Abstract Title: N=1

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Abstract: N=1 is the story of one child- my second born son diagnosed at age 3 with severe type 1 Gaucher Disease. In 1983 there were no treatments and such a diagnosis was a death sentence. It is a story of a mother-physician researcher at the USA National Institutes of Health, Dr Roscoe Brady, and a visionary entrepreneur, Henri Termeer-who were brought together under the guiding hand of the divine to save this child's life. This child, on the edge of high cardiac output failure, was given an enzyme extracted from human placenta, and modified to increase uptake by macrophages.The enzyme rapidly improved symptoms including reducing hepatosplenomegaly, increasing hemoglobin and platelet counts, decreasing bone pain, improving energy. This N of 1 led to clinical trials and FDA drug approval in 1991, followed by approvals in Israel and sequentially other countries. Eventually, the drug was synthesized using recombinant methods and is currently being produced by various pharmaceutical companies. In 1984, my family created the National Gaucher Foundation to provide patient education, advocacy, and research funding. NGF became a model for other LSD patient groups globally. the success of Gaucher treatments led to the whole field of LSDs, and the extensive academic research, pharmaceutical drug development, patient advocacy groups, and humanitarian outreach programs that exist today.

Area of expertise: Diagnosis and Treatment of Gaucher Disease including adjunctive anti inflammatory interventions