Balancing the Sweetness of the Family Between Monogenic Hypoglycemia and Multifactorial Hyperglycemia

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A breezy summer morning, a routine day with chiming OPD bells, ever attentive attendants calling out patients' names and patients awaiting their turn to enter the OPD chambers. As we were going through our daily routine, a middle-aged couple walked in with their pretty 7-year-old girl and a portly 20-year-old boy. The couple were married for the past 22 years and had their first son, 1 and half years into marriage. As their son became 6 months old, he started throwing seizures. Ever since, the couple had been roaming from pillar to post; from homoeopathy to Ayurvedic to allopathic medicine, they had tried it all. The final diagnosis was eventually unravelled at our institute when their son turned 3 years of age. He was diagnosed to have glycogen storage disorder (GSD) type III through a liver biopsy. Days passed with the family trying everything on earth towards making sure that their son had a normal life. During this roller coaster ride, the couple had gone through four terminations of pregnancies as they were too overwhelmed to welcome a new-born. Days passed into months and months into years and the couple had finally made a call towards extending their family when their son entered his teens.A tiny little angel entered their family, and their happiness knew no bounds. Unfortunately, fate does not care about anyone's plans. At 6 years of age, the little girl suffered from loss of weight and during workup was found to have autoimmune thyroiditis and type I diabetes mellitus. Balancing the two disorders of sugar homeostasis in two kids, monogenic hypoglycemia in one child and multifactorial hyperglycemia in another was indeed challenging and left the couple totally shattered. But still, they did not lose hope and they had decided to fight it out. They did genetic testing of their son which confirmed the diagnosis of GSD III. The treatment options were re-discussed with stress on the need of adhering strictly to the treatment for good outcome. The children are being managed carefully, doing well and the family is happy and well-adjusted now. Twenty-five percent risk of recurrence of glycogen storage disorder in the next offspring was briefly mentioned with the background thought that they seemed to have completed the family, the eldest son being twenty years old, doing relatively well and preparing for his medical entrance exams. They were further counselled that no genetic testing is recommended in the second child as it is a multifactorial disease.As a part of a concluding clockwork note, we had asked the couple if they had any further queries for which we got a reply from the father in a very hesitant manner about his desire to extend his family.In fact, the couple had come to the genetics department solely for this reason.They were worried about the possibility of recurrence of both the disorders. A detailed discussion about the risk of recurrence of monogenic glycogen storage disorder and multifactorial diabetes mellitus followed.

Referrals for genetic counselling for multifactorial diseases like neural tube defects are common but uncommon for disorders like schizophrenia, diabetes, celiac disease, etc. We pay little less attention to the issue of recurrence for multifactorial disorders as compared to the monogenic disorders. Prenatal diagnosis for these multifactorial diseases is not possible. But for the family, each disease matters and it does not depend on the number of genes in the etiology or the figure of recurrence. They had come with the hope of ensuring that the next child would be a healthy individual who would eventually take on the responsibility of looking after his/her elder siblings. After detailed counselling, this family understood that prenatal diagnosis for glycogen storage disorder will be possible but not for diabetes mellitus for which the risk of recurrence is also relatively low.

Following this session,they were also given an option of screening for common genetic disorders. There was a prompt and swift response from the couple and during the next OPD visit, they had diligently followed all the instructions and had arrived with all the necessary reports. The interaction with the family was pleasant and an enriching experience in genetic counselling and emphasized that age is just an issue of mind over matter and desires have no fixed timelines. Good communication from both sides is the key to successful genetic counselling.This was indeed an eye opener for us and taught us the importance of discussing the reproductive options in all cases despite the number of children in the family and age of the parents and children, and address the risk of recurrence for multifactorial disorders. Hence,we decided to pen it down.....