

Holy glycosylation! New 'bat signal' flags distressed cells in childhood genetic diseases

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Just as Gotham City uses the Bat Signal to call for Batman's aid, a new tool developed by scientists from the Sanford-Burnham Medical Research Institute in La Jolla, California, should serve as the cellular equivalent for children with glycosylation disorders, sometimes called "CDG syndromes." In a new report appearing online in *The FASEB Journal*, scientists describe how they used a green fluorescent protein to identify the presence of genes—known and unknown—associated with a wide variety of glycosylation-related diseases. By being able to identify exactly which genes are defective, researchers can develop treatments and therapies to correct the root causes of these diseases rather than merely treating the symptoms.

Glycosylation is an enzymatic process that coats proteins, lipids or other organic molecules with sugar molecules. It helps cells "stick" together, and proteins fold and work properly, among other things. When this process does not function correctly, it causes diseases involving intellectual disability, digestive problems, seizures and low blood sugar.

"We hope this glowing protein will help light the path for the discovery of new [genes](#) that cause genetic disorders in children," said Hudson Freeze, Ph.D., a senior researcher involved in the work from the Genetic Disease Program at Sanford-Burnham Medical Research Institute in La Jolla, California. "It's not Harry Potter's magic wand, but we hope it will offer a way to test for new therapies in these kids. They're counting on us."

To make this advance, Freeze and colleagues engineered cells from children with glycosylation disorders so the cells would glow to indicate when there was a glycosylation problem related to a defective or missing gene. Once the problematic, glowing cells were "rescued" by inserting a healthy gene into the cell or correcting a defective gene's function, the cells stopped glowing. This new tool may be used in high-throughput screening to identify therapeutic molecules that improve glycosylation in defective cells, including stem cells. In addition, this advance may serve as the foundation for a new diagnostic tool for patients.

"These glowing proteins serve as a hotline between distressed cells and researchers hoping to restore their normal function," said Gerald Weissmann, M.D., Editor-in-Chief of The [FASEB Journal](#). "Knowing when and where there are [cells](#) with flawed glycosylation pathways should allow researchers to rapidly screen for compounds that may have therapeutic potential."

More information: Marie-Estelle Losfeld, Francesca Soncin, Bobby G. Ng, Ilyas Singec, and Hudson H. Freeze. A sensitive green fluorescent protein biomarker of N-glycosylation site occupancy. *FASEB J.* [doi:10.1096/fj.12-211656](https://doi.org/10.1096/fj.12-211656)

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