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A GENETIC CAUSE OF BREATHING ABNORMALITIES AND SEVERE ABDOMINAL DISTENSION

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Rett syndrome is an X-linked dominant progressive neurologic developmental disorder and one of the most common causes of intellectual disability in females . It is caused by mutations in methyl-CpG-binding protein 2 (MECP2), a ubiquitously expressed factor.[1] The patients usually exhibit autistic behaviors, gradual loss of speech, characteristic stereotyped hand movements, intellectual disability, ataxia, seizures, autonomic dysfunction (constipation, gastroesophageal reflux, aerophagia, hyperventilation, breath-holding episodes and Cardiovascular autonomic dysfunction). [2,3]Patients usually survive till adulthood. We report a 5-years-old girl with Rett syndrome with worsening breathing abnormalities, over several months, including irregular breathing, breath-hold or apnea, gasping, alternative hyper and hypoventilation. These breathing abnormalities were associated with daily recurring episodes of air swallowing, severe abdominal distension and bloating (Figure/video 1). The episodes were exacerbated with stress and anxiety. The patient’s general condition at admission was good, weight was 14.2 kg (10th percentile), height 109 cm (50th percentile) and head circumference 48 cm (3th percentile). Physical examination showed excessive air swallowing associated with visible severe abdominal distension. Ultrasonography did not reveal any organomegaly or fluid presence in the abdomen. No abdominal pain or other gastrointestinal symptoms were present. Electrocardiogram was normal. A routine basic metabolic panel was within normal limits. The patient underwent a complete standardized neuropsychiatric examination that considered the recurring episodes of air swallowing as a stereotypical symptom such