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ORIGINAL ARTICLE

Dermatosis in down syndrome

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Abstract

Down syndrome is the most common chromosomal disorder, affecting 1 in 700 live births. It has typical physical features and multisystem anomalies that include the skin. This study aims determine the dermatological characteristics among patients attended at a Down syndrome outpatient clinic. Cross-sectional study conducted by the review of medical records of patients attended in 2018. Data were submitted to descriptive analysis using Statistica® software. Dermatologists worked at the Down syndrome outpatient clinic one day per week. They evaluated 838 patients, of which 474 (56.6%) were male and 364 (43.4%) were female. The most frequent dermatoses were xerosis (57.6%), follicular keratosis (26.3%), papular urticaria (17.6%), palmoplantar hyperkeratosis (11.9%) and sudammine (9.1%). Diagnosing and treating dermatoses in Down syndrome patients are an integral part of the clinical follow-up of this population, and skin self-care should be encouraged from childhood.

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INTRODUCTION

Down syndrome (DS) is the most common chromosomal disorder, characterised by 95% regular trisomy 21 and 5% in the form of translocation and mosaic[1]. Because of the presence of extra genetic material from chromosome 21, children with Down syndrome have multiple malformations, medical conditions and cognitive impairment[1,2]. Congenital heart diseases are present in 50% of DS patients, and of these 24.8% need corrective cardiac surgery. The clinical and surgical treatment has increased the survival of these patients. Over recent decades, their life expectancy has increased from 25 years in 1983, to 49 years in 1997, to 65 years in 2010[1,3].

The most frequent dermatological conditions in DS are listed in Table 1[4].

DS is associated with several dermatological problems. Premature ageing is one of the phenotypic features of DS and is reflected in age-dependent skin changes including premature graying of the hair, hair loss, and skin wrinkling. Skin disorders are more prevalent in DS than in the general population, and they include alopecia areata (often severe), vitiligo (often associated with endocrine disorders), milia-like calcinosis cutis, syringoma (mainly from adolescence) and onychomycosis. Atopic dermatitis, on the other hand, is not the most frequent in the general population[1].

The skin of the newborn DS patient is soft, thin and similar to that of the general population; however, they may present cutis marmorata, malar erythema and nevus simplex on the lumbosacral skin, all of which are considered clinical signs of DS and remain unchanged with age. During childhood, due to changes in keratin, the skin becomes thick, dry and rough, with an increased frequency of xerosis, keratosis pilaris and palmoplantar[5,6].

METHODOLOGY

This study was approved by the Research Ethics Committee of the Clinical Hospital of the Federal University of Paraná (Brazil Platform No. 04542712.3.0000.0096). It was conducted in patients with Down syndrome at a public referral center that was created in 1997 in Curitiba, South Brazil. The purpose of the interdisciplinary team in this outpatient clinic is to ensure comprehensive health care for these patients and support for their families.

Of the 4,500 patients at the outpatient clinic, the medical records of 838 were included. We excluded adult patients. Included patients were seen in 2018 and evaluated by dermatology.

RESULTS

There were 838 patients, and their distribution by age group is shown in Table 2.

The sample included 463 (55.3%) male and 375 (44.7%) female patients. They had their first dermatologic consultation within a median of 2 months. The parents had a mean age of 32 years, ranging from 26 to 40 years. Sixty per cent had an elementary and high school education and a monthly income of 1 to 2 times the minimum wage. They came from Portuguese-speaking families with an income of 2.5x the minimum wage.

Table 1. Dermatoses in down syndrome patients.

Skin and cutaneous appendages phenotypic characteristics	Malar erythema
	Nevus simplex on the lumbosacral skin
	Cutis marmorata / livedo reticularis
	Café au lait spots
	Xerosis
	Palmoplantar hyperkeratosis
	Fine and sparse hair
	Synophrys
	Ungual dystrophy
	Photosensitivity and premature skin aging
	Scrotal tongue
Autoimmune cutaneous diseases	Vitiligo
	Alopecia areata
Inflammatory skin diseases	Seborrheic dermatitis
	Blepharitis
	Atopic dermatitis
	Psoriasis
Cutaneous sign of peripheral insulin resistance	Acanthosis nigricans
Bacterial skin infections	Folliculitis
	Hidradenitis
Viral cutaneous infections	Warts
	Molluscum contagiosum
Fungal infections	Tineas
	Superficial mycoses
Common dermatoses	Calcium-like cysts of milia
	Crusted scabies
	Serpiginous perforating elastosis
	Syringoma
	Keratin abnormalities
	Frictional tylosis
	Angular cheilitis
	Onychophagia
	Cutaneous trauma

Table 2. Distribution of patients with down syndrome by age group.

AGE GROUP	n=838
	N
0 to 5 years	147
6 to 10 years	327
11 to 20 years	474

Regarding the perinatal characteristics, a higher prevalence of women with a previous gestation was observed, most had prenatal care, and rates of vaginal and surgical delivery were similar. A history of spontaneous abortion was documented in 21% of the mothers. The patients were mostly term-delivered and were breastfed in 75% of the cases. Simple trisomy was observed in 94.6%, translocation in 3.1% and mosaicism in 2.3%.

The frequency of the dermatoses of our patients is in Table 3.

Table 3. The frequency of dermatoses of down syndrome patients.

DERMATOSE	n	100%
Xerosis	500	57,6
Follicular keratosis	230	26,5
Papular urticaria	103	11,9
Palmoplantar hyperkeratosis	153	17,6
Sudammine	79	9,1
Livedo reticularis	71	8,2
Seborrheic dermatitis	70	8,1
Folliculitis	70	8,1
Nevus	50	5,8
Malar erythema	46	5,3
Atopic dermatitis	45	5,2
Onychomycosis	35	4,0
Acne vulgaris	33	3,8
Candidiasis	32	3,7
Mongolian spot	25	2,9
Impetigo	23	2,6
Alopecia areata	21	2,4
Nevus simplex (lumbosacral)	20	2,3
Lichenoid reactions	19	2,2
Angular cheilitis	19	2,2
Superficial mycosis	18	2,1
Café au lait spot	15	1,7
Diaper dermatitis	15	1,7
Salmon spot	14	1,6
Scabies	14	1,6
Blepharitis	13	1,5
Hidradenitis supurativa	13	1,5
Molluscum contagiosum	13	1,5
Acanthosis nigricans	12	1,4
Psoriasis	10	1,2
Miliaria	8	0,9
Vitiligo	8	0,9
Ichthyosis	7	0,8
Syringoma	7	0,8
Xerosis	7	0,8
Follicular keratosis	4	0,5
Papular urticaria	3	0,3
Palmoplantar hyperkeratosis	3	0,3
Sudammine	2	0,2

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DISCUSSION

We evaluated our 838 patients with DS and compared them with those of other studies (Table 3)[1,7,9]. The most frequent dermatoses were xerosis (57.6%), follicular keratosis (26.3%), papular urticaria (17.6%), palmoplantar hyperkeratosis (11.9%) and sudammine (9.1%).

Reviews of other studies show variations in the frequency of dermatoses in DS patients[1,8,9]. The most common dermatoses in follow-up patients were similar to the ones found by El Achkar et al. (2007)[7], with a significant difference only in xerosis and malar erythema. Also, Rork et al. (2020)[10] found higher frequency of folliculitis, seborrheic dermatitis and hidradenitis supurativa, because they included young adults in their study.

The frequency of skin diseases in DS patients varies with age. Xerosis, keratosis pilaris and palmoplantar hyperkeratosis increase in frequency in infancy with age[5,7].

Chromosomal abnormalities, as seen in DS, have specific dermatological characteristics. The skin of the newborn with DS is soft and thin, but palmoplantar hyperkeratosis begins as early as infancy. During childhood, the skin becomes thick, dry and rough, with generalised xerosis commonly associated with keratosis pilaris. In the mucous membranes, macroglossia, tongue extrusion, scrotal tongue, and angular cheilitis can be observed[7].

Livedo reticularis (or cutis marmorata), telangiectatic nevus of the lumbar region and malar erythema are considered phenotypic dermatoses of DS[5,6].

The onset of numerous autoimmune, inflammatory and infectious dermatoses is facilitated by alterations in cellular and humoural immunity, with a decreased number and function of B and T lymphocytes and impaired phagocytosis. DS most commonly exhibits keratin abnormalities such as keratosis pilaris and xerosis[6].

Early skin ageing and photosensitivity are common features of the syndrome. The mechanisms are not yet well understood, but may be related to the effects of free radicals[7].

The most frequently described dermatoses in DS are cutis marmorata (livedo reticularis), xerosis, palmoplantar hyperkeratosis, cheilitis, seborrheic dermatitis, folliculitis, tinea pedis, onychomycosis, scabies (Norwegian mange), atopic dermatitis, alopecia areata, vitiligo, psoriasis (severe form), pityriasis rubra pilaris, syringoma, serpiginous perforating elastosis and verticis girata cutis[4,5,7,10-14]. Ryan et al. (2020)[15] included images of many dermatoses in patients with DS in their article.

Table 4. Comparison of the frequency of dermatoses of down syndrome patients.

DERMATOSIS	BERMUDEZ et al. (2018) n=838	RORK Et al. (2020) n=101	EL ACHKAR et al. (2007) n=382	MALT et al. (2013)
	%	%	%	%
Xerosis	57.6	21.8	30.6	
Follicular keratosis	26.5	7.9	7.8	
Papular urticaria	17.6		13.6	
Palmoplantar hyperkeratosis	11.9	5.9	17.8	10–40.8
Sudammine	9.1		13.9	
Livedo reticularis	8.2	6.9	10.5	2–12.6
Seborrheic dermatitis	8.1	26.7	4.4	3–36
Folliculitis	8.1	30.7	3.4	10.3–21.1
Nevus	5.8	5.9	8.1	
Malar erythema	5.3		15.4	1.4–4.9
Atopic dermatitis	5.2	20.8		4.4–67.8
Onychomycosis	4.0	9.9	1.0	
Acne vulgaris	3.8	10.9		
Candidiasis	3.7		1.0	
Mongolian spot	2.6		14.7	
Impetigo	2.4		2.1	2.9–11
Alopecia areata	2.3	6.9	0.3	
Nevus simplex (lumbosacral)	2.2		2.9	
Lichenoid reactions	2.2		1.0	
Angular cheilitis	1.7	6.9		2.4–25
Superficial mycosis	1.7			1.9
Café au lait spot	1.6		6.8	
Diaper dermatitis	1.6		6.0	
Salmon spot	1.5		6.8	
Scabies	1.4		0.5	
Blepharitis	1.2		0.5	
Hidradenitis supurativa	1.2	22.8		
Molluscum contagiosum	0.9		0.5	
Acanthosis nigricans	0.9		0.5	
Psoriasis	0.8	8.9	0.5	
Miliaria	0.8		0.8	
Vitiligo	0.3		0.3	1.9–3
Ichthyosis	0.3			
Syringoma	0.2			6–39.2

Medial telangiectatic vascular nevi are capillary vascular malformations often observed at birth. They occur mainly on the face or neck as a single lesion or as multiple macules that affect more than one site simultaneously[13]. In 1990, Metzker and Shamir reported a medial telangiectatic vascular nevus in the sacral region along the midline, and they referred to this particular variety as “butterfly-shaped” and frequent in chromosomopathies. They can also be located occipitally, and they persist into childhood and adulthood[10].

Diagnosing and treating dermatoses in DS patients is an integral part of the clinical follow-up of this population, and skin self-care should be encouraged from childhood.

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