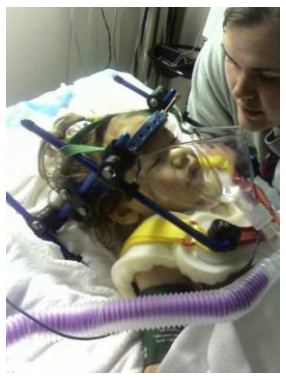
JOSSLEN'S JOURNEY

Klippel-Feil Syndrome – (N.O.R.D.)

Klippel-Feil syndrome (KFS) is a rare skeletal disorder primarily characterized by abnormal union or fusion of two or more bones of the spinal column (vertebrae) within the neck (cervical vertebrae). Some affected individuals may also have an abnormally short neck, restricted movement of the head and neck, and a low hairline at the back of the head (posterior hairline). The disorder is present at birth (congenital), but mild cases may go undiagnosed until later during life when symptoms worsen or first become apparent.

KFS can be associated with a variety of additional symptoms and physical abnormalities. Including: scoliosis spina bifida occulta, raised scapula (Sprengel's deformity), rib defects, skeletal malformations of the ear, nose, mouth and larynx, hearing impairment and cleft palate, malformations of the head and facial (craniofacial) area; anomalies of the urinary tract and/or kidney or structural abnormalities of the heart, webbing of the digits and digital hypoplasia. In addition, in some cases, neurological complications may result due to associated spinal cord injury.

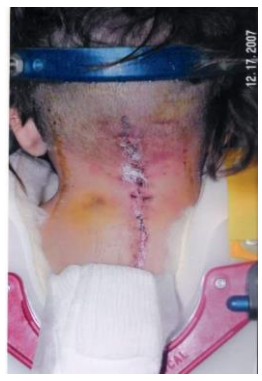
KFS was originally described in the medical literature in 1912 by doctors Maurice Klippel and Andre Feil. The exact incidence of the disorder is unknown, although reports estimate that the condition occurs in approximately one in 42,000-50,000 live births.



Yes, you read that correctly, One (1) in 42,000-50,000 live births.

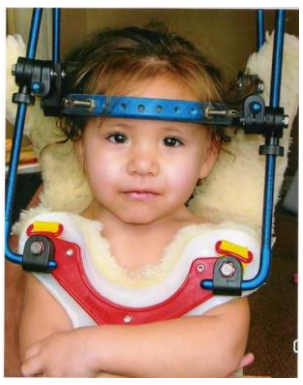
They key words in that sentence are “one” and “live”. (**Live**) Most babies born with KFS are lost upon delivery due to those spinal bone fragments severing the spinal cord or brain stem. (**One**) The miracle, the lucky, the gift, Josslen.

In 15 years, I can count on just one hand, the number of people who have ever heard the words Klippel-Feil Syndrome that didn’t need an explanation on what it was. Don’t worry, I didn’t know either until 2007. Joss was diagnosed with KFS when she was 23 months old. Though there were signs prior to her official KFS diagnosis. Corrective eye surgery at 12 months old. Oral surgery at 14 months old because her teeth would not erupt. Fused Ribs. Missing ribs, heart murmur. But it wasn’t until we noticed a lump starting to form between her neck and shoulder that we insisted on further testing.



What do you do when you are given a diagnosis that you have never even heard of – Research and Pray. Visit after visit, different doctors, different hospitals, different specialist, same response to my question “Have you ever dealt with this condition or performed the type of required life-saving surgery?” – NO!

That is until October 2007. A drive from Michigan to Illinois. The destination address 2211 N. Oak Park Ave, Chicago, IL. The name on the outside of the building **Shriner’s Hospital for Children**. The Chief o Staff, Dr. Purnendu Gupta. The very last time I had to ever ask “Have you ever dealt with this condition or performed the type of required life-saving surgery?”. The answer was yes. Only not just yes, he said “Yes BUT…..I feel that a partner, another surgeon, would be able to perform this required surgery better than I can.” What? Are you kidding me? A surgeon humble enough to say that another surgeon may be better than himself, to say without so many words that my child is more important than his pride. Within the hour, we met Dr. Steven Mardjetko for the first time.



Surgery was scheduled for December 12, 2007. Josslen had just turned 2 on October 13, 2007.

There is no amount of research, talking, praying, crying, wishing that can actually prepare you for the day of surgery. Surrounded by the love and support from all the most important people in my life and I still found it hard to breath for those 8 hours of surgery. A complete fusion of her upper neck, 4 screws into the bottom of her skull, 2 metal rods that protected that fusion going down her back and 6 screws into her spine, Halo in place.

It was almost Christmas and I had just received a Christmas miracle.

For this 2-yr old and all the other patients there during this timeframe, regardless of their condition, pre or post-surgery, those donated Christmas gifts gave each of them a reason to smile and just that smile, gave each of those parents a chance to stop holding their breath, if even for just a moment. Trust me, it matters. Your donation, your gift, your toy, however big or small, matters.

Josslen’s Journey has been heartbreaking and breathtaking all at the same time. The resilience, the courage, the determination of this, now, 15-year old is nothing short of a miracle. Josslen refuses to let KFS define her and instead challenges herself, pushes the boundaries, and embraces each one of her Perfect Imperfections!

My favorite part of it all……
tell her she “can’t” and watch her prove you wrong.