



5p- Syndrome Day May 5

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Fact Card about 5p- Syndrome:

- 5p- Syndrome (Five P Minus), also known as Cri du Chat Syndrome, 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 get diagnosed at birth because providers notice the main characteristics, but some individuals do not get diagnosed until later when they miss physical and cognitive milestones.
- 5p- Syndrome was discovered 60 years ago (1963) by Dr. Jerome Lejeune in France.
- Most cases of 5p- Syndrome are "sporadic" or "de novo" - which means it just happened. However in 10% of the cases, the occurrence was due to an unbalanced translocation caused by a balanced translocation rearrangement of a parent's chromosomes.

For more information please visit www.fivepminus.org or www.criduchat.org
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- Individuals with 5p- Syndrome are on the Mild to Severe spectrum with regard to developmental delays.
- Individuals tend to have a slow growth rate. On a typical growth chart you will most likely find an individual with 5p- 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 aspirate during feeding which 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 verbalization oftentimes communicate with sign language or Augmentative and Alternative Communication Devices
- 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 5p- Syndrome have tendencies that are often associated with Autism.
- In general, individuals with 5p- Syndrome are happy, loving, sociable and friendly; sometimes a little **too** friendly and tend to get into other people's space.
- Safety education continues throughout life.
- Many individuals with 5p- 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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