Michelin tire baby, Congenital absent radii without thumb aplasia, Dandy walker syndrome and the syndrome of congenital facial palsy and unilateral anotia. In addition, six children patients had newly recognized syndromes (five boys and one girl). Many of the patients in this series were previously described or reported.

Conclusion: This one-year Iraqi pioneering experience in the fields of clinical genetics and clinical dysmorphology should herald the end of the Dark Age in these fields in Iraq.

2. Keywords: Rare; Genetic disorders; Dysmorphic syndromes; Iraq children

3. Introduction

We have previously described our extensive experiences with clinical genetic and dysmorphology in a plethora of publications [1-20]. Table 1 shows the genetic disorders we have previously observed, reported, or studied. The occurrence of oculo-cerebro-renal syndrome and nephropathic cystinosis have

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been well documented and studied in Iraqi children [4,5]. Some other genetic renal diseases have also been studied and reported [6-11].

Table 1: The genetic disorders we have previously observed, reported, or studied [20].

Commoner disorders
Thalassaemias and hemophilias [Specialized clinics and centers have been established for these disorders]. Down syndrome
Common or relatively common disorders
Achondroplasia Polycystic kidney disease Duchenne muscular dystrophy Werding Hoffman disease Gaucher disease, Mucopolysaccharidosis
Genetic renal diseases
Oculo-cerebro-renal syndrome Nephropathic cystinosis Nephronophthisis Adult polycystic kidney disease.
Polycystic disease of kidneys and liver.
Primary oxalosis.
Cystinuria.
Autosomal recessive steroid resistant syndrome.
Autosomal recessive proximal renal tubular acidosis.
Autosomal recessive distal renal tubular acidosis X-linked dominant hypophosphatemic rickets Laurence-Moon-Biedl syndrome.
Uncommon, rare and very rare genetic disorders
Wilson's disease Diastrophic dysplasia *Klinefelter syndrome* associated with renal Fanconi syndrome Coffin Siris syndrome Cutis laxa type II (Debre type) Aicardi syndrome Autosomal recessive autism.
Rare dysmorphic syndrome
Mostyn Embrey Syndrome.
New dysmorphic syndrome
A new dysmorphic associated with acrocephaly, seizures, long spindle fingers and cherry red spots.

In a previous study [21] we reported 43 patients (29 males, 14 females) with uncommon, rare and very rare genetic and hereditary disorders who were observed during three-year period (2016-2018). Their ages ranged from 5 days to 17 years. Table 2 shows the number and sex of each genetic and hereditary disorders observed in that study. In that series [20], (Table 3) very rare genetic disorders included:

The thirty-six case of cutis laxa type II (Debre type) in the world and the second case in Iraq. The case was also most probably the first case to be associated with atrial septal defect [22,23].

Table 2: The number and sex of each genetic and hereditary disorders observed [2].

Disorder	Male	Female	Total
Duchenne muscular dystrophy	5		5
Charcot Marie Tooth disease	2	2	4
Wohlfart Kugelberg Welander	3		3
Wiskott Aldrich syndrome	2		2
Hereditary (Familial) spastic paraplegia		2	**2**
Berry-Treacher Collins syndrome	1		1
Ekman-Lobstein syndrome	1		1
Morquio syndrome	1		1
Werding Hoffman disease	1		1
Prader-Labhart-Willi syndrome	1		1
Hypohidrotic ectodermal dysplasia	1		1
Lesch Nyhan syndrome	1		1
Phenylketonuria	1		1
Homocystinuria	1		1
Pediatric Huntington disease	1		1
Sanjad-Sakati-Richardson-Kirk Syndrome	1		1
Von Recklinghausen syndrome	1		1

The first case of Sanjad-Sakati-Richardson-Kirk syndrome in Iraq which was the case number 104 in the world [24,25]. The first case of Townes Brocks syndrome in Iraq which was the case number 130 in the world [26,27]. The second case of Coffin Siris syndrome in Iraq which was also the case number 170 in the world [28,29].

The aim of this paper is to report the genetic disorders and dysmorphic syndromes observed during the year 2019.

4. Materials and Methods

During the year 2019, 37 patients (25 males and 12 female) with genetic disorders and dysmorphic syndromes and were studied at the Children Teaching Hospital of Baghdad Medical City. Their ages ranged

from 2 days to sixteen years.

Table 3: The number and sex of each genetic and dysmorphic disorders observed.

Disorder	Male	Female	Total
Down syndrome (Figure 1)	6	2	8
Cornelia De Lange Syndrome (Figure 2)	4	1	5
Fragile X-syndrome (Figure 3)	2		2
Prader Willi syndrome (Figure 4)	2		2
Noonan syndrome (Figure 5)	1	1	2
Goldberg Shprintzen Syndrome (Figure 6)	2		2
Facioscapulohumeral muscular dystrophy (Figure 7)		1	1
Virchow Seckel Syndrome (Figure 8)		1	1
Mowat Wilson syndrome associated with pseudo rocker bottom feet deformity (Figure 9)	1		1
Toriello-Carey Syndrome (Figure 10)		1	1
Ruprecht Majewski-Bosma syndrome (Figure 11)		1	1
Extended Michelin tire baby syndrome associated with undescended testis, mental retardation and hearing impairment (Figure 12)	1		1
Congenital myotonic muscular dystrophy (Figure 13)		1	1
Congenital absent radii without thumb aplasia (Figure 14)		1	1
Dandy walker syndrome (Figure 15)	1		1
The syndrome of congenital facial palsy and unilateral anotia (Figure 16)		1	1
A new dysmorphic syndrome associated with reverse slanting of split eyebrows and palpebral fissures (Figure 17)	1		1

5. Results

Eight patients had Down syndrome (6 boys and 2 girls), 5 patients had Cornelia De Lang syndrome (4 boys and one girl), 2 male patients had Fragile X-syndrome, 2 male patients with Prader Willi syndrome, two patients had Noonan syndrome (a boy and girl), 2 brothers had Goldberg Shprintzen syndrome. Ten patients each had Facioscapulohumeral muscular dystrophy, Virchow Seckel Syndrome, Mowat Wilson syndrome, Toriello-Carey Syndrome, Ruprecht Majewski-Bosma syndrome, congenital myotonic muscular dystrophy (Congenital dystrophia myotonica), extended Michelin tire baby, Congenital absent radii without thumb aplasia, Dandy walker syndrome and the syndrome of congenital facial palsy and unilateral anotia. In addition, six children patients had newly recognized syndromes (five boys and one girl). Many of the patients in this series were previously described or reported [29-40].

Patients with Down syndrome (Figure 1) included two infants, a boy and a girl with atrial septal defect. Two boys with Down syndrome had alopecia areata. One of the boys with Down syndrome had mild overweight.

Figure 1: Patients with Down syndrome included two infants, a boy and a girl with atrial septal defect. Two boys with Down syndrome had alopecia areata. One of the boys with Down syndrome had mild overweight.

Figure 2: Five patients with Cornelia De Lange syndrome were observed during the year 2019.

Figure 3: Two patients with Fragile X-syndrome were observed during the year 2019.

Figure 4: One of the two patients with Prader Willi syndrome with characteristic facial features and hypogonadism.

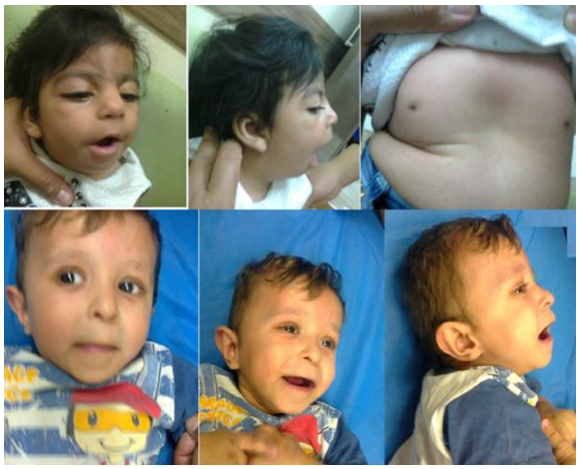

Figure 5: Two unrelated patients with Noonan syndrome were observed. The girl had widely space nipples.

A girl and a boy with Noonan syndrome were observed (Figure 5). The girl had predominantly cerebral manifestations without congenital heart defect. CT-scan showed ventriculopathy with evidence of brain atrophy. The boy had undescended testes, small atrial septal defect and closing patent ductus arteriosus.

Figure 6: Two brothers with Goldberg Shprintzen Syndrome were observed during the year 2019.

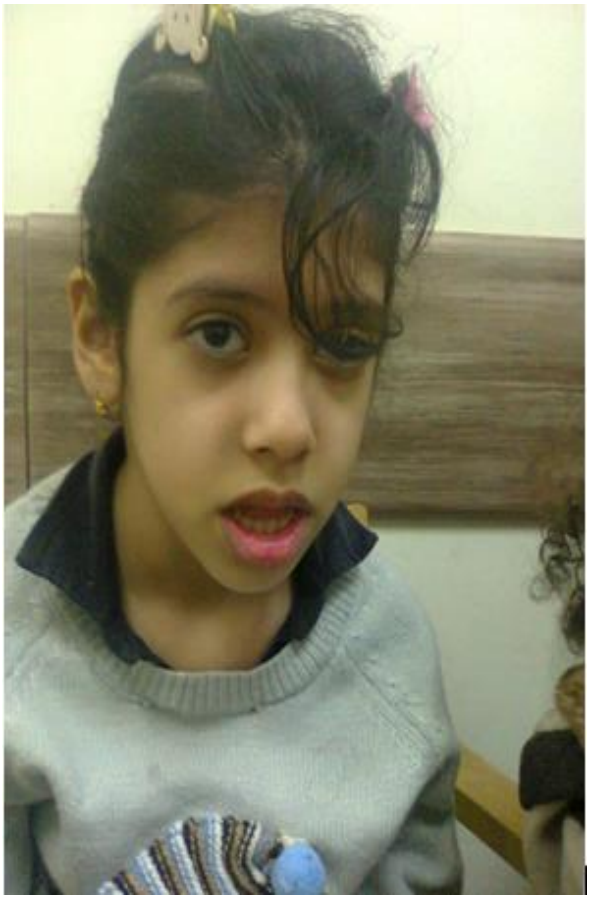

Figure 7: A girl with Facioscapulohumeral muscular dystrophy were observed during the year 2019.

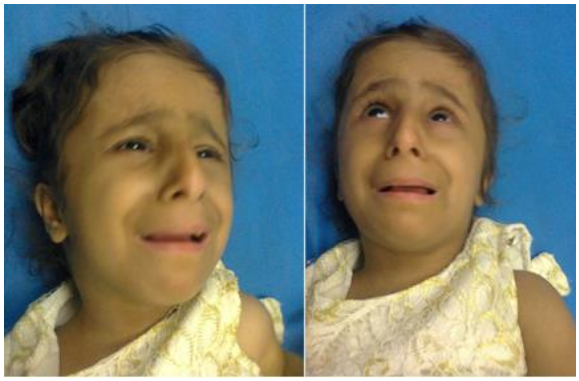

Figure 8: A girl with Virchow Seckel Syndrome.

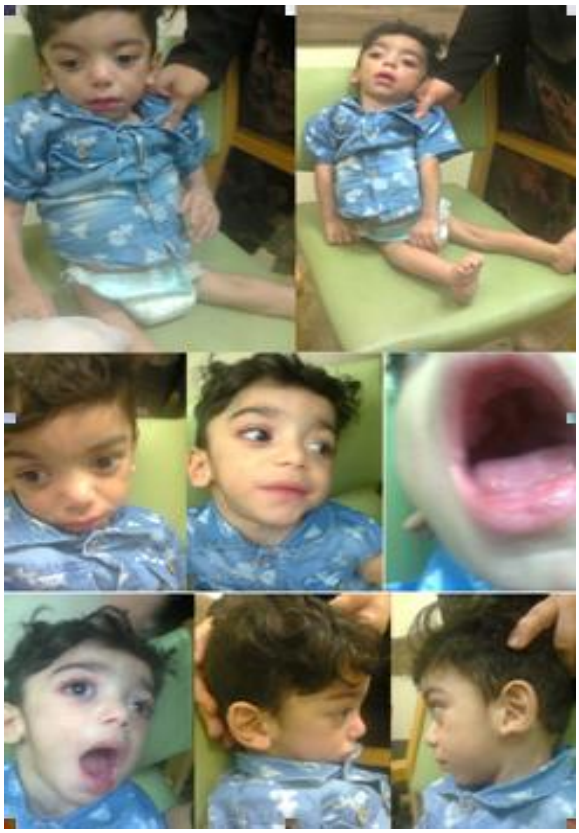

Figure 9: A boy with Mowat Wilson syndrome associated with pseudo rocker bottom feet deformity.

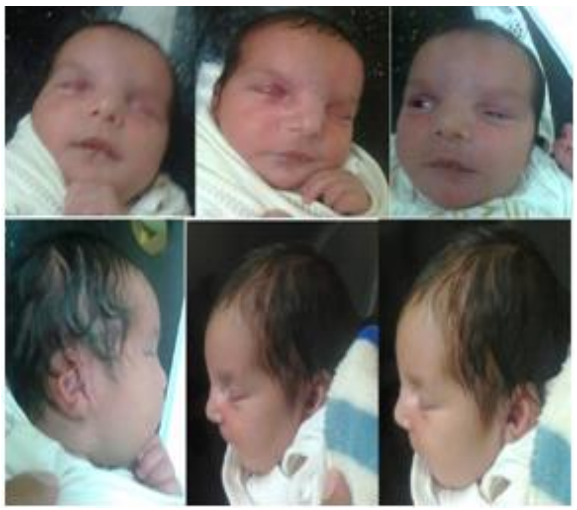

Figure 10: The girl with Toriello-Carey Syndrome.

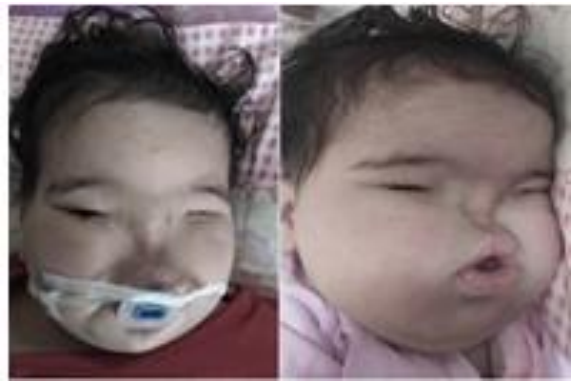

Figure 11: The girl with Ruprecht Majewski-Bosma syndrome. She had complete nasal agenesis causing difficulty in breathing requiring the use of airway piece and orogastric tube feeding. At the age of six months, it was to remove the airway piece and orogastric tube with establishment of normal breathing.

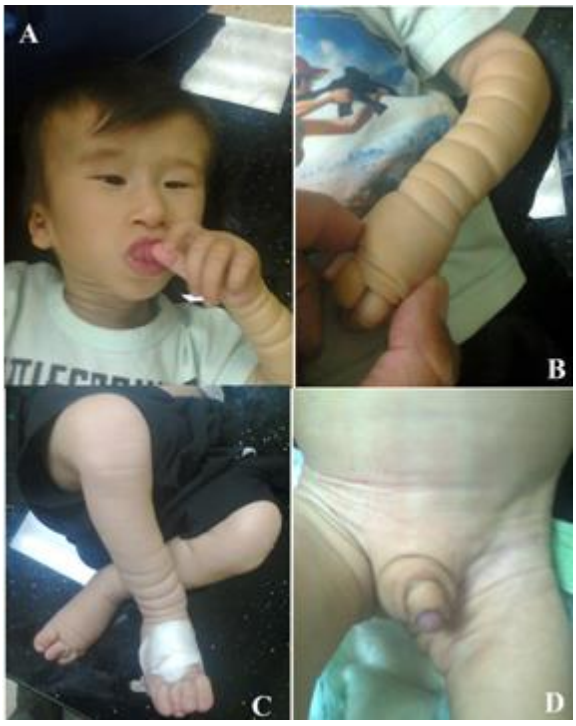

Figure 12: The case of the extended Michelin tire baby syndrome associated with significant non-cutaneous abnormalities undescended testis, mental retardation and hearing impairment.

Figure 13: The girl with congenital myotonic muscular dystrophy. At the age of four years, the girl was markedly hypotonic and was showing no spontaneous movements. She was unable to sit without support.

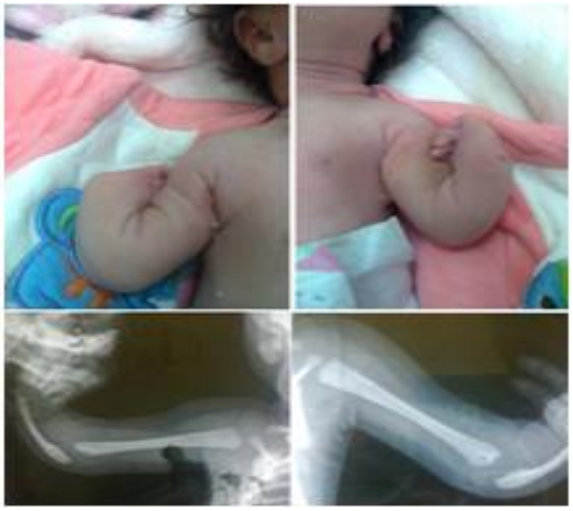

Figure 14: A two-day old female neonate with congenital absent radii without thumb aplasia.

Figure 15: A boy with Dandy walker syndrome with unusual presentation and unusual radiologic sign on CT-scan. At the age of five years, the boy was unable to sit unsupported.

Figure 16: The girl with syndrome of congenital facial palsy and unilateral anotia.

Figure 17: The boy with a new dysmorphic syndrome associated with unique eyebrows abnormalities consisting of splitting with a relatively thick upward slanting medial parts and thin non-slanting lateral parts in association with downward slanting palpebral fissures, bilateral convergent squint, hypertelorism with flat mid-face, epicanthic folds and large ears.

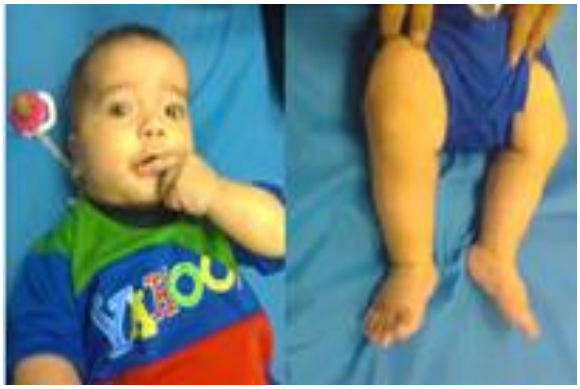

Figure 18: A boy with a new dysmorphic syndromic association consisting of congenital partial hemihypertrophy, low set ears, hypertelorism and epicanthi folds.

Figure 19: The boy with a new dysmorphic syndrome associated with psychomotor retardation, low set ears, retrognathia, facial dysmorphism (narrow palpebral and upslanting palpebral fissures and thin upper lips) and schizencephaly.

Figure 20: The boy with a new genetic syndrome associated with mental retardation, periventricular white matter hyperintensity on brain magnetic resonance imaging, retinitis pigmentosa and optic atrophy. At the age of fourteen years, the boy was able to walk, but he had psychomotor retardation with poor attention to the environment.

Figure 21: The boy with new dysmorphic syndrome associated with facial dysmorphism, mental retardation, triphalangeal toes and unilateral renal agenesis. He had distinctive facial features characterized by hypertelorism, broad nasal bridge and absent naso-frontal angle.

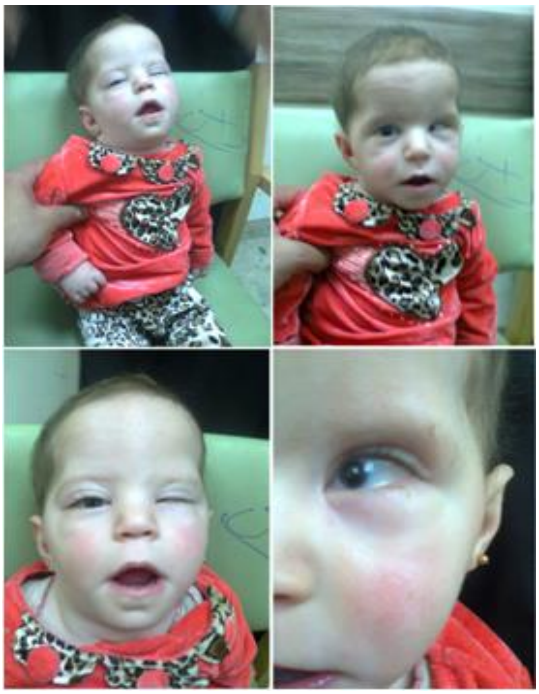

Figure 22: The girl with a new dysmorphic syndrome associated with mental-growth retardation, microphthalmia of the left eye, convergent squint of the microphthalmic eye, microcornea, iris and uvea colobomata, transient ocular hypopigmentation and contralateral optic disc colobomata and dilated third ventricle. She also had low set ears and prominent forehead. At the age of eighteen months, she was hypotonic with poor head control and was unable to sit on the chair without slipping.

6. Discussion

This one-year Iraqi pioneering experience in the fields of clinical genetics and clinical dysmorphology should herald the end of the Dark Age in these fields in Iraq. In addition to describing six new genetic and dysmorphic syndromes [41-49], this experience included describing the third case of the extended Michelin tire baby syndrome associated with undescended testis, mental retardation and hearing impairment [35], the first case of Mowat syndrome associated with deformity of the feet " Pseudo rocker bottom feet" [36], the 58th case of Toriello Carey syndrome which was associated with colpocephaly on brain MRI and CT-scan [37], the thirty fourth and thirty fifth cases of Goldberg Shprintzen syndrome [38], the case number 52 of Ruprecht Majewski-Bosma syndrome which was associated with atrial septal defect [39], the fourth case of the syndrome of microtia/anotia syndrome and congenital facial palsy without other abnormalities in the world [42].

7. Conclusion

This one-year Iraqi pioneering experience in the fields of clinical genetics and clinical dysmorphology should herald the end of the Dark Age in these fields in Iraq.

8. Acknowledgement

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