



Thyroid Dysfunction and Hashimoto Thyroiditis in Children with Down Syndrome: A Mexican Referral Center Experience

José Francisco Figueroa Pérez¹, Montserrat Vázquez García¹, Karla A. Flores Arizmendi¹

Instituto Nacional de Pediatría – Universidad Nacional Autónoma de México.



1.- BACKGROUND

- Children with Down syndrome (DS) have the highest prevalence of endocrine disorders among genetic conditions, with thyroid dysfunction representing the most frequent abnormality.
- Autoimmune thyroid disease develops earlier and more frequently than in the general pediatric population owing to trisomy 21-associated immune dysregulation, chronic interferon hyperactivation, and oxidative stress.
- Because clinical manifestations often overlap with the DS phenotype, current international guidelines recommend lifelong biochemical thyroid surveillance.



2.- OBJECTIVE

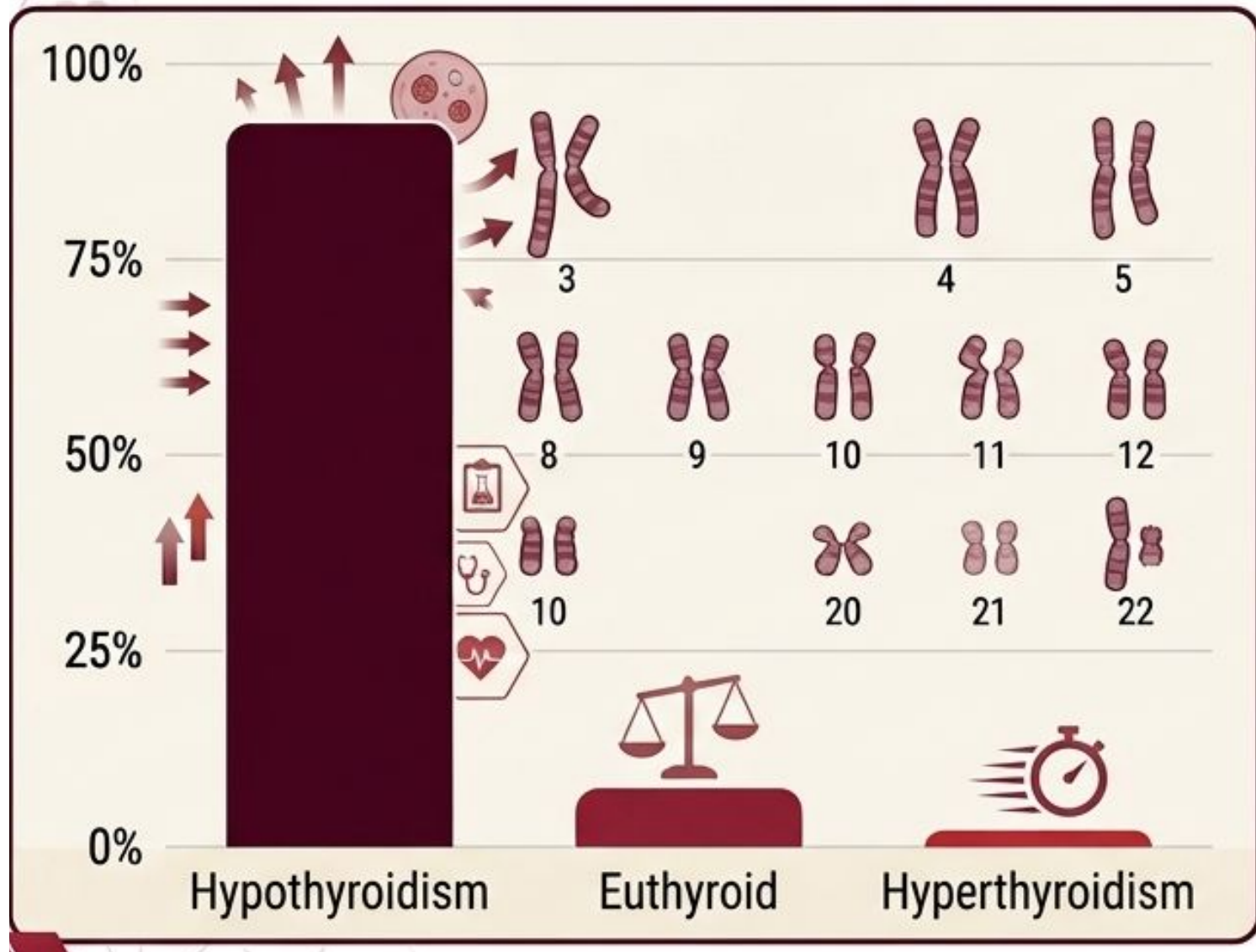
To describe the prevalence, clinical characteristics, thyroid autoimmunity, nutritional status, and growth outcomes of children with Down syndrome followed at a national referral center and compare these findings with current international evidence.

3.- METHODS

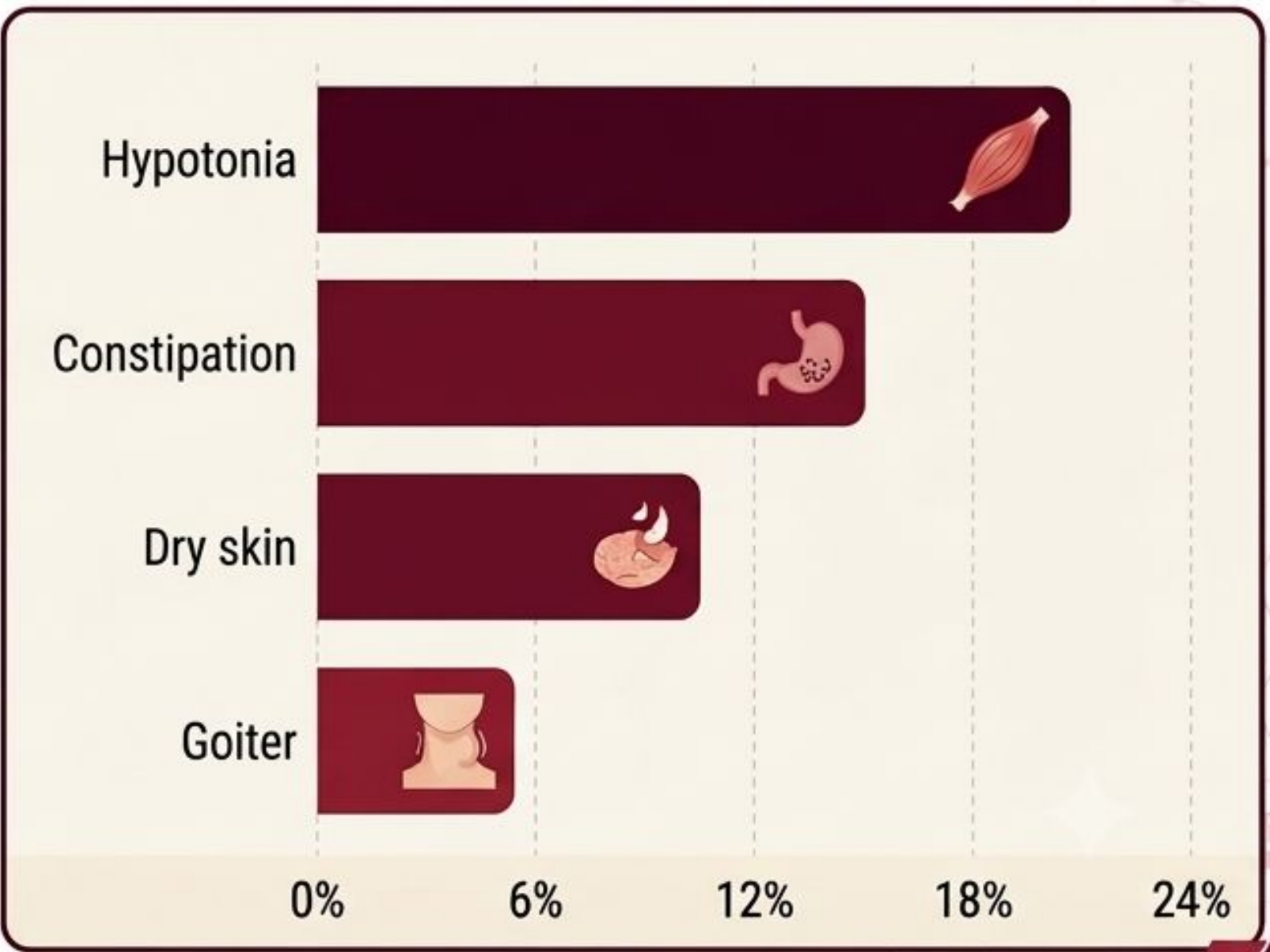
Retrospective observational study of 100 children with DS evaluated between 2013–2019. Clinical, biochemical, anthropometric and nutritional variables were analyzed using Down syndrome-specific reference standards.

4.- RESULTS

DISTRIBUTION OF THYROID DISORDERS



CLINICAL MANIFESTATIONS



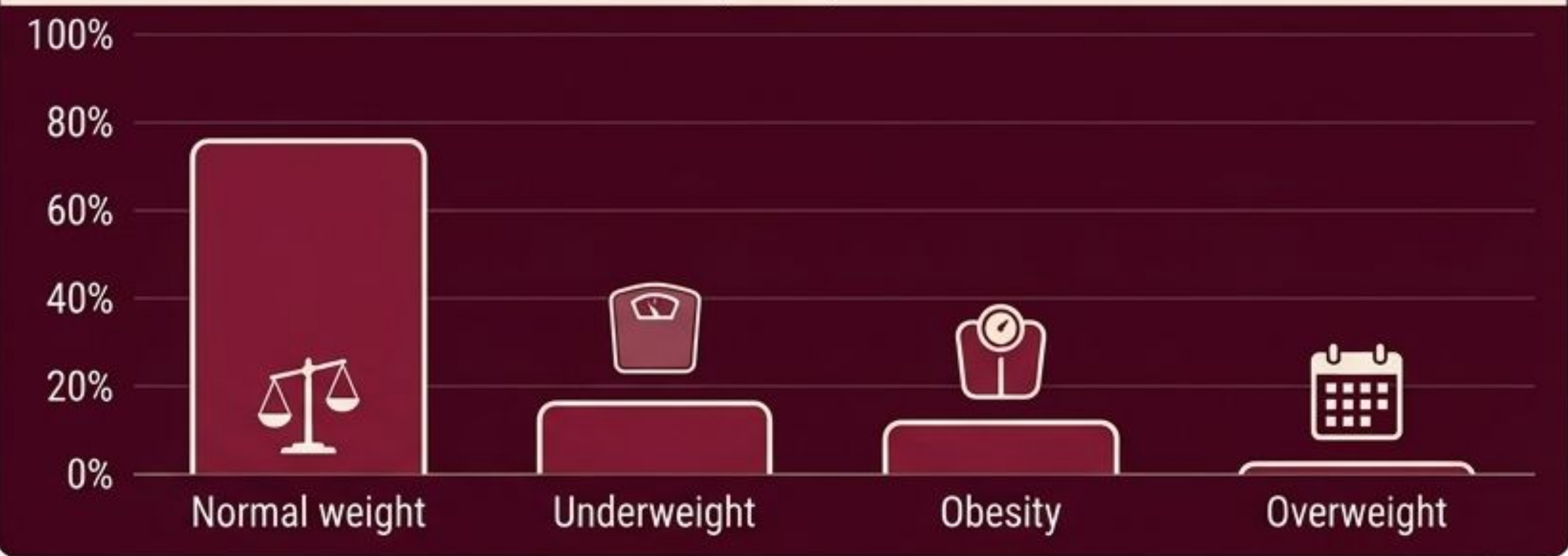
KEY FINDINGS

VARIABLE		RESULT
Total cohort		100
Mean age at thyroid diagnosis		≈3 years
DETAILED FINDINGS		
Hashimoto thyroiditis		24%
Male antibody positivity		14%
Female antibody positivity		10%
Mean age of antibody positivity		3.3 years
Interval from thyroid dysfunction to antibody positivity		7–9 months
First-degree family history		12.5% (3/24)

PRESENT STUDY COMPARED WITH INTERNATIONAL EVIDENCE

STUDY	MAIN FINDING	STUDY FINDING INTEGRATION	MEXICAN COHORT RESULT
AAP (2022)	Lifelong thyroid surveillance		✓ Supported
DSMIG	Symptoms are unreliable		✓ Confirmed
Pierce et al.	Autoimmunity increases with age		Autoantibody positivity identified at ~3 years
International cohorts	Anti-TPO predicts progression		Similar findings
Mexican cohort	–		24% Hashimoto, early diagnosis, limited family history

PERCENTAGE DISTRIBUTION OF NUTRITIONAL STATUS (N=100)



18.5% PATIENTS WITH HYPOTHYROIDISM HAD SHORT STATURE

5.- CONCLUSIONS

Hypothyroidism was the predominant thyroid disorder (91%). Hashimoto thyroiditis affected one in four children and appeared early. Symptoms and family history were poor predictors of disease. Routine biochemical surveillance remains essential for early diagnosis and treatment.

6.- CLINICAL RELEVANCE

Nearly one in four children with DS developed autoimmune thyroid disease despite a very low frequency of positive family history. Classical clinical manifestations were uncommon, and almost one in five children with hypothyroidism presented with short stature.

7.- LIMITATIONS

Single-center retrospective study. Autoantibody measurements were unavailable for all patients. Longitudinal outcomes were not evaluated.

8.- FUTURE DIRECTIONS

Prospective multicenter studies are needed to better define the natural history of autoimmune thyroid disease in Latin American children with DS.

9.- KEY CLINICAL MESSAGE

Routine biochemical thyroid screening should not depend on symptoms or family history. Early surveillance enables prompt diagnosis of autoimmune thyroid disease, supports normal growth, and may reduce additional neurodevelopmental burden in children with DS.

REFERENCES

Bull MJ, et al. *Pediatrics*. 2022.
Tsou AY, et al. *JAMA*. 2020.
Zalzal HG, & Lawlor CM. *JAMA Otolaryngol Head Neck Surg*. 2023.
Konishi A, et al. *Clin Endocrinol*. 2025.
DSMIG. *Thyroid Guidelines*. 2020.
Global Down Syndrome Foundation. *Medical Care Guidelines*. 2020.
Vázquez García M. *UNAM–National Institute of Pediatrics*. 2020.