

NPTR VIDEO CATALOG ENTRY

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| Disease | Biotinidase deficiency. Vitamin and cofactor metabolism defects. |
| Title | BabySeq: Partial Biotinidase Deficiency |
| Link | https://www.youtube.com/watch?v=CruC-7B-T4E |
| Key words | Patient/family experience. Newborn screening. Diet/treatment. |
| Description | The patient's mother describes her experience in encountering her daughter's diagnosis through newborn screening and a clinical diagnostic research study. She also mentions the treatment. |
| Length (min:sec) | 1:33 |
| Speaker(s) Background | Mother. |
| Objectives | 1. Describe the experience of encountering a diagnosis through newborn screening and a clinical research study. |
| Educational utility | Provides a personal narrative about encountering a disease but should be accompanied by significant medical teaching to increase understanding about the disorder. |
| Technical aspects | Excellent audio quality and good video 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 contains comments that promote the BabySeq research study. This video does not otherwise promote a commercial product, treatment, and/or medical device. |
| Source | Genomes2People. |

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| Author | JA. |
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