Isabelle

Isabelle's story is certainly a remarkable and inspiring one. It's evident that she and her family have faced numerous challenges and difficult decisions along the way. The discovery of the complete deletion of the Cathepsin A enzyme on the CTSA gene, leading to galactosialidosis, presented a severe early-infantile form of the condition.

The family's determination to save Isabelle's life led them to pursue a bone marrow transplant (BMT) at Charité in Berlin, which was the first of its kind for her condition. The chemotherapy and stem cell transplantation aimed to correct the missing enzyme in her body, and it appears that the treatment was successful in improving her metabolic function and enzyme levels.

However, Isabelle experienced kidney failure approximately one year after the BMT. While it's challenging to pinpoint the exact cause, galactosialidosis could have contributed to this complication. She currently relies on peritoneal dialysis and requires a new kidney for a more lasting and effective treatment.

Despite the challenges she faces, Isabelle continues to shine brightly in her family's eyes. She has shown progress in certain areas, such as sitting independently, using her hands to communicate her needs, and making vocalizations. While she may be delayed in meeting some milestones, her family believes she understands everything and has confidence in her abilities.

Isabelle's journey is not only significant to her and her family but also to the scientific community. Her unique case and treatment process will be closely followed, as they may provide valuable insights into the treatment of galactosialidosis in the future. Isabelle's resilience and the support of her family are truly inspiring, and they continue to hope for her well-being and development while seeking a suitable kidney for her ongoing care.