***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 |