embryos biopsied and frozen on day 5 (n=61). The average age of patients having a fresh embryo transfer on day 6 is 32±3.2 and having frozen embryo transfer is 30±3.7 years old. The results showed that pregnancy rates were not significantly different between frozen embryo transfer and fresh embryo transfer (59% vs. 52% respectively, P value >0.05). Nevertheless, as well as indicating that not only is frozen embryo transfer as good as or better than fresh embryo transfer, frozen embryo transfer can also have advantages in in-vitro fertilization in allowing optimal embryo transfer planning for couples.

**Keywords:** Array comparative genome hybridization (aCGH); in vitro fertilization, fresh blastocyst transfer; frozen-thawed blastocyst transfer


**AB096. Pharmaco-genetic guided personalized medicine: discovery of a maturity onset diabetes of the young (MODY2) novel mutation [S441W in glucose kinase (GCK) gene] by next generation sequencing (NGS)**

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**Background and objective:** Monogenic diabetes or maturity onset diabetes of the young (MODY) is characterized by young-onset (<45 years old), non-insulin dependence and a strong family history (autosomal dominant mode of inheritance). The major candidate genes include HNF4α (MODY1), glucose kinase (GCK) (MODY2) and HNF1α (MODY3). MODY is an attractive model for pharmaco-genetics because an accurate diagnosis may inform specific choice of anti-hyperglycemic therapy for better clinical outcome. We report a slim lady (BMI 22.4 kg/m²) with Type 1 diabetes diagnosed based on abnormal fasting glucose and oral glucose tolerance test at age 21. She was started on multiple daily insulin injections (total daily dose 18-22 units/day) with good glycemic control (HBA1c 6.2%). Glutamic acid decarboxylase (GAD) autoantibody was negative. On occasions when she ran out of insulin supply, there was no incidences of diabetic ketoacidosis. We aim to identify proband with monogenic diabetes phenotype to perform high through-put exonic mutation screening using next-generation sequencing (NGS) on an extended panel of candidate genes (i.e., beyond GCK, HNF1A and HNF4A) for these individuals. We will also recruit other members within the pedigree for segregation analysis to strengthen causality of discovered variant (this is necessary primarily because more variants-of-unknown-significance are expected to be observed in an extended gene panel).

**Methods:** DNA from peripheral blood cells was subjected to high-throughput targeted nucleotide sequencing for all 16 known MODY candidate genes including exons, untranslated (UTRs) and promoter regions using the Ampliseq kit (Life Technologies).

**Results:** We discovered a novel non-synonymous mutation (c.1322C>G, p.Ser441Trp) in the GCK gene, which was confirmed by bi-directional Sanger’s sequencing. The mutation is predicted to be functionally deleterious using multiple bioinformatics tools (e.g., SIFT and PolyPhen-2). In accordance with clinical practice guideline, insulin replacement was successfully discontinued with no deterioration of glycemic control.

**Conclusions:** The successful treatment-conversion based on genotype exemplifies how pharmaco-genetics can improve disease-stratify to inform diagnosis and treatment. This can translate into improved clinical outcome and quality of life.

**Keywords:** Diabetes; maturity onset diabetes of the young (MODY); mutation; sequencing