***Intrauterine growth restriction and hypertrophic cardiomyopathy as prenatal ultrasound findings in a case of leprechaunism***

**Supplementary Material**

For Perge et al. *‘Intrauterine growth restriction and hypertrophic cardiomyopathy as prenatal ultrasound findings in a case of leprechaunism’*.

**Supplementary table: Characteristics of the mutation identified in the *INSR* gene**

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **Chromosome****Position (hg19)** | **Gene** | **dbSNP rsID** | **cDNA** | **Protein** | **Allele Counts WT/Mutated (MAF)****in Public Databases** | **In silico Pathogenicity** | **ACMG****classification** |
| **Refseq** | **Nucleotide****Change** | **Refseq** | **Amino Acid****Change** | **gnomAD****(N= 141,456)** | **EVS****(N=6,503 )** | **GME****(N = 2099)** | **Polyphen 2****(score)** | **Provean****(score)** | **Sift****(score)** | **CADD****score** | **UMD predictor****(score)** |
| chr19:7267376 | *INSR* | - | NM\_000208 | c.632C>T | NP\_000199 | p.T211I | Absent | Absent | Absent | Probably damaging(0.999) | Deleterious(-4.04) | Damaging(0.002) | 24.4 | Pathogenic(75) | Pathogenic |