



# Disorders of Gut-Brain Interaction in Children With Down Syndrome

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**Article:** Disorders of gut-brain interaction in a national cohort of children with Down syndrome  
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Down syndrome is associated with multiple physical and psychiatric comorbidities caused by chromosomal abnormality. The incidence is around 1 in 600-700 live births worldwide and is not different according to race or nation. Because of multiple health problems including congenital heart disease, congenital gastrointestinal (GI) obstruction, hematologic or endocrine disorders and high susceptibility of infection, quality of life (QoL) of pediatric patients with Down syndrome and their caregivers is dependent on the severity of the comorbidities.<sup>1</sup> According to the present article, besides the congenital abnormalities, disorders of gut-brain interaction (DGBIs) could also contribute the QoL of the patients and their caregivers.<sup>2</sup>

DGBIs, previously called functional gastrointestinal disorders (FGIDs) have been diagnosed using the Rome IV criteria. Rome IV criteria for symptom based diagnosis of FGIDs for child and adolescent have been reported previously.<sup>3</sup> Due to the possibility of coexistence of other medical conditions that could result in GI symptoms, the application of the Rome IV criteria (21.2%) resulted in a significantly lower prevalence of FGIDs in children than the application of Rome III criteria (23.7%,  $P = 0.004$ ).<sup>4</sup> In a representative community sample, pediatric FGIDs were common (25.0%),

with functional constipation being the most common FGID in children and adolescents (14.1%) among pediatric FGIDs. QoL assessed using PedsQL4.0 in pediatric patients with FGIDs in the representative community also decreased (71.69 vs 87.16,  $P < 0.001$ ) as in the present study.<sup>5</sup> Although the population composition in the representative community was not disclosed, subjects might be heterogeneous ( $n = 1255$ ). They might have a small number of Down syndrome patients, considering the incidence of the chromosomal abnormality. The present study showed higher DGBIs prevalence in this homogenous population of Down syndrome (52.0%, aged 4-18 years) than the previous heterogeneous community population (25.0%, aged 4-18 years).<sup>2</sup> Among FGIDs, pediatric Down syndrome patients most commonly struggled with functional constipation (36.0%) as in previous reports.<sup>5,6</sup>

GI changes occurred in 300 (49.0%) of 1207 patients with Down syndrome, excluding GI symptoms caused by congenital digestive tract malformations such as duodenal atresia, Hirschsprung disease, or imperforate anus. Chronic constipation was the most prevalent among various GI changes.<sup>7</sup> Chronic constipation in Down syndrome is associated with hypotonia, short segment of Hirschsprung disease, hypothyroidism, celiac disease and mental

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problems such as intellectual disability or autism.<sup>8</sup> Although functional constipation was the most common DGBI in children with Down syndrome in the present study, caution was necessary to manage chronic constipation because it was often caused by diverse comorbidities.<sup>2</sup> Sometimes malformations might be detected late, especially in case of refractory constipation despite appropriate measures.<sup>9</sup>

Diagnosis of DGBIs and assessment of QoL were investigated with online questionnaires using the Rome IV Diagnostic Questionnaire (Rome IV Diagnostic Questionnaires: theromefoundation.org) and the Peds QL4.0, respectively.<sup>2,10</sup> Using questionnaires might have limitations to measure prevalence because of the possibility of different interpretations or agreement of similar symptoms in FGIDs.<sup>11</sup> Limitations of the present study were a small number of patients included and an entirely racial deviation to white despite using a national cohort and diagnosis of DGBI from a questionnaire based on caregivers' opinions. Still, the results were helpful to understand GI symptoms in Down syndrome regarding underlying medical conditions and DGBIs. Health care of comorbidities raised the estimated survival for patients with Down syndrome up to 60 years in 2020. However, due to the lack of awareness of FGIDs in Down syndrome and low QoL of their caregivers, more professional health care might be needed for the GI symptoms and QoL of their families.<sup>12,13</sup>

Although GI symptoms in pediatric patients with Down syndrome are associated with diverse physical or mental comorbidities, the high prevalence of DGBIs and the low QoL in families of children with Down syndrome in the present study suggest the need for promoting adequate management of DGBIs as well as comorbidities.

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