Treating diseases in the womb: In medical first, doctors treat Pompe disease before birth

A toddler is thriving after doctors in the U.S. and Canada used a novel technique to treat her before she was born for a rare genetic disease that caused the deaths of two of her sisters.

Ayla Bashir, a 16-month-old from Ottawa, Ontario, is the first child treated as fetus for Pompe disease, an inherited and often fatal disorder in which the body fails to make some or all of a crucial protein.

In a case study published [November 9] in the New England Journal of Medicine, doctors describe an international collaboration during the COVID-19 pandemic that led to the treatment that may have saved Ayla’s life – and expanded the field of potential fetal therapies. The outlook for Ayla is promising but uncertain.

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Doctors have treated fetuses before birth for three decades, often with surgeries to repair birth defects such as spina bifida. And they’ve given blood transfusions to fetuses through the umbilical cord, but not medicines. In this case, the crucial enzymes were delivered through a needle inserted through the mother’s abdomen and guided into a vein in the umbilical cord. Ayla received six biweekly infusions that started at about 24 weeks of gestation.

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