

Hello Chair Griffith and the and the Senate Finance Committee. My name is Kristin Anzelc and I was born May 2, 1985 in Austin, Texas. My mom reported that I had a normal birth - much like my neurotypical Brother and Sister (I am the Middle of 3 kids). However, immediately after birth I was sent to the NICU for 6 weeks. I never really learned how to latch correctly, so had to use a special bottle (could never be breastfed). I was said to be neurologically at-risk and Failure to Thrive. I was always considered a mystery to doctors.

Fast forward to 1995 when we moved to Baltimore, Maryland for my dad's job. Due to us wanting answers we were immediately followed at Johns Hopkins. May 1996 I will forever remember this weekend. I was attending a Camp sponsored by Hopkins and it was at this Camp where I met Dr. Eric Wolfsburg and where I first heard of Kabuki Syndrome. I had many of the classic symptoms and there is only 100 Cases in the World (at that time – now it is thought that 1:32000 births are affected). From 1996-2002 I saw Dr Wolfsburg but in 2002 we stopped due to nothing new ever happening.

From 2002 - 2012, a lot happened to me medically and personally. I was still being followed for my ITP (diagnosed in 2001 and resolved in 2003). I also had 3 knee surgeries (2002, 2003, and 2005). Between 2002 and 2012, I managed to graduate from High School, attend Community College, AND Graduate from Stevenson University.

July 2012, my mom hears from someone in her Pilates class at our local gym that Johns Hopkins (Hopkins) was doing the testing on the specific genes for Kabuki now that they had been identified in 2010. We immediately got an appointment with Genetics at Hopkins. The new geneticist asked us who my Immunologist was and I had none, but my allergist did the Testing of my Immune System – and the results that came back from that testing were surprising - it showed that I had no Immune System (which was when we were followed by an Immunologist at Hopkins - which was the beginning of my CVID Diagnosis – a serious immune issue). We realized after the testing of the Immune System that is why I was constantly getting sick as a child – but no one had ever even THOUGHT to check my Immune System until this Geneticist asked us that question/ Additionally, I had the Genetic Testing done to confirm the Kabuki diagnosis and I tested positive for type 1. Additionally, Kabuki can lead to other

health issues - like heart conditions, orthopedic issues, low muscle tone and sometimes problems with communication. Those with Kabuki have mild - extreme cognitive delay (mine is more mild - my cognitive age is very similar to my biological age but some with Kabuki struggle more cognitively).

Additionally, those with Kabuki can have breathing issues. I remember in 2013, I was having issues with breathing, so my Immunologist connected me with Pulmonology at Hopkins. I had various tests done that Summer and into the Fall but all that showed us NOTHING. November 2013 I had a standard follow up with my Pulmonologist and at this point I was very close to death (due to not being able to get a deep breath and being sick for a month). From his office he admitted me to the Hospital in Critical Condition. After a few more days of testing (and me being on IV Antibiotics) my doctor made the decision to put me on Rituximab. This was a battle between my Pulmonologist and the "insurance company". Every day we were hearing that "the insurance company will not approve it" (and this is AFTER my dad had talked to the person at the Insurance Company - he negotiated the contracts for the teachers for his job AND the Insurance Company had talked to the doctor directly at that point). We found out later that it was Hopkins Pharmacists that did not want to approve it, but ultimately it was approved.

May 2015, I attended the first ever Kabuki Research Conference held at Hopkins (where I was also on the Young Adult Panel). This was my first time ever meeting others with Kabuki. 2 years later we heard that 2 adults I had met at the Kabuki Conference in 2015 had passed away due to Lung Issues. I know had I not been where I am at – there would have been a 3rd – ME. I consider myself lucky that I live about half an hour from Baltimore – and therefore close to many prestigious medical centers (Johns Hopkins and University of Maryland).

Much of the time those with Kabuki can have a shortened lifespan because of all we go through. I consider myself lucky that my dad's previous company kept agreed to allow me to have my own Insurance through them because of being a Dependent with Disabilities. Having been a Warrior from birth till now and going through multiple medical issues - I have seen how incredibly Vital Healthcare access is. Today I ask you to support a rare disease advocacy council

to support both myself and others like me with a Rare Disease. Thank you for your time.