

# Medical research team cures fatal infant disease molybdenum cofactor deficiency type-A

September 4 2015

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Monash Health's pioneering treatment for babies with a disease that was once universally fatal has been validated by research published today in prestigious journal, *The Lancet*.

"We don't use the word cure very often in medicine, but this research shows that under the right circumstances this is a cure," Dr Flora Wong said.

Dr Wong, a Monash Health neonatologist and researcher with Monash University and the Hudson Institute of Medical Research, was part of a global research project that has shown a disease that had no hopes for survival can now be treated effectively. "We now know that early and careful treatment of [babies](#) born with this rare condition can lead to survival with good long-term developmental outcomes," Dr Wong said.

In 2009 Monash Health made medical and legal history by curing 'Baby Z,' who was the first baby to ever survive molybdenum cofactor deficiency Type-A, a previously incurable disease that had no prospects of survival. Babies born with the disease did not survive past early infancy.

Molybdenum cofactor deficiency Type-A is a rare condition that causes toxic sulphites to rapidly accumulate in the brain of newborns, causing seizures and the death of brain tissue, leading to death.

After Baby Z's diagnosis with the illness, Monash Health neonatologists led by Dr Alex Veldman identified a possible treatment, cPMP (cyclic pyranopterin monophosphate). It had never before been used in humans and required unprecedented and fast-tracked legal approvals before being administered.

The treatment rapidly improved Baby Z's sulphite (S-sulfocysteine) levels. Although she has experienced ongoing health issues, the innovative treatment allowed her to become the first human to ever survive the horrific condition. Please note that Baby Z's identity remains suppressed for legal reasons.

Data showed that S-sulfocysteine levels rapidly improve once treatment commences. The new research published today has shown that the compound is even more effective than previously thought. With earlier administration of the treatment before brain injury ensues, we have seen

babies survive this illness with no significant complications whatsoever. "Because this is a rare condition we took a global approach, and treated 11 babies affected by molybdenum cofactor deficiency Type-A from around the world with the compound. It was safe, and we've shown that early [treatment](#) can be amazingly effective over a long time," Dr Wong said.

Provided by Monash University

Citation: Medical research team cures fatal infant disease molybdenum cofactor deficiency type-A (2015, September 4) retrieved 13 May 2024 from <https://medicalxpress.com/news/2015-09-medical-team-fatal-infant-disease.html>

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