

From Dr. Ian Krantz. February 2010

There are a few aspects to this difficult question. Life expectancy in PKS has not been formally looked at but what we do know is this: There seems to be two or three main time points to consider. For the very severely affected newborn with a diaphragmatic hernia or severe congenital heart defect or other severe structural or functional anomaly this will have the most significant effect on morbidity and mortality and many newborns succumb early from these congenital differences.

After that I would say the most concerning issues that impact life expectancy are undiagnosed/untreated medical issues, such as reflux, malrotation of the intestine or seizures. We advocate for aggressive clinical evaluation's for these issues and if present for aggressive treatment as this will not only improve health and life expectancy but also optimize learning and happiness for the children (any parent should understand the impact that untreated gastroesophageal reflux, for example, can have on a child resulting in constant/recurrent pain and discomfort, risk for aspiration and damage to the esophagus).

For the child with PKS who has overcome these neonatal issues and is being managed effectively for the commonly associated medical issues, lifespan should be long or close to normal. For any individual with chronic medical issues like seizures or cognitive impairment there is an overall impact on life expectancy but not a large one. We know of some individuals with PKS in their 40s and 50s, and it is likely that there are many older individuals out there who have never been diagnosed since the type of testing available in the past decade or so is primarily used in the pediatric setting.

Like most issues concerning rare diagnoses like PKS, the primary point of entry for us to learn about these issues is through the parents and support groups like PKS Kids. It would be great for parents to report the ages of their kids as well as for parents who have lost a child with PKS to report how old their child was when they passed away and what the cause of death was. This could form the initial database that can eventually be used to better answer this question as well as to help identify causes of death in individuals with PKS so that we can better identify those problems early and avoid bad outcomes.