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RESULTS

Eight outpatient dermatology clinics participated (bolded above). There were 1,529 patients (49% female; 51% male). The majority of patients were children 0-12 years old (51%), followed by adolescents 13-17 years old (25%) and adults 18 years or older (24%).

Table 1 captures the most common overall ICD-10 diagnoses and then stratifies diagnoses by age group. Eczematous/xerotic, adnexal, and autoimmune dermatologic conditions are the most reported. The most common cutaneous infections were onychomycosis and tinea pedis, which occurred in all age groups. Alopecia areata was second to eczematous dermatitis as the most common skin condition in children 0-12 years.

Six patients were diagnosed with non-melanoma skin cancer. No patients were diagnosed with melanoma.

66 patients (4.3%) were treated with high risk medications (ICD-10 code Z79.899) for a variety of skin conditions (**Table 2**).

Table 3 lists the top 3 diagnoses with the highest frequency for return outpatient visits.

DISCUSSION

This multi-site retrospective review of ICD-10 codes confirms findings of prior single-site retrospective reviews: eczematous, adnexal, and autoimmune skin conditions are the most common findings in individuals with DS at outpatient dermatology visits.²⁻⁵

Trisomy of chromosome 21 and the resulting downstream effects on the immune system, including dysregulation of interferon and amyloid precursor proteins, likely account for these skin findings.⁶⁻⁷

It may be prudent for clinicians to monitor for progression from folliculitis to hidradenitis suppurativa (HS), especially in adolescents 13-17 years who are 3.4 times more likely than children 0-12 years to progress from folliculitis to HS.

Limitations, include variance in coding practices, evolving medical coding guidelines, uneven age distribution, and poor translatability from diagnosis coding to disease severity.

CONCLUSION

This study provides guidance for clinicians and researchers to prioritize screening, diagnosis and management, and basic science and clinical research of common skin conditions in people with DS.

BACKGROUND

Down syndrome (DS) is the most common chromosomal condition in the United States and Canada with an estimated worldwide incidence of one in every 1000 births.¹ While the majority of dermatology literature has focused on case reports of rare skin conditions diagnosed on individuals with DS, more recent single-site retrospective reviews have revealed the burden of common diagnoses such as adnexal (folliculitis, hidradenitis suppurativa), autoimmune (alopecia areata), eczematous (seborrheic dermatitis), and infectious (dermatophyte) conditions.²⁻⁵

OBJECTIVES

Review demographics and outpatient visit International Classification of Diseases, tenth revision (ICD-10) codes of children and adults with DS evaluated at a dermatology practice.

METHODS

Eligible patients had an ICD-10 diagnosis of DS and an outpatient dermatology visit between January 2011 and December 2021. Nurse visits were excluded. All participating sites reported demographics (sex and age at visit(s)) and associated ICD-10 diagnosis codes. Since ICD-10 codes were not instituted until October 1st, 2013, some sites chose to exclude visits until this date or manually converted ICD-9 codes to equivalent ICD-10 codes if their electronic medical record system did not automatically. Data was stored in Dartmouth Hitchcock Health REDCap database. Statistical analysis was conducted with R version 4.2.1.

Table 1. Top ICD-10 codes reported in all patients with Down syndrome stratified by age group

ICD-10 Codes and Diagnoses		Overall (n=1,529)	Children (n=776)	Adolescents (n=386)	Adults (n=366)
Eczematous and Xerotic	L20.84, L20.9, L30.9 Eczematous dermatitis	397 (26.0%)	238 (30.7%)	77 (19.9%)	82 (22.4%)
	L21.9 Seborrheic dermatitis	239 (15.6%)	74 (9.5%)	76 (19.6%)	89 (24.3%)
	L85.8 Keratosis pilaris	194 (12.7%)	91 (11.7%)	63 (16.3%)	40 (10.9%)
	L85.3 Dry skin	151 (9.9%)	79 (10.2%)	40 (10.3%)	32 (8.7%)
	L71.0 Perioral dermatitis	100 (6.5%)	68 (8.8%)	16 (4.1%)	16 (4.4%)
	K13.0 Angular cheilitis	41 (2.6%)	17 (2.2%)	16 (4.2%)	8 (2.2%)
Adnexal	L73.8, L73.9 Folliculitis and other specified follicular disorders	295 (19.3%)	109 (14.0%)	112 (28.9%)	74 (20.2%)
	L73.2 Hidradenitis suppurativa	200 (13.1%)	44 (5.7%)	94 (24.3%)	62 (16.9%)
	L70.0 Acne vulgaris	167 (11.0%)	44 (5.7%)	73 (18.9%)	50 (13.7%)
Autoimmune	L63.8, L63.9 Alopecia areata	178 (11.6%)	135 (14.6%)	24 (6.2%)	19 (5.19%)
	L80 Vitiligo	66 (4.3%)	51 (6.6%)	10 (2.6%)	5 (1.4%)
Papulosquamous	L40.9 Psoriasis	102 (6.7%)	44 (5.7%)	27 (7.0%)	31 (8.5%)
Neoplasm	D22.9 Melanocytic nevi, atypical nevi	101 (6.6%)	28 (3.6%)	25 (6.8%)	48 (13.1%)
	D23.9 Benign neoplasm	67 (4.4%)	25 (3.2%)	14 (3.62%)	28 (7.7%)
Cutaneous Infection	B35.1 Onychomycosis	90 (5.9%)	33 (4.3%)	31 (8.0%)	26 (7.1%)
	B35.3 Tinea pedis	77 (5.0%)	16 (2.1%)	30 (7.8%)	31 (8.5%)
	B07.8, B07.9 Verruca vulgaris and other viral warts	77 (5.0%)	37 (4.8%)	20 (5.2%)	20 (5.5%)
	L02.92 Furuncle	36 (2.4%)	7 (0.9%)	16 (4.1%)	13 (3.6%)
Other	Z79.899, High-risk medication use	66 (4.3%)	23 (3.0%)	21 (5.4%)	22 (6.0%)

Table 2. Dermatologic conditions associated with high risk medication use (Z79.899)

ICD-10 Codes and Diagnoses N (%)	Patients (N = 66)
L70.0 Acne vulgaris	23 (34.8%)
L73.2 Hidradenitis suppurativa	21 (31.8%)
L20.9, L30.9 Eczematous dermatitis	12 (18.2%)
L40.9 Psoriasis	9 (13.6%)
L63.8 & L63.9 Alopecia areata	4 (6.1%)
L80 Vitiligo	3 (4.6%)

Table 3. Dermatological conditions with the most frequent clinic visits

ICD-10 Code	Number of Visits Avg (SD), Median [Range]
L40.9 Psoriasis	5.27 (6.4), 2.0 [1 – 27]
Z79.899, Other long term (current) drug therapy	4.62 (4.6), 3.0 [1 – 21]
L73.2 Hidradenitis suppurativa	4.42 (5.4), 2.0 [1 – 38]



Figure 1. Clinical presentation of commonly reported skin conditions (A) Xerosis, (B) Seborrheic dermatitis, (C) Alopecia areata, (D) Folliculitis, (E) Hidradenitis suppurativa, (F) Onychomycosis

REFERENCES

- Graaf Gd, Buckley F, Skotko B. *Estimation of the Number of People with Down Syndrome in the United States*. Genetics in Medicine. 2017; 19: 439-447. <https://doi.org/10.1038/gim.2016.127>
- Wentworth AB, Hand JL, Davis DM, Tollefson MM. Skin concerns in patients with trisomy 21 (Down syndrome): A Mayo Clinic 22-year retrospective review. *Pediatr Dermatol*. 2021 Nov;38 Suppl 2:73-78. doi: 10.1111/pde.14764. Epub 2021 Aug 18. PMID: 34409638.
- Garg A, Strunk A, Midura M, Papagermanos V, Pomerantz H. Prevalence of hidradenitis suppurativa among patients with Down syndrome: a population-based cross-sectional analysis. *Br J Dermatol*. 2018;178(3):697-703.
- Rork JF, McCormack L, Lal K, Wiss K, Belazarian L. *Dermatologic conditions in Down syndrome: A single-center retrospective chart review*. 2020; 37(5): 811-816. <https://doi.org/10.1111/pde.14214>.
- Firsowicz M, Boyd M, Jacks SK. Follicular occlusion disorders in Down syndrome patients. *Pediatr Dermatol*. 2020;37(1):219-221.
- Huggard D, Doherty DG, Molloy EJ. Immune dysregulation in children with Down syndrome. *Front Pediatr*. 2020;8:73.
- Blok J, Jonkman M, Horvath B. The possible association of hidradenitis suppurativa and Down syndrome: is increased amyloid precursor protein expression resulting in impaired notch signalling the missing link? *Br J Dermatol*. 2014;170(6):1375-1377.