Fact Card about Cri du Chat Syndrome:

• Cri du Chat Syndrome, also known as 5p- (Five P Minus) occurs when there is a loss of genetic material on the short arm of the fifth chromosome.

• It’s main characteristic is the kitten-like cry (soft cry) that is due to a underdeveloped larynx. As the child grows and the larynx gets stronger the sound slightly deepens;

• Other characteristics may include low birth weight, small head circumference, small chin, low set ears, epicanthal folds in the inner corners of the eyes, broad nose, low muscle tone, cognitive delays, and expressive speech delays.

• This rare genetic disorder affects 1:50,000 births. Most individuals are diagnosed at birth because providers notice the main characteristics; but some individuals do not get diagnosed until later because parents and/or providers investigate the reason for their child’s developmental delays.

• Cri du Chat Syndrome was discovered 50 years ago (1963) by Dr. Jerome Lejeune in France.

• Most cases of Cri du Chat Syndrome are “sporadic” or “de novo” which means it just happened. However in 10% of the cases the occurrence was an unbalanced translocation caused by a balanced translocation rearrangement of a parent’s chromosomes.

• Most individuals with Cri du Chat Syndrome will have a normal life expectancy.
• Individuals with Cri du Chat Syndrome are on the Mild to Severe spectrum with regards to development delays.

• Individuals tend to have a slow growth rate. On a typical growth chart you will most likely find an individual with Cri du Chat Syndrome in the 50th percentile or lower. Because of this slow growth many individuals are fed by g-tube. Many individuals also have reflux and aspiration during feeding and can account for slow growth.

• Due to the low muscle tone and cognitive delays, most children with the syndrome will be delayed in reaching their physical milestones of turning over, sitting, crawling and walking. Some will not be able to accomplish these physical milestones.

• Due to the cognitive delays and expressive speech delays, children with the syndrome may not be able to speak verbally or may be considered unintelligible. Those who do not acquire vocalization often times communicate with sign language or an augmented device.

• Problem behaviors are associated with the syndrome. Some behaviors include overactivity, restlessness, impulsiveness, inattentiveness and distractibility. It is very common for individuals to hit, pinch, pull hair, kick and bite to get attention. There is a relationship between poor expressive communication and behavior problems.

• Receptive language is 1.5 to 2 years greater than expressive language.

• Many individuals with Cri du Chat Syndrome have Autism-like tendencies.

• In general, individuals with Cri du Chat Syndrome are happy, loving, sociable and friendly. Sometimes a little too friendly and tend get into other’s space. This could be misconstrued as an aggressive gesture.

• Safety education continues throughout life.

• Many individuals with Cri du Chat Syndrome have difficulty with sleeping.

• Some individuals with the syndrome have seizures from silent to Grand Mal.

• Respiratory illnesses including asthma like symptoms, frequent colds and ear infections are very common.

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