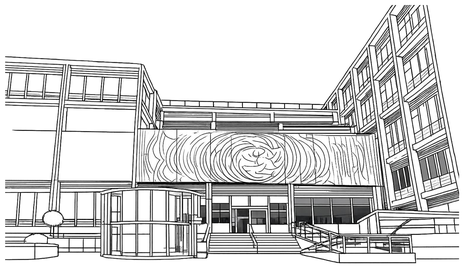

Cholestasis in Children with Down Syndrome: Clinical Characteristics, Etiology, and Outcomes in a Pediatric Cohort

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BACKGROUND

Neonatal cholestasis affects approximately 1 in 2,500–5,000 live births and occurs more frequently in children with Down syndrome (3.9%). While biliary atresia is the leading cause of neonatal cholestasis in the general population, the etiologic spectrum in Down syndrome appears to be distinct.

OBJECTIVE

To describe the clinical, biochemical, etiological, and outcome characteristics of children with Down syndrome diagnosed with cholestasis in a pediatric cohort.

METHODS

A retrospective observational study was conducted at the Clínica Down of the Instituto Nacional de Pediatría, Mexico City. Medical records of patients with Down syndrome and cholestasis evaluated between January 2010 and June 2026 were reviewed. Only patients with complete clinical data were included. Demographic characteristics, age at onset and diagnosis, clinical manifestations, comorbidities, etiologies, treatment, and outcomes were analyzed.

RESULTS

KEY FINDINGS

14	Patients with Down syndrome and cholestasis were included.
64.3%	Male patients.
2 days	Median age at cholestasis onset.
13 days	Median age at diagnosis.
100%	Patients presented with jaundice.
85.7%	Patients had congenital heart disease.
Neonatal hepatitis	Most frequent etiology, followed by giant cell neonatal hepatitis.
0%	Extrahepatic biliary atresia cases identified.
92.9%	Patients achieved complete resolution of cholestasis.
7.1%	One patient died.

Fourteen patients with Down syndrome and cholestasis were included. Males accounted for 64.3% of cases. The median age at cholestasis onset was 2 days of life, while the median age at diagnosis was 13 days of life. Fig 1. Clinical manifestations are shown in Figure 2.

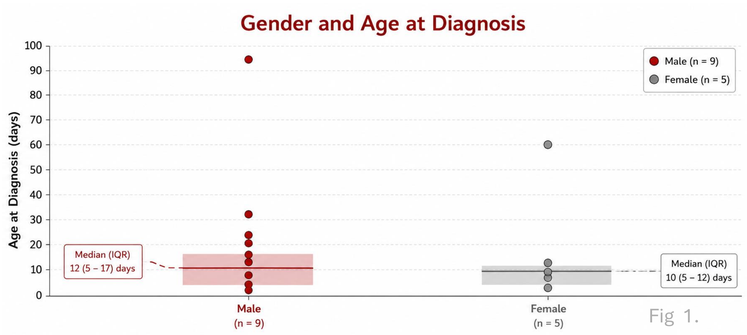

Fig 1.

Each point represents one patient.
Horizontal line indicates the median; shaded box represents the interquartile range (IQR).

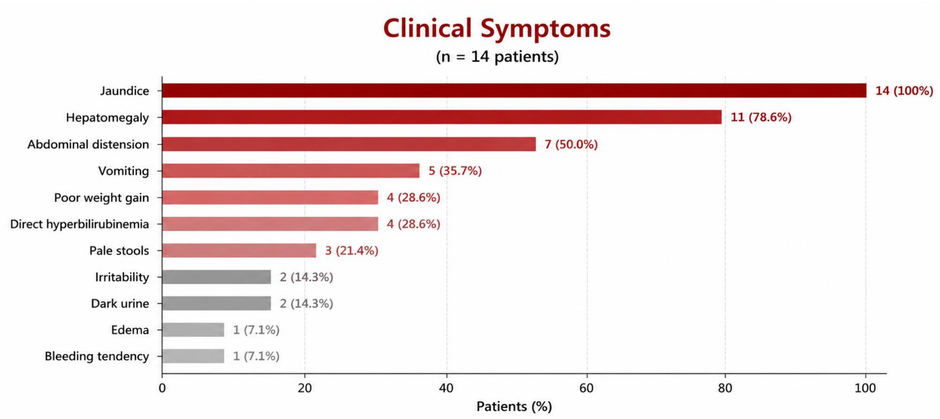

Fig 2.

Data are presented as number of patients and percentage.
Some patients presented more than one symptom.

Congenital heart disease was the most common comorbidity (85.7%), followed by sepsis, pneumonia, and hypothyroidism (Fig 3).

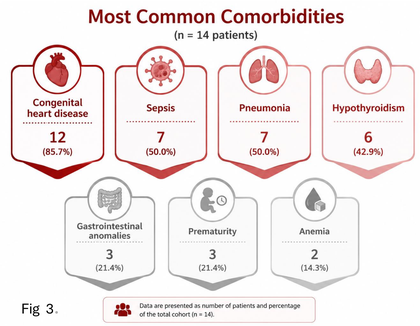

Fig 3.

Data are presented as number of patients and percentage of the total cohort (n = 14).

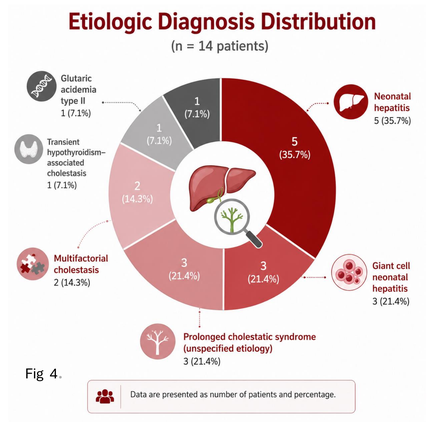

Fig 4.

Data are presented as number of patients and percentage.

The most frequent etiologies were neonatal hepatitis and giant cell neonatal hepatitis, followed by prolonged cholestatic syndrome of unspecified etiology and multifactorial causes (Fig 4).

Clinical outcomes were favorable in most cases, with resolution of cholestasis in 92.9% of patients. One patient died.

CONCLUSIONS

Cholestasis in Down syndrome predominantly presents during the neonatal period and is most commonly associated with congenital heart disease, infections, and hypothyroidism. Neonatal hepatitis was the leading etiology, and no cases of extrahepatic biliary atresia were identified in this cohort. Most patients experienced complete resolution, supporting a generally favorable prognosis. Larger multicenter studies are needed to further define risk factors and optimal management strategies.

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