There has never been a report of a family having a second child with PKS that I am aware of. So empirically based on that we would say the risk is as close to zero as possible. It can never be completely "zero" since there is always the possibility of there being a second "de novo" event (the chance of which would be around the population risk of having a child with PKS times itself or: 1/20,000 x 1/20,000 or 1/400,000,000). In some other diagnoses we have seen recurrence due to germ line mosaicism - cases in which parents do not carry the genetic change in most cells in their body but do carry the change in a group of their germ cells (e.g. sperm or eggs) and can result in subsequent affected children - but again to stress we have NEVER seen this in PKS.

The other complicating issue to consider in all of this is that some kids with PKS clinical fathers actually have trisomy 12p (a duplication of all of some of 12p material - instead of a triplication that we see in typical PKS) - in some of these case the extra 12p material can be the result of a balanced chromosomal rearrangement in a parent and therefore can recur in subsequent pregnancies - but again most families in this scenario will know that risk and it is quite different than the more typical tetrasomy 12p (resulting in that extra isochromosome) seen in PKS.

Dr. Ian Krantz Jan 2013