



Features	<i>HNF4A</i>	Fanconi Bickel	Mother	Son
Macrosomia	7/14 (50%)	-	-	-
Hypoglycemic hyperinsulinism	18/18 (100%)	-	+	+
Maturity Onset Diabetes of the Young/Diabetes	6/10 (60%)	-	+	N/A
Atypical renal Fanconi syndrome	17/17 (100%)	- Renal tubular dysfunction	+	+
Hypophosphatemic Rickets	8/9 (89%)	+	+	-
Nephrocalcinosis	8/14 (57%)	-	-	+
Hepatomegaly	6/16 (38%)	+	+ Liver cysts	+
Splenomegaly	2/11 (18%)	+	+	-
Hearing loss	2	-	-	-
Iris and chorioretinal coloboma	1	- Cataracts	-	+
Penile Chordee and hypospadias	1	-	-	+