

# Glossary

## Phelan-McDermid Syndrome Related Medical Terms

<b>Arachnoid Cyst</b>	fluid-filled cyst in the brain
<b>Bitemporal Narrowing</b>	size of the head above the eyebrows is narrow
<b>Brain Atrophy</b>	part of the brain is underdeveloped
<b>Bulbous Nose</b>	tip of the nose is round or bulb-shaped
<b>Cellulitis</b>	inflammation or infection of the skin causing redness, swelling and tenderness
<b>Commando Crawl</b>	crawls using only the arms
<b>Cyclic Vomiting</b>	sudden repeated attacks of severe vomiting that can last from hours to days
<b>Deep Tendon Reflexes</b>	involuntary jerks produced when certain spots on the limbs are tapped with a rubber hammer
<b>Dehydration</b>	a significant loss of fluid (can result from diarrhea or vomiting)
<b>Delayed Myelination</b>	sheath that covers the nerve fibers did not form at the usual stage of development
<b>Dilated Renal Pelvis</b>	outlet of the kidney is enlarged where urine is passed into the ureter
<b>Dolichocephaly</b>	a long, narrow head
<b>Down slanting Palpebral Fissures</b>	the eye opening appears slanted downwards from the inner corner of the eye to the outer corner of the eye
<b>Dysplastic Ears</b>	ears seem simple, underdeveloped, or the upper part of the ear does not seem folded over enough
<b>Dysplastic Fingernails/Toenails</b>	underdeveloped, thin, easily peel off
<b>Enlarged/Dilated Ventricles</b>	cavity within the brain is enlarged
<b>Epicanthal Folds</b>	fold of skin covers the inner corner of the eye
<b>Facial Asymmetry</b>	right and left sides of the face seem out of proportion to each other
<b>Fifth Finger Clinodactyly</b>	pinkie is slightly bent inwards toward the ring finger
<b>Flat Midface</b>	middle part of the face below the eyes and above the mouth seems depressed or flattened
<b>Frontal Lobe Hypoplasia</b>	front part of the brain is underdeveloped

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<b>Full Brow</b>	forehead seems broad
<b>Gastroesophageal Reflux</b>	contents of the stomach flow back into the esophagus due to a muscle problem
<b>high arched palate</b>	The palate is unusually presented in a high arch
<b>Hyperextensible Joints</b>	joints move beyond normal range with little effort
<b>Hyperreflexia</b>	overactive reflexes, excess reaction of muscles when stimulated
<b>Hyperthyroid</b>	overproduction of the thyroid hormone by the thyroid gland
<b>Hypoplastic Cerebellum</b>	reduced number of cells in the cerebellum of the brain
<b>Hyporeflexia</b>	underactive reflexes, weak reaction of muscles when stimulated
<b>Hypothyroid</b>	insufficient production of the thyroid hormone by the thyroid gland
<b>hypotonia</b>	Decreased tone of the skeletal muscles characterized by weakness and “floppiness”.
<b>Impulsiveness</b>	acting out apparently without thinking of the consequences
<b>Lax Ligaments</b>	increased flexibility
<b>Long Philtrum</b>	vertical groove that goes from the middle of the upper lip to the middle of the nasal septum is long
<b>lymphedema</b>	The swelling of a body part caused by the abnormal accumulation of lymph fluid.
<b>macrocephaly</b>	Large head for which the cause may be unknown. It is not the same as fluid on the brain, which is typically called hydrocephaly and may require a shunt.
<b>Malocclusion</b>	teeth are not aligned properly
<b>Microcephaly</b>	small head size for age
<b>mild dilation of cerebral ventricles</b>	The ventricles of the brain are enlarged and may be caused by the collection of cerebrospinal fluid and may require a shunt.
<b>mild facial dysmorphic features</b>	This refers to minor facial anomalies that occur fairly frequently and don't pose a significant health risk, such as ptosis, epicanthal folds, or down slanting palpebral fissures. One of these features as an isolated finding in an individual would not be of concern since they occur fairly frequently in the general population. However, the presence of two or more minor anomalies may prompt a search for major defects and may aid in the diagnosis of a particular syndrome.
<b>Polycystic Kidneys</b>	kidneys are enlarged due to the presence of multiple cysts

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<b>Precocious Puberty</b>	early development of mature sexual characteristics such as pubic hair, breasts etc.
<b>Prominent Ears</b>	ears seem large or stick out from the head
<b>Prominent Nasal Alae</b>	flared nostrils
<b>Ptosis</b>	droopy upper eyelids so the eyes appear partially closed
<b>Sacral Dimple</b>	dimple at the top of the tail bone
<b>simian crease</b>	A single crease (line) along the middle of the palm of the hand. Usually there are three major creases on the palm - a crease that goes from the pinkie side of the palm and curves up toward the index finger, another crease that starts on the pinkie side of the hand and goes between the index finger and the thumb and the third crease that goes from above the thumb toward the wrist. In individuals with a single palmar crease, there is a straight line across the palm – It was initially called a simian crease because it was thought to resemble a monkey’s hand – now that term is insensitive. This feature is now commonly called a single palmar crease.
<b>Single Palmar Crease</b>	one crease extends across the palm of the hand instead of the three creases that are usually seen
<b>Skin Tags</b>	small, fleshy growths on the skin
<b>Sleep Apnea</b>	temporary interruption of breathing during sleep
<b>speech difficulties</b>	Growling, low guttural utterances
<b>Strabismus</b>	wandering eye, the eye seems misaligned
<b>syndactyl</b>	webbing, or an extra fold of skin between the digits – in 22q13 deletion, we sometimes, but not always, see webbing between the second and third toes.
<b>Tactile Sensitivity</b>	sensitive to the touch
<b>Tongue Thrusting</b>	pushes the tongue against the teeth when swallowing
<b>Two-Thirds Toe Syndactyly</b>	webbing or extra fold of skin between the 2nd and 3rd toes
<b>Upslanting Palpebral Fissures</b>	the eye opening appears slanted upwards from the inner corner of the eye to the outer corner of the eye
<b>vesicoureteral reflux</b>	(VUR) refers to a condition in which urine flows from the bladder, back up the ureter, and back into the kidneys.
<b>Wide Nasal Bridge</b>	the bridge of the nose seems broader than usual