

NPTR VIDEO CATALOG ENTRY

Disease	Glutaric aciduria type I (GA1). Organic acidemias.
Title	Noah & Ruby's story – living with GA1.
Link	https://youtu.be/tTcF7MLUICo
Key words	Glutaric aciduria type I. Organic acidemias. Patient/family experience. General disease overview. Symptoms. Pathophysiology. Biochemistry. Newborn screening. Diet/treatment. Prognosis.
Description	Ruby and Noah are two children with glutaric aciduria type 1; Noah, the older child, presented clinically with an episode of severe clinical decompensation, resulting in long-term neurologic sequelae. His younger sister, Ruby, was identified shortly after birth and treated immediately; she is growing and developing normally. The parents describe their experience of raising two children with this disease, highlighting the importance of being positive and setting goals. Three metabolic experts, two physicians and one dietitian, explain the disease, the treatment approach, and the benefit from an early identification through newborn screening and onset of therapy. Viewers can observe some of Noah's dystonic posturing and movements.
Length (min:sec)	9:38
Speaker(s) Background	Parents. Physicians and dietitian (metabolic experts).
Objectives	<ol style="list-style-type: none"> 1. Describe the characteristic course of untreated GA type I vs early recognized/treated GA type I. 2. Identify the treatment plan for managing a patient with GA type I. 3. Characterize the multi-disciplinary approach necessary for managing such patients.
Educational utility	Provides a personal narrative about living with the disease but would be more effective if accompanied by some medical teaching.
Technical aspects	Excellent audio and visual quality.

Relevant/ Target Audience	Health professionals and specialists. Trainees and students in the health professions. Other stakeholders who have an interest in the inborn errors.
Promotional aspects	This video does not promote a commercial product, treatment, and/or medical device.
Source	NIHRtv - NIHR CLAHRC Yorkshire.
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