

Nemours Children's Study Demonstrates Babies Prenatally Diagnosed with Lethal Osteogenesis Imperfecta May Survive with Medical Interventions

Study indicates that the severity of the condition is challenging to predict in utero



Ricki S. Carroll, MD

WILMINGTON, Del. (March 4, 2025) — For decades, the rare bone condition osteogenesis imperfecta has been diagnosed on a spectrum, from mild to lethal. New research by Nemours Children's Health physicians suggests that many babies whose cases are prenatally classified as "lethal" survive and may benefit from ongoing advances in treatment.

Osteogenesis imperfecta (OI) is a rare genetic disorder that occurs in 1 of every 15,000 to 20,000 births. Also known as "brittle bone disease," OI causes fractures or broken bones and bone instability and fragility. Patients with severe forms of the condition may experience significant respiratory problems or death.

Ricki S. Carroll, MD, Physician on the Skeletal Dysplasia and Palliative Care teams at Nemours Children's Hospital, Delaware, explained that while patients with mild forms of OI are often not diagnosed until they experience multiple fractures during childhood, those with severe cases are often diagnosed in utero, when fractures or unusual bone measurements are detected via ultrasound. The most severe cases are labeled as "lethal."

"Parents facing this prenatal diagnosis may be counseled that the disease will limit their child's life, and that they should opt to forego life-sustaining interventions at birth," Carroll said.

As a palliative care specialist, Carroll had observed that many babies who were prenatally diagnosed with OI responded well to treatment in the neonatal intensive care unit (NICU). In a new study published in *JBMR Plus*, Carroll and colleagues examined the cases of 18 infants with OI who were cared for in the Nemours NICU between September 2019 and August 2024. Twelve had been given a prenatal diagnosis of "lethal" or "possibly lethal" OI. (Four were diagnosed with less severe forms of OI, and severity-specific diagnostic information was not available for two.) All 18 infants survived to hospital discharge, and 16 are alive today with minimal breathing or feeding support needs.

Carroll explained that prenatal prognosis in OI is based on criteria including decreased chest-to-abdomen and femur length-to-abdomen ratios. However, those measurements may not be accurate due to fractures or the bowed legs that are a characteristic of OI.

Carroll recommends that babies with a known OI diagnosis be delivered in an advanced delivery unit, with interventions prepared to support children with life-threatening conditions. For example, in order to survive, most infants with OI will need respiratory support and pain control due to fractures sustained in utero or through the birth process. Skilled nursing care is also needed for the infants, limiting movement to allow their injuries to heal. Babies may be prescribed bisphosphonate to strengthen their bones.

The 18 babies in Carroll's study experienced a range of health issues and early interventions. Two babies died of respiratory ailments, at 7 months and 13 months of age.

Among the others, seven required respiratory support at discharge, and nine required feeding support. Most were weaned off such support in the following 18 months. Five have had orthopedic procedures including realignment or rodding. The patients have achieved a wide range of mobility milestones—for example, of the eight patients aged 2 and up, one walks without support; one walks with support; one scoots, stands, and uses a power wheelchair; two crawl, scoot and stand with support; and the others are rolling with support.

Carroll credited the multidisciplinary support these OI patients received at Nemours for their survival and wellness.

"Babies born with OI do face challenges," she said. "But for families who pursued supports early on, we saw that infants born with a possibly lethal diagnosis were able to survive and actually do very well."

In February 2024, Viana Kelly of Jersey City, New Jersey, gave birth to her son Koa, who had been prenatally diagnosed with lethal OI. He was scheduled to be discharged home on hospice, until she heard about the Nemours OI team. Kelly transferred Koa's care to Nemours Children's Hospital, Delaware, when he was just 11 days old. After nearly two months in the hospital, Koa went home. He recently celebrated his first birthday. He is active and happy, scooting, playing and enjoying trips to the zoo.

"I saw progress in him every day," Kelly said. "There was a time when I thought, 'This is where it ends.' But with time and with the care he received in the NICU at Nemours, he is doing really well."

Carroll said Koa's case exemplifies how advances in OI treatment can help patients survive and achieve a good quality of life.

"Our findings show that it is difficult to prognosticate the severity of OI prenatally, and that it is reasonable to offer

interventions to families who wish to pursue them,” Carroll said.

Carroll said she and colleagues at Nemours Children’s already avoid the term “lethal,” instead classifying OI cases as “mild, moderate, or severe.” She said prenatal testing can provide guidance on the severity of the disease, but may not provide a fully accurate picture.

“This important research questions physicians’ ability to predict lethality in utero. Our suggestion is that all infants with OI should be offered medical intervention as an option, which is not currently the case,” said Mahim Jain, MD, Medical Director, Orthogenetics, Nemours Children’s Health.

Carroll and Jain said future research will examine the role of all aspects of caring for infants with OI, from delivery to physical therapy. Given the rarity of the disease and the lack of physicians with experience treating severe cases at birth globally, they aim to continue to publish research and disseminate their findings to further advance treatment and results.

About Nemours Children’s Health

Nemours Children’s Health is one of the nation’s largest multistate pediatric health systems, which includes two freestanding children’s hospitals and a network of more than 70 primary and specialty care practices. Nemours Children’s seeks to transform the health of children by adopting a holistic health model that utilizes innovative, safe, and high-quality care, while also addressing children’s needs well beyond medicine. In producing the highly acclaimed, award-winning pediatric medicine podcast Well Beyond Medicine, Nemours underscores that commitment by featuring the people, programs and partnerships addressing whole child health. Nemours Children’s also powers the world’s most-visited website for information on the health of children and teens, Nemours KidsHealth.org.

The Nemours Foundation, established through the legacy and philanthropy of Alfred I. duPont, provides pediatric clinical care, research, education, advocacy, and prevention programs to the children, families and communities it serves. For more information, visit Nemours.org.

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