

Patient	PKHD1 sequencing	Panel sequencing (1): Panel v.1 (2): Panel v.2	MLPA PKHD1	MLPA HNF1B	PKD1 sequencing
1/88512	+	+(1)	+	+	+
2/88412	+	+(1)	+	+	+
3/89313	+	-	-	-	-
4/135812	+	-	-	-	-
5/117712	+	-	-	-	-
6/147912	+	+(1)	+	+	+
7/135912	+	-	-	-	-
8/134013	+	+(1)	+	+	+
9/137113	+	-	-	-	+
10/94312	+	+(1)	+	+	+
11/117812	+	+(1)	+	+	+
12/88612	+	+(1)	+	+	+
13/136012	+	+(1)	+	+	-
14/88712	+	+(1)	+	+	+
15/88812	+	-	-	-	-
16/86512	+	-	-	-	-
17/86312	+	-	-	-	-
18/88312	+	-	-	-	-
19/76512	+	+(1)	+	+	+
20/88912	+	+(1)	+	+	+
21/86412	+	-	-	-	-
22/71615	+	+(1)	+	+	+
23/37815	+	-	-	-	-
24/09416	+	Commercial lab	+	-	-
25/104016	-	+(1)	-	-	+
26/51515	-	+(1)	-	-	+
27/44916	-	+(1)	-	-	+
28/118418	-	-	-	-	+
29/123818	-	+(2)	-	-	-
30/173718	-	+(2)	-	-	-
31/166818	-	+(2)	-	-	+