

Comorbidity Burden in Children and Adolescents with Down Syndrome

Attending a Tertiary Pediatric Hospital in Chile, 2016–2024

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Background

Children and adolescents with Down syndrome (DS) experience a high multisystem comorbidity burden and frequently require longitudinal multidisciplinary care. Local pediatric data from Chile remain scarce.

Global incidence: 1.42/1,000 live births, decreasing.
Chile: 2.87/1,000 live births (2001–2005), increasing.

Objective

To describe cumulative comorbidity prevalence and multimorbidity across organ systems in ambulatory patients with DS.

Methods

- Retrospective cross-sectional study. Electronic medical record review.
- Hospital Dr. Luis Calvo Mackenna, Santiago, Chile. Outpatient multidisciplinary clinic. From Jan, 2017 to Jun, 2024.
- n = 144 patients with DS.
- Multimorbidity: ≥2 affected organ systems.

Table 1. Study population

Characteristic	n / %
Total patients	144
Female / Male	42% / 58%
Age 0–5 / 6–12 / ≥13 yr	35% / 33% / 32%
Prenatal diagnosis (n=76)	21.0%
Free trisomy 21 (n=90)	93.0%
Premature (<37 wk) /Extreme preterm(<32 wk)	18.1% / 3.5%

Strengths and limitations

Strengths: clinically detailed tertiary cohort; multisystem classification; age-group comparisons.

Limitations: retrospective design, partial variable availability, and referral bias inherent to a tertiary center.

Clinical interpretation

These findings support structured surveillance, integrated care pathways, and local prioritization of underdetected domains such as sleep and cardiometabolic comorbidity.

Results

Graph 1. Comorbidity prevalence by organ system (%)

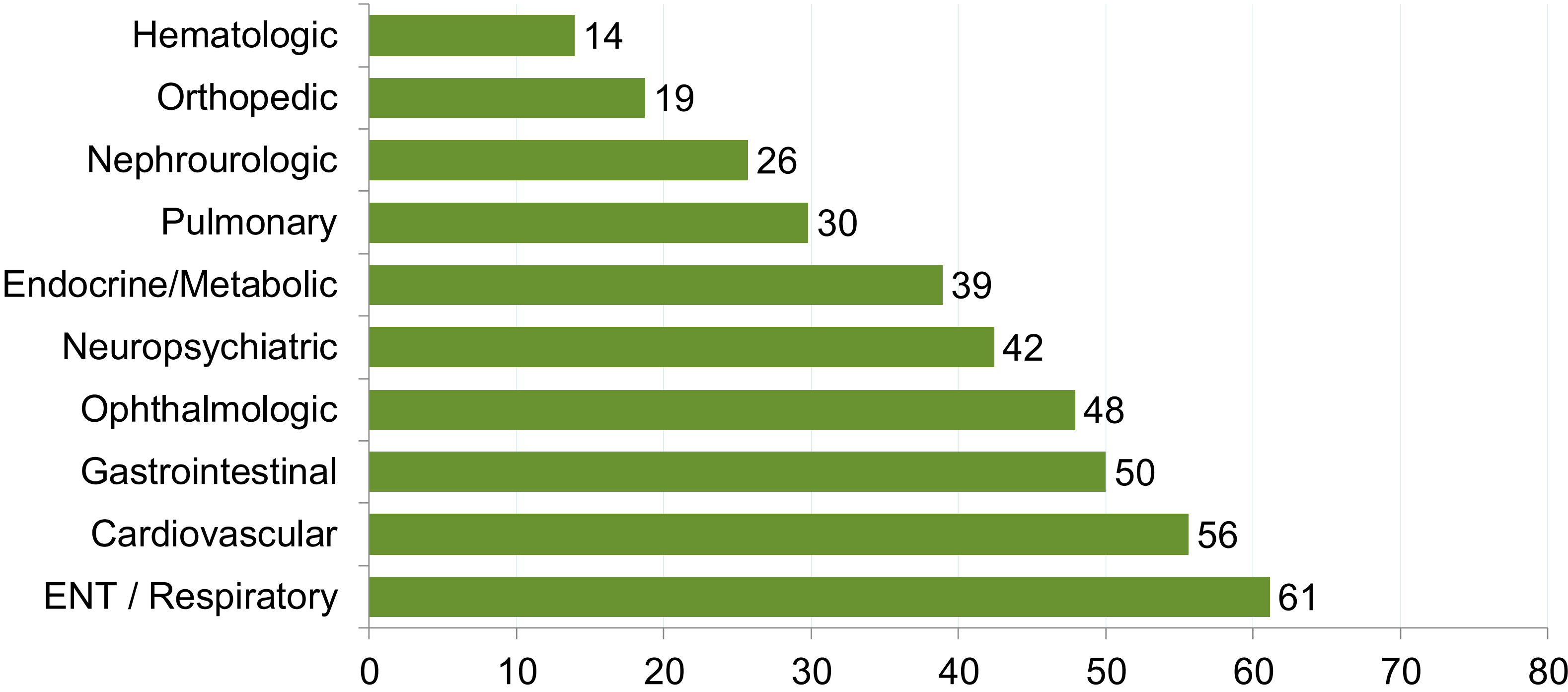


Table 2. Most common specific comorbidities

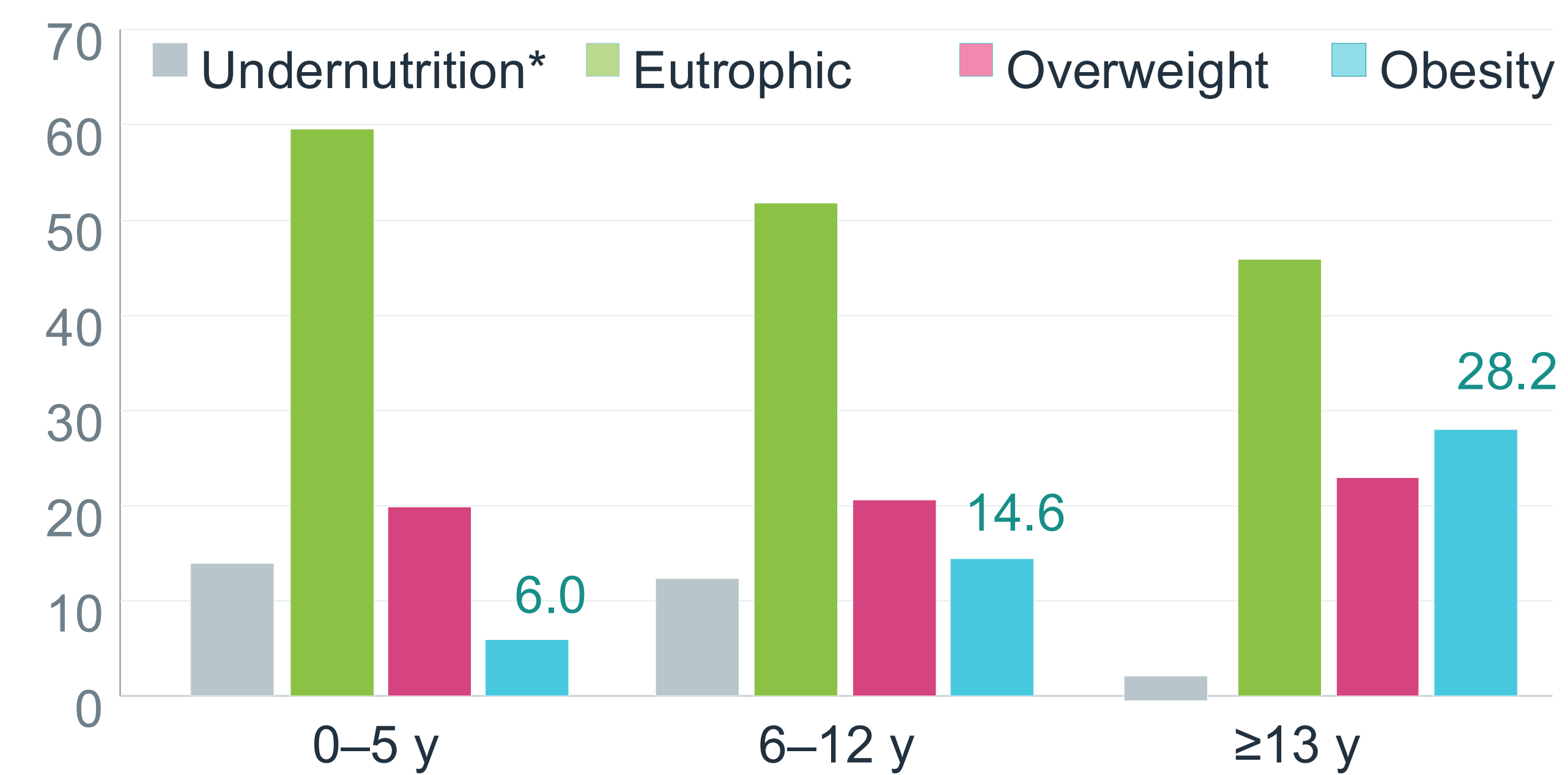
System / Diagnosis	n	%
ENT		
Allergic rhinitis	52	36.1%
SRBD / OSA	38	26.4%
OME / chronic OM	29	20.1%
Hearing loss	16	11.1%
Cardiovascular		
Any congenital HD	80	55.6%
AV canal (isolated)	18	12.5%
VSD (isolated)	15	10.4%
PDA (treated)	14	9.7%
Tetralogy of Fallot	7	4.9%
Gastrointestinal		
Constipation	40	27.8%
Swallowing disorder	27	18.8%
GI malformations	16	11.1%
Ophthalmology		
Refractive errors	41	28.5%
Strabismus	27	18.8%
Neuropsychiatry		
ADHD	22	15.3%
Behavioral disorder	19	13.2%
ASD	14	9.7%
Endocrine / Metabolic		
Hypothyroidism	35	24.3%
Insulin resistance	7	4.9%
Other key findings		
Ferritin <50 ng/mL	66	45.8%
Vit D deficiency/insuf.	34/104	32.7%
Obesity (≥13 yr)	12/44	27.3%

96.5%
with ≥1
comorbidity

3.9
mean affected
systems/patient

76.4%
followed by ≥3
specialties

Graph 2. Nutritional status by age group



* Underweight + risk of undernutrition.

Key messages and Discussion

- 96.5% had at least one comorbidity; mean burden: 3.9 affected systems per patient.
- The most affected systems were ENT (61%), and cardiovascular (56%) and GI (50%).
- Lower hearing loss (11.1% vs 47–75%).** Only chart-recorded diagnoses; mild/conductive losses often missed. OME reached 20.1%, indicating a significant undiagnosed conductive component.
- Lower OSA/SRBD (26.4% vs 50–75%):** Registry-based diagnosis without systematic PSG. Real sleep disorder burden is likely substantially underestimated due to limited access to polysomnography.

Conclusions

This tertiary pediatric cohort showed a very high comorbidity and multimorbidity burden. Findings support context-adapted surveillance, standardized screening, and better registry capture.

In Down syndrome, multimorbidity is not an added complication; it is the baseline condition from which care must be organized.