

Caregiver's Guide

*Raising an individual with
5p- and Cri du Chat Syndromes*



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Introduction and Acknowledgments

Caregiving for a child with intellectual and physical disabilities is not an easy task. Caregiving for a child with a genetic disorder that you've never heard of, and is so rare that your doctor has never seen a child with it, can leave you overwhelmed and seeking out answers to questions that are hard to find.

The 5p- Society has created this Caregiver's Guide to assist you in raising your child or adult with 5p- Syndrome and Cri du Chat Syndrome. This guide is dedicated to Kent Nicholls and the original founders of the 5p- Society.

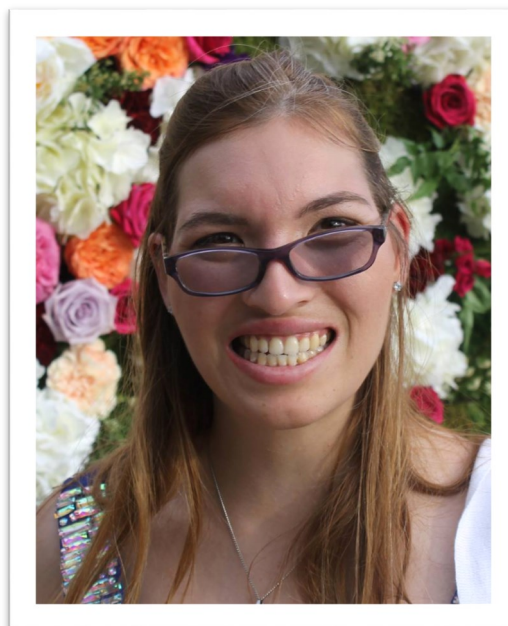
I wish to thank the over 100 families who participated in the grueling questionnaires that provided the basis of each of the chapters in this guide.

I would also like to thank my daughter, Katie, who provided me with the true meaning of unconditional love and without her I would not have met, counseled, or assisted hundreds of families around the world.

This guide is a living document. As the world around us changes, so does caring for a individual with 5p-, with new therapies, educational opportunities, research and medical advances.

Warmly,

Laura Castillo
Executive Director
5p- Society



"Having a family member with 5p- syndrome is a challenge and an opportunity not unlike supporting any individual in your family. You must accept them as they are; learn about them as an individual; nurture their strengths; find help for their weaknesses; encourage their growth; and love them unconditionally. They will not be what you want them to be. They will be the best of what they can be. They will definitely be a blessing."

Kathy Rapp



Inside this guide

As you read through this guide, keep in mind that no two children born with 5p- Syndrome(5p-)/Cri du Chat Syndrome (CdCS) are alike. There will be similar characteristics. We can put ten children with the same breakpoint in a room together and you can see the similar characteristics, but you will also see individuals all over the spectrum, from severe to mild. Over 100 families responded to five questionnaires. The responses have been thoroughly reviewed and turned into a working guidebook.

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What is 5p- Syndrome?

Simply put, 5p- Syndrome is a deletion (-) of the short arm (p) of the fifth (5) chromosome. Each of us has 46 chromosomes, two of each chromosomes 1 through 22. The 23rd set of chromosomes is the sex chromosome. Two "x" chromosomes is a female and one "x" and one "y" is a male. Chromosomes make us uniquely who we are. One set of chromosomes comes from the father and one from the mother. Upon conception and first cell division in utero an unusual mishap occurs that causes 5p-. There are several hundred genes on the "p" arm of the 5th chromosome. Genes provide the instructions for making proteins in our bodies. (To understand more about the genes involved in the 5p chromosome please read *5p Deletions: Current Knowledge and Future Directions*, Nguyen JM, et al. American Journal of Medical Genetics Part C semin Med Genet 9999:1-15 (2015))

In 90% of the cases, there is no known reason for this to happen and is called a "de novo" or unfortunate occurrence. **Please keep in mind there is nothing that you did wrong as a parent for this to happen — it just did.** Statistically, one in 100 individuals are born with a genetic disorder.

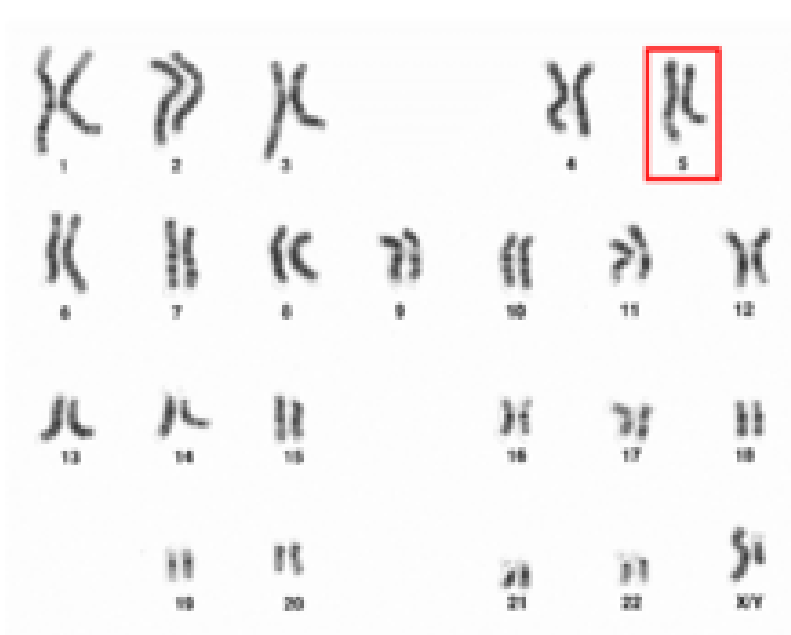
In 10% of the cases, one of the parents has a **balanced translocation**. This means that the parent had the 5p- deletion, but the deleted material was attached to another chromosome. Upon conception the 5p- portion of that parent's chromosome and the deleted material did not continue together and therefore the child was born with an **unbalanced translocation**. In most cases, parents with a balanced translocation are unaware that they have it until they have a child born with 5p- and undergo testing. If a parent has a balanced translocation they have a one in four chance of having another child with 5p-. (As a side note, of the over 100 families who participated in the Caregiver's Guide questionnaires, 10% reported that their child had an unbalanced translocation.)

There are other types of 5p- deletions that are extremely rare:

Mosaicism—is when the deletion occurs later in the cell division process and therefore not all cells in the body are affected by the deletion.

Ring Deletion—is when both arms of the deleted chromosome reunite.

Interstitial Deletion—is a 5p- deletion but occurs within the 5p chromosome and does not include the terminal end of the chromosome.



Cri du Chat Syndrome

Cri du Chat Syndrome was diagnosed in 1963 by Dr. Jerome Lejeune, a French researcher who also recognized Down Syndrome. Cri du Chat (pronounced “kree do shaw”) is French for Cry of the Cat. Dr. Lejeune recognized this characteristic in three patients at an institution. Further research revealed that the area on the 5p chromosome responsible for this characteristic is found at the 5p15.3 band. Cri du Chat Syndrome is diagnosed when the child has a deletion that includes the 5p15.2 band. This band also includes the CNNTD gene known to be responsible for intellectual delays. A child can have a 5p– deletion and not have Cri du Chat Syndrome if the deletion does not include the 5p15.2 band (i.e. 5p15.31). Most 5p– deletions are terminal, which means that the deletion starts somewhere within the 5p region to the terminal end. The end point of the 5p chromosome is 5p15.33.

Diagnosing 5p– Syndrome

5p– Syndrome can only be diagnosed by either a blood test or saliva swab. It is not discovered prenatally through routine test such as a CVS or ultrasound. It can be diagnosed prenatally by an amniocentesis or through new specific testing done by blood tests. Of those individuals who participated in the questionnaires, only 2% of the parents knew about the diagnosis prior to giving birth.

There are several tests that can be ordered by your doctor to correctly diagnose the syndrome. The first is a simple blood test called a karyotype. Although not used very often today, the test was used extensively from the 1960’s to early 2000’s to diagnose genetic disorders. Then came the FISH test that looked at the chromosome through a high powered microscope and specific dyes. Both the karyotype and the FISH provide a diagnosis to a specific band on the “p” arm. Currently all testing is done by what is called a microarray. The microarray is able to not only detect the deleted band chromosome but can detect the deletion down to a specific location on a band.

Of those who answered the questionnaires, the average age of a child when he or she was diagnosed with the syndrome was 3 months. Diagnosing was as early as right after birth to 10 years of age. We do know that we have individuals in the 5p– Society who have been diagnosed as early as prenatally and as late as in their 40’s or 50’s. You might ask how that can be, diagnosed so late in life. Keeping in mind that Cri du Chat Syndrome was not officially named until 1963, there would be quite a few individuals with developmental disabilities who never had a “diagnosis” until such time that they had genetic testing done for any particular reason. Many adults are diagnosed after they have given birth to a child who has the syndrome.

It is very important for parents to be tested after the diagnosis of their child with the syndrome. This is important because it will rule out a translocation and for peace of mind that future pregnancies will not be at an increased risk of having another child with the syndrome. Parents can then make informed decisions about future family plans.

Is there a cure for 5p– Syndrome?

Unfortunately there is no cure for 5p– Syndrome. Because of the dynamics of the “p” arm of the chromosome and the different genetic make up of each individual, it would be almost impossible to re-create the missing piece. Research is being sought after though to see if amino acids or other nutrients can be provided to a child with the syndrome to assist with improving the prognosis and cognitive development.

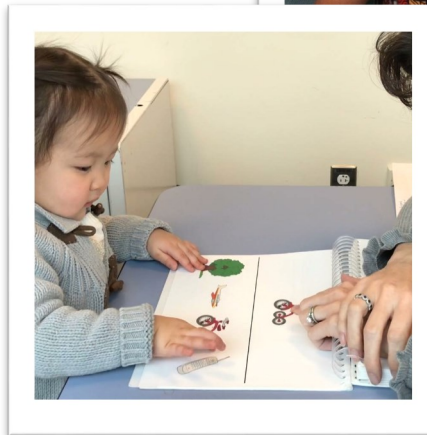
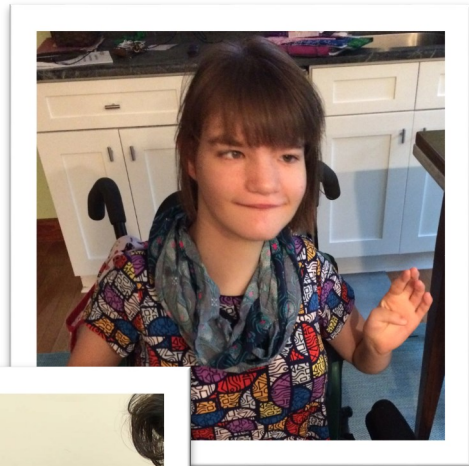


Characteristics

The main characteristic of Cri du Chat Syndrome includes the high-pitched kitten like cry that occurs in infancy. This too has been found on the 5p15.2 band and is caused by the under-development of the larynx.

Other characteristics include:

- ◆ Small head circumference (microcephaly)
- ◆ Small chin
- ◆ Wide-set eyes
- ◆ Flat nasal bridge
- ◆ Epicanthal folds in corner of eyes
- ◆ Skin tags
- ◆ Low set ears
- ◆ Cleft palate
- ◆ Laryngeal cleft
- ◆ VSD or ASD
- ◆ Low muscle tone
- ◆ Low birth weight
- ◆ Dislocated hip
- ◆ Constipation issues



Based on the answers from the questionnaires received, 90% reported that their child had the high-pitched cry, small head circumference, low muscle tone, low birth-weight (average 5lbs 10oz) and feeding difficulties; 70% of the children had low set ears; 25% of the children had skin tags; and 10% were reported to have seizures. Other characteristics noted on the questionnaires, not listed above: simian crease on the palm of the hand, club foot, knuckle displacement on hands and a stridor when breathing.

The breakpoints of the participants were reported all throughout the 5p chromosome; however, 50% of the deletions were in the range of 15.3 to 15.1. There were two reported interstitial deletions and seven unbalanced translocations. Of the unbalanced translocations, the additional affected chromosomes were 7p, 6p, 17q and 22q (and a few unknowns) and the father had the balanced translocation in 5 of the 7 occurrences.



Medical Considerations

Parents responded to questions about their child's medical history. They reported that approximately 60% of children born with the syndrome required surgery to repair one or more of the below medical issues between the ages of birth to three.

Height & Weight: <5% of normal growth chart; slow growth (please inquire with the 5p- Society for growth charts specific to children with 5p- Syndrome)

If you see a * next to any series of words, it means "very frustrating parents" moment can occur.

Head, Mouth, Neck & Throat

Head: Small circumference;

Most children born with the syndrome will have a smaller than normal head circumference. The pediatrician will continue to monitor this and take measurements at each visit. He or she is measuring to make sure there is growth occurring.

Eyes: Strabismus, low vision, visual corrective devices (glasses) for both nearsighted and farsighted, blindness

Children with 5p- Syndrome have low muscle tone (see below) and this includes the muscles of the eyes. Some doctors will patch one eye if it is found to be weaker than the other eye. Many children with 5p- Syndrome do wear glasses; as young as 1 year of age. It is difficult to keep glasses on a young one and be prepared to replace them often.* You will eventually find the right fit for your child. Some parents reported that the flexible frames and wrap around the ear arms work the best for the young children. Very few children with the syndrome have been diagnosed with blindness.

ENT: Ear infections, eustachian tubes, congestion, sinusitis, laryngomalacia (stridor), laryngeal cleft, adenoidectomy, tracheostomy, paralyzed vocal chords, swallowing issues, constantly sick

All children get sick; unfortunately, children with 5p- Syndrome get sick more often than the average child. Ear infections tend to happen a lot and because of this tubes are generally put into the ears to assist with draining of excess fluids. As the child grows, the tubes are replaced (for larger tubes) until such time that the child no longer needs the assistance of the tubes. Along with the ear infections are sinus infections and congestion. The use of a dehumidifier can help along with steaming. Doctors may also allow the use of decongestants (over the counter). You may hear your child breathe with what is called laryngomalacia or a stridor; this is very common in children with the syndrome. Laryngomalacia is a congenital softening of the tissues of the larynx (voice box) above the vocal cords. This is the most common cause of noisy breathing in infancy. The laryngeal structure is malformed and floppy, causing the tissues to fall over the airway opening and partially block it. This does tend to go away as the child grows. Please do have an ENT check to make sure that the vocal chords are developed correctly. Some children have paralyzed vocal chords. Laryngeal cleft can also occur and can be repaired. In some instances children with the syndrome have had their tonsils and adenoids removed. Swallow tests are also done if a child is unable to properly swallow food because he or she cannot tear the food down into a manageable bolus. In some instances children with the syndrome are unable to breathe properly and a trach is needed to be inserted into the windpipe. This can be very scary for a new parent. We have a handful of parents who are willing to talk with you about the procedure and caring for a child with a trach.



Mouth: Cleft lip or cleft palate, small jaw, crowding of teeth, teeth misalignment, overbite, grinding of teeth, brushing issues,* dental cleanings, dental surgeries, braces

If a child is born with a cleft lip or cleft palate, the doctor will discuss with you the management and repair of the cleft. Children with 5p– Syndrome usually have a small jaw, and because of this there will be crowding of the adult teeth (not seen as much in baby teeth).

Make sure to take your child to the dentist early on to help alleviate the fear of dental work. However, approximately 50% of children with the syndrome do have dental cleanings done under anesthesia in the hospital for specific reasons. You will know after a few visits with the dentist if your child can tolerate regular visits or if he or she will need to be sedated. Many children with 5p– Syndrome have had extractions done of their adult teeth to relieve the overcrowding. Only about 10% of children with the syndrome have had braces to correct their overcrowding and have done so quite well. The children tend to be older when they get braces.

Children also tend to put their hands or items in the mouth and drool a lot*. They could be sensory seeking. For extensive drooling, some children have had surgery to correct.

Heart & Lungs:

Heart: VSD, ASD, PDA, heart murmur, tetralogy of fallot

The above listed conditions VSD, ASD and PDA might be found on a newborn with 5p– syndrome. Very few of the children required surgery. In most cases the condition healed itself with time. Pediatricians will make sure that they are constantly monitoring the heart.

Lungs: Primary Ciliary Dyskinesia (PCD); chronic pneumonia, bronchitis, asthma

In 2014, a discovery was made that *some* individuals with 5p– were co-diagnosed with PCD. PCD is found on the same band that Cri du Chat Syndrome is located: 5p15.2 (DNAH5). Chronic lung infections and sinus infections contributed to the diagnosis. The only way to truly diagnosis this is to have a sampling of the ciliary in the lungs. PCD will not go away and is a lifelong and chronic disorder. It is also very disabling and can cause a shortening of life expectancy. If your child has constant lung infections including pneumonia and sinusitis, it would be the recommendation of the 5p– Society to question your doctor about possible link with PCD. Likewise, many children with 5p– Syndrome also have severe asthma and require daily inhaler treatment or nebulizer treatments, especially when sick with a lung infection.

Gastroenterology:

Gastrointestinal: Reflux, gagging, vomiting, constipation, hernia, colic, j-tube, g-tube, ng-tube

Just about every child with 5p– has some sort of gastrointestinal problem. Most common and occurring in approximately 75% of the children is constipation*. Parents have tried just about every over the counter medication, holistic cure, therapy treatment (for example massage therapy) or prescription to assist with the movement of the bowels. Keep in mind that the child has low muscle tone and the intestines are a muscle. This issue continues into adulthood. It is very common, although not desirable, for an individual with the syndrome to have a bowel movement every three days. You will eventually learn the habits of your child. Of course we don't want your child to be in pain or for the constipation to cause other medical issues, so it is always wise to tell your pediatrician about the issue.

Gagging and vomiting, although not as common as constipation, can occur. A lot of times the gagging is a sensory response. Vomiting could be more serious and should be brought to the attention of your doctor. Reflux is common with children who have the syndrome and can be dangerous. If you feel that your child has reflux please ask the doctor to do some tests to make sure that fluids are not going into the lungs.

A large number of children are unable to take any foods by mouth because of swallowing issues or vomiting. A pediatrician will make a recommendation to see a feeding specialist to see if there is something that can be done to enhance the child's feedings. If feedings are not tolerable, then the doctors will encourage a feeding tube. There are



different types of feeding tubes: ng tube, g-tube and j-tube. If you have questions about any of the “tubes,” the roles that each of them have and how children with the syndrome do with the tubes, please feel free to contact us and we will get you in touch with one of the parents. There is also a really great organization that you can connect with that can assist with any questions you have about the different types of tubes called Feeding Tube Awareness.

Extremities—Hands & Feet:

Hands: Small, simian crease, knuckle misalignment, weak, cold, blue

Children with the syndrome tend to have poor circulation in the hands and feet and you will notice your child’s hands colder than usual and maybe turning blue at times. You will also notice that they have a difficult time picking up items with their pincher grasp and will instead sweep the item with their hand to pick it up. Make sure that when you talk with the occupational therapist that they work with you on improving their grasp. Many children with the syndrome also have a difficult time with writing skills. They may not like to color or hold a crayon or pencil. Repetition, encouragement and trying different sized items can assist with this. Do not become too discouraged by the lack of ability to use their hands correctly; this is very common and your child will figure out what will be his or her best way to do things. A lot of children do learn to fasten a button and a snap, zip up a zipper and tie their shoes.

Feet: Small, pronation, flat feet, club feet, weak, cold, blue

Children with the syndrome will also have quite a few issues with their feet. Pronation tends to be one of the main issues. Many of our children will need to wear some form of orthotics to assist with keeping the ankles strong and maintaining good posture for standing and walking. Few children with the syndrome are born with club feet and/or flat feet.

Muscular:

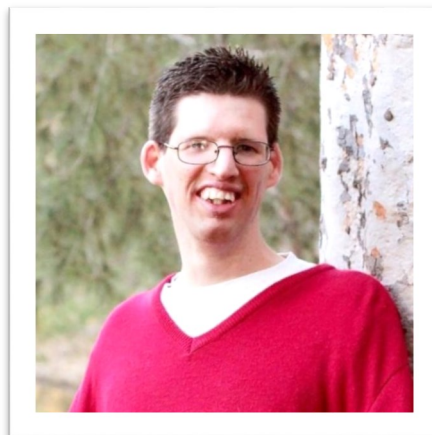
Low muscle tone:

Poor suck, tires easily, dislocated hips, floppy doll, slow reflexes, weak head control, scoliosis, delay in rolling over, delay in sitting, delay in crawling, delay in pulling to stand, delay in standing and delay in walking. Delay in potty training.

Low muscle tone includes both the large voluntary muscles in the legs, buttocks, abdomen, neck and arms, but also includes the involuntary muscles of the tongue, esophagus, vocal chords, stomach, intestines, lungs and heart.

Don’t be discouraged by all the delays!! Just know that there will be delays. Once a child learns the skill, or gets strong enough to complete the skill, he or she will better and better at the skill every day.

Your child will engage in a lot of exercises with his or her occupational and physical therapists. Do your homework and continue to work with your child when you can. Core muscle exercises will help with sitting, standing and walking.



A few words from our parents

"Trust yourself, fight hard, and never underestimate your child."

"It is difficult, yet rewarding. I have seen it develop the characters of her siblings--they are more patient, less judgmental, and more loving than their peers. I have days where I mourn the life she (and I) would have had, but there are less of those than the ones where I revel in her joy and kisses and complete lack of guile."

"You make the best decisions you can at the time with the knowledge you have. Don't get upset with yourself years later when you think well I didn't do XYZ therapy. At the time you did what you could do."

"Remember families hold the long view. Just because a therapist or teacher when your child is six says you should do XYZ, you have to decide if it is the right decision for your child. Teachers/therapists might be around for one year or a few years, but families are there for a lifetime."

"You have to do what is best for your child within the context of your family."

"This disability is a very small part of your child. Do not let it define their world or yours."



Therapeutic Considerations

As soon as you get your diagnosis, contact your state agency that oversees your child's development and institute **early intervention** as soon as possible. Four months is a typical waiting period (each state is different).

PLEASE NOTE THAT EACH STATE IS DIFFERENT AND YOU WILL NEED TO DO YOUR DUE DILIGENCE TO FIND OUT WHAT YOU NEED TO DO TO GET THE SERVICES YOU NEED. Some parents paid out of pocket or used their private insurance to increase the amount of therapy that their child received. This is a personal choice to make. Many children with the syndrome only receive the state funded aid that they are able to get with much success. As with any program, you as parents or caregivers should work with your child as much as possible to assist the therapist in the child's development.

Your child will need the following therapies:

Occupational Therapy:

An occupational therapist should be able to work on feeding, holding a bottle, motor movement, fine motor and gross motor skills. Occupational therapists will also work on sensory issues that your child may or may not develop. You should request that they come to the house 2-3 times a week from birth (or onset of early intervention to 3 years of age). Some states will have in-home therapy until the child reaches 20 months and then will have you bring the child to a facility for more structured learning and to get him or her ready for preschool.

Physical Therapy:

A physical therapist will work on gross motor skills and strengthening of the muscles. Both physical therapy and occupational therapy should be done simultaneously for as long as you can obtain these services.

Speech Therapy:

A speech therapist may or may not be needed at the earliest stage but introduced closer to one year of age. The occupational therapist can work on speech therapy as well and should be working on any feeding issues that your child is having. A speech therapists should work on feeding, mouth movement and breath control. It is the recommendation of the 5p– Society that you engage your child in sign language as soon as possible to have a vehicle of communication. All three therapists should also work with your child on sign language as the main form of communication until the child is able to produce sound. Not all children with the syndrome can talk or will develop speech. If you want to look into programs to assist with sign language, please visit www.signingtime.com. If you'd like to assist your child with breath control, please look into www.talktools.com. (Please note that the 5p– Society does not endorse these programs, but have many families who have had great success with these programs.)

As your child grows the therapy needs will change. You may begin to include:

Behavior Therapy

To help with:

- Sleep: difficulty staying asleep for long periods of time
- Arching back: could be caused by gastrointestinal discomfort
- Sensory input: sounds and touch tend to upset them.
- Vestibular movement seeking: head banging, twirling string
- Communication: child may bite, kick, hit, pull hair to get attention



Throughout your child's life you will learn about other therapy options. Parents who participated in the questionnaires reported that their children participated in the following therapies. Please keep in mind that what works for one child may not work for another. The 5p– Society does not endorse any of these therapies; they are listed for your reference.

Applied behavior analysis (ABA) Therapy

If your child has a dual diagnosis of Cri du Chat and Autism you can ask for an evaluation for your child to obtain ABA Therapy. The parents of children who have received this therapy are happy with the outcome. ABA is an applied science devoted to developing procedures which will produce observable changes in behavior.

Hippotherapy

A licensed, specially-trained physical therapist, occupational therapist or speech language pathologist may use a horse as a treatment tool within their therapy session, just as they might use a ball, balance beam, physio ball or toy. This helps to strengthen the child's core muscles. Many parents report that their child's mobility improves after this therapy.

Music Therapy

Music has been found to be an effective tool for music therapists through extensive research. It is beneficial for any individual, both physically and mentally, through improved heart rate, reduced anxiety, stimulation of the brain, and improved learning.

Behavior Modification Therapy

Behavior Modification Therapy is a therapeutic approach designed to change a particular undesirable negative behavior. By using a system of positive or negative consequences, an individual learns the correct set of responses for any given stimulus.

Cranial Sacral Therapy

Cranial Sacral Therapy, also known as craniosacral therapy, is a form of bodywork or alternative therapy using gentle touch to manipulate the synarthrodial joints of the cranium. A practitioner of cranial sacral therapy may also apply light touches to a patient's spine and pelvic bones.

Aquatic Therapy (our kids love the water)

Aquatic Therapy is physical therapy that takes place in a pool or other aquatic environment under the supervision of a trained healthcare professional.

Anat Baniel Method Therapy

Anat Baniel Method is a neuromovement method that helps increase strength and flexibility by using different movement exercises and the Nine Essentials. You can learn more about this therapy by going to www.anatbanielmethod.com.

Alternative and Holistic Treatments:

Essential Oils to assist with sleeping and behavioral issues

Coconut Oil to aid with constipation

Autism and 5p– Syndrome

It was discovered that one of the genes found on the p arm of the 5th chromosome also causes autism. As many of you know, children and adults with the syndrome do have autistic characteristics. It doesn't necessarily mean though that they have autism. If you feel that your child has autism, bring up the concern to your pediatrician for testing and confirmation of the dual diagnosis. If your child is diagnosed with both Autism and 5p– Syndrome, make sure that you have it documented and presented to the school/facility. This way your child can get all the services available to him or her with the autism diagnosis.



Prescribed Medications

Although it is not desirable to have to put your child on medication (for any reason), sometimes medications are needed to help the child manage his or her behaviors or other medical ailments. Below is a list of medications compiled by the parent reported surveys. Please keep in mind what works for one child may not work for another and that medications are constantly changing. Behavior medications can be for a variety of behavior issues, including depression, anxiety, OCD, social anxiety, panic attacks, skin picking, sensory issues. This list could grow old very quickly.

Abilify - behavior depression	Ethosuximide - seizures	Protonix - reflux
Adderall - ADHD	Ex Lax - constipation	Prozac - behavior
Albuterol - asthma	Eye drops - eyes	Pulmicort - colitis, ulcers
Allergy Medications - allergies	ferrum - iron supplement	Quillivant XR - ADHD (chewable)
Aloe Vera Capsules - constipation	Fluoxetine - OCD behavior	Qvar - asthma
Amitriptyline - behavior anxiety	Gabapentin - seizures	Ranitidine - reflux
Amoxicillin - infections	Gaviscon - digestion stomach	Ranitidine with gaviscon - severe reflux
Antibiotics as needed	Glycopyrrolate - peptic ulcers	Reglan - gastroenteritis issues
Ativan - seizures	Heparin - blood thinner	Remeron - depression
Atropine Sulfate Ophthalmic - eye	Hydrazine - sleep disorder	Restoralax - constipation
Atrovent - asthma	Hydrocortisone - itching and skin inflam	Risperadol - behavior
Axid - reflux	Hypertonic Saline - respiratory disease	Ritalin - ADHD
Bactrim - infections	Kepakote - bipolar (behavior)	Salbutamol - bronchial inhaler
Bethanechol - urinary/bladder problem	Keppra - seizure	Seroquel - behavior
Budesonide - colitis	Klonopin - behavior anxiety	Sertraline - OCD, social anxiety
Buspar - anxiety	Lactulose - constipation	Singulair - asthma
Buspirone - anxiety	Lasix - diuretic	Strattera - ADHD
Calcium - vitamin	Levobutanol - asthma	Tenex - ADHD
Carafate GI Distress - ulcers/reflux	Levothyroxine - hypothyroidism	tobramycin neb treatments - asthma
CBD Oil - behavior	Lexapro - behavior depression	Trazodone - depression
Celexa - depression	Losec - reflux	Trileptal - seizures
Cetirizine - allergies	Luvox - OCD behavior	Valerian Root - sleep disorders
Chlorpromazine - behaviors	Methlyphenidate - ADHD	Valium - behavior
Cimetidine - reflux	Methyl Folate - vitamin	Venlafaxine - behavior
Citalopram - depression	Mexium - reflux	Vesicare - bladder relaxant
Clonidine - hypertension/ADHD	Miralax - constipation	Vitamin B12 - vitamin
Codeine - pain/cough	Movicol - constipation	Vitamin B-6 - vitamin
Cogentin - anti-tremor	Mucomyst - cough	Vitamin D - vitamin
Cortoderm - itching & inflammation	Multi Vitamin - vitamin	Vyvanse - ADHD
Cyproheptad - antihistamine	Nexium - reflux	Wellbutrin - depression
daily vitamins - vitamins	Omeprazole - reflux	Xopenex - asthma
Depakote - seizure	Paroxetine - behavior OCD, anxiety	Zantac - relax
Dexedrine Spansules - ADHD	Paxil - behavior	Zoloft - OCD, behavior
Dextroamphetamine - ADHD	Penicillin - infection	Zyprexa - behavior
Digoxin - heart meds	Phenobarbital - seizures	Zyrtec - allergies
Dimetapp PM - cold/cough	Poly vi Sol - iron supplement	
Divalproex - seizure	Prevacid - reflux	
Doxycycline - infections	Prilosec - GERD, relax	
Dulera - asthma	Probiotics—constipation	
Erythromycin - infections	Propranolol - high blood pressure	



Educational Considerations

Will my child learn? What should I expect from my child and his/her instructors? What are goals? What is an IEP? A lot of questions will run through your head (and will be asked by your close family and friends). Having a child with a disability is tough. We are not going to sugarcoat it; it's tough. You have to not only figure out what is best for your child, but you will constantly be questioned about your decisions. You will even question yourself. Let's talk a little bit about educating your child with 5p– Syndrome. We will look at this in age groups.

Birth to Three

After the diagnosis of your child, you will want to start your child with the different types of therapies available. We listed those above in the *Therapeutic Considerations* section of this guide. From ages birth to about 2, therapists will most likely come to your house. The goals that you and the therapists come up with will be mostly strengthening and fine and gross motor skills. There will also be goals to improve cognitive and speech and language skills. When you talk with the therapists about the goals you'd like to see your child reach, keep in mind that they should be broken down into small steps. A goal can be as simple as tracking an object with the eyes. Therapists may only come to the home 1x or 2x's a week. It will be your responsibility as a caregiver to continue the steps to help your child reach his or her goals (i.e. do your homework). Repeat, Repeat, Repeat. Then Repeat again! What we have learned about our children is that they have great memories. The more repetitions that you have your child perform will hopefully help him or her be able to attain the skill you are working on.

From age 2 1/2 to 3, it may be the recommendation to send the child to an infant stimulation program at a facility. You will be responsible for transporting your child to the facility and many of these programs are "Mommy & Me" type where the parent and the child learn together. If your child receives Medicaid you may have transportation covered (you may want to check on this in your state/area). This can be tough if you have other children at home, unless they can provide child care for your other kids.

5p– Syndrome is a spectrum disorder. Children with the syndrome will be somewhere on the spectrum. The amount of the child's deletion most likely will not be able to tell you where on the spectrum your child will land. The attitude you take towards your child's development will assist in the overall understanding and acceptance of who your child becomes. Having the attitude that you will do whatever you can to help your child reach his or her maximum potential is a much better attitude to have than expecting that therapists and teachers teach your child how to be a scholar. Not only does this take a big weight off your shoulders and the shoulders of the teachers and therapists, but you can then appreciate the successes of your child, big or small, throughout your child's life. Also, don't think that your child cannot learn or cannot hear you (unless they are diagnosed with deafness). They CAN learn and they CAN hear you. They understand a lot more than you think.

Research studies show that a child with the syndrome has a higher receptive language than expressive language. Many of our children will not learn to actually talk or vocalize, but they are still learning and understanding things.

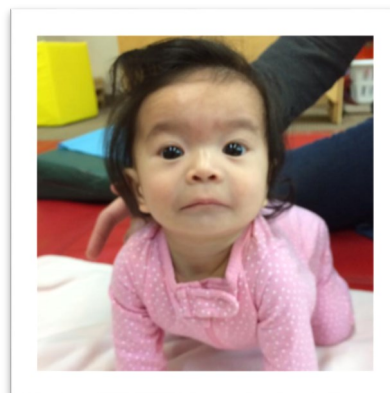
Children who are profound or severely affected by the syndrome will most likely have difficulty with fine motor skills, expressive language and mobility. They will be in diapers their entire life and probably in a wheelchair. They most likely will not be able to eat by mouth and require continuous feeds. They still need to have outside stimuli to continue to learn; however, it may be a very very slow growth. Try not to get discouraged. You are your child's best advocate. Don't stop requesting services. Many families who have a child that is profound-severe home school their child. Others continue to keep them in Special Education or Infant Stimulation programs for the profound-severe child. One thing they will always give you . . . Unconditional love.

Children who are mild-moderate on the spectrum will most likely have difficulty communicating their needs. It is imperative that speech therapy be continued as long as possible. Review and revise your child's goals to continuously create a positive experience for your child. If the goals are too hard to attain at the present time, your child may become defiant and discouraged causing undesirable behaviors to emerge. If your child attains a goal quickly, then add to it. Don't be too discouraged yourself if your child is having difficulty attaining a goal. Repeat, Repeat, Repeat and then Repeat again. It can take our children longer than expected to finally "get" it (developmentally delayed).



Reported goals: Birth to Three

- Tracking objects with eyes
- Responding to own name
- Making choices - choosing a toy preference
- Choosing drink over food for example
- Learn to sit unaided
- Improve upper body strength
- Lifting head while on tummy
- Signing for communication needs
- Drink from a bottle
- Learn to crawl
- Produce sounds
- Roll over



The parents were asked what types of obstacles they ran into that hindered their child's development from Birth to Three, and many parents reported illnesses, hospitalizations, surgeries, insurance, financial or time commitments (parents work full time).

When asking the parents if they would have done anything differently in those first three years, many parents reported that they wouldn't have changed anything. They felt that they did what they needed for their child at that time. There were a few parents who wished they would have started to homeschool their child at this time. Others felt that the goals that they wanted for their child were dismissed by the therapists. A few suggested to integrate the children with their typical peers in local activities instead of so much therapy. As you can see from these responses, there really isn't a right or wrong way to begin the process. It will evolve as your child grows.

Elementary School

As your child reaches the age of three, he or she will go into the public school system to continue his or her developmental needs, including, but not limited to, OT, PT, Speech and Language, Adaptive PE, Social Skills, and Self help Skills. Your case worker should assist you in the process. First you will need to register your child with the home school district by calling the district and telling them you need your child to be evaluated for special needs. You will have to produce documentation. You will get assigned a representative at the school district to help you navigate the system (at least you should be assigned a representative — you might have to ask for one). Your child will have to be tested in various areas (try not to take this too personal, they don't know who you are or who your child is — yet — they will eventually, but not at this initial stage). After your child is tested, you will be called into a meeting called an IEP (Individualized Education Plan). At this time the representative will go over the testing result and discuss a road map for you to navigate for your child and his or her future goals for the next year.

This is a great time for you to have your ideas on paper of what you think you'd like for your child to attain. The goals discussed are annual goals. No need to think too far into the future, just immediate goals you'd like your child to work on. You will also discuss at this time what environment will work best for your child: an integrated environment, a segregated environment, a combination of the two, or perhaps a program where the majority of the individuals have little to no mobility but they are cognitively higher functioning. If your child is cognitively higher functioning but lacks speech and language skills, then they have environments that work extensively on speech and language. Ask about all the different types of programs that are available. **DO NOT SIGN THE IEP YET.** Schedule a follow up IEP so that you can visit all the different programs of interest. Bring your questions to ask the instructor(s) that are of concern to you. Keep an open mind while visiting these programs. You will quickly weed out the ones that will not work for your child as you know your child best. Before reconvening with the group, discuss with the district representative who you'd like at the meeting, including the teacher of the classroom you have decided to enroll your child in. Be prepared, there will most likely be anywhere from 5-10 individuals at the meeting.

After introductions at the follow up IEP meeting, take the time to tell the group who your child is: the diagnosis, what you know about the diagnosis, your child's strengths and weaknesses, what makes him/her happy and what



can cause your child distress. Also talk about what you do in case of behavior meltdowns. Perhaps talk about a future behavior management plan if one is needed (most likely one will be needed). You should discuss what your immediate goals are and how they can align with the goals that will be presented in the meeting. If you don't agree with a goal or think a different goal would be better, discuss it. You can also add into the IEP that you'd like to meet in six months to discuss how things are going and if things need to be changed. Only sign the IEP if you are in complete agreement with it. If there is anything you don't agree with, don't sign it. Don't forget to discuss transportation. Also, don't forget to discuss the ability to visit the classroom and how you can help in the classroom. Teachers do like it when parents show an interest in the classroom.

After your initial IEP you will have an annual IEP with your child's teacher and usually a district personnel. You can bring anyone you want to the IEP, but you will need to let the teacher know ahead of time. Every three years (3 years old, 6 years old, 9 years old, 12 years old, 15 years old, 18 years old and 21 years old) the IEP is called a Tri-Annual IEP. These IEPs are longer than the annual IEPs because your child will be re-evaluated to check his or her overall progress. **PLEASE NOTE THAT YOU CAN CALL AN IEP MEETING AT ANYTIME YOU FEEL THAT THE NEEDS OF YOUR CHILD ARE NOT BEING ADDRESSED AS PER THE IEP OR IF YOU FEEL A CHANGE NEEDS TO BE MADE TO THE CURRENT IEP.**

Reported goals: Elementary

Cognitive Goals: recognize name, colors, letters and numbers. Ability to use electronic device such as iPad. Focus and attention, being able to perform a one step command. Squeeze toys, pull apart/push together toys, fingertip grasp, scribbling, increase stabilization/manipulation skills, imitate basic pretend play behavior, explore cause/effect toys, stack blocks, match objects, learning over, under, around, front and back.

Gross Motor Goals: floor mobility (crawling, rolling over, supine to sit), sitting unaided, balance, sit to stand, pull to stand, stand with assistance, stand unaided, walking with assistance, walking unaided, use of walker if needed, riding a bike, jumping, catching a ball, rolling a ball, and many others.

Fine Motor Goals: Raking of hands/fingers to move an object, pincher grasp, pick up food, grasp a marker, self feed, ability to use loop scissors, use of tongs, hold a pencil/pen correctly, attempt to write horizontal, vertical and circular lines, use a zipper, manipulate a button, thread a hole, pulling on pants, putting on a shirt, putting on socks, unscrew a cap, screw on a cap, place pegs in a hole, stack blocks, move items from one container to another, and many others.

Social Skills Goals: Engage in appropriate parallel play, make eye contact, being able to communicate his or her needs through gestures, and signs, vocalize appropriately to interact appropriately with his or her peers, spatial awareness, meaningfully point to choose between two pictures, understanding the concept of sharing, sit appropriately in class, church or a restaurant, learn to take turns, learning to keep hands to his or herself, play games and many more

Speech and Language Goals: Sign language, Picture Exchange Communication System (PECS), use of iPad, switches, produce or imitate developing consonants and/or vowel sounds, say one word consistently, say two words, say a short sentence, work with feeding, taking small bites of food, chewing food, and many more.

Parents reported some of the activities they did at home to help increase and improve their child's development:

- The use of the iPad for cause and effect, use of manipulatives, sand paper cut outs of letters and numbers
- Created a PECS, books, tried to continue the exercise we had learned in Birth to 3
- Shaving cream on the tile wall in the bathtub, speech therapy with animals and boats etc. in the bathtub, making the vowel and consonant sounds by giving the object a voice
- The use of flash cards at home, the use magnetic letters to help her form words on the refrigerator, the use of foam letters and numbers in the bathtub
- Practice writing, no letters but only lines; replacing the beans with a spoon from a one to another
- We worked on gross motor by setting up paths for her to walk in her walker. We learned basic sign language together to give her another method of communication. We worked a lot on puzzles and getting her to focus on one thing at a time.
- We would count and say the alphabet and use toys and puzzles. We watched Sesame Street, especially Elmo and educational videos with cartoon characters such as Barney.



- We use Gemini for speech therapy. We also use TheraPutty to help with low muscle tone in his hands. We use flash cards for letters and numbers and make our home environment similar to that of a homeschool environment. We make every experience something to learn from.
- We would count and try to get him to show us how many fingers, or we would tell him colors.
- Looking at picture books, practice sound & word pronunciation, placing shapes in correct slots, rhyming games
- We used special toys, blocks and other manipulatives; enrolled in music groups, sports groups etc.; watched Sesame Street video's that sang the alphabet; anything with music kept his attention.
- Always, every day. Developmental toys, play with siblings, outings in the community, every family meal, etc. If not for our own efforts at home, and seeking more than the school district was offering, she would not have gotten/ used gait trainer (we took her to community center hallways and helped her increase endurance and skill over 9 months) and consequently walked at the age she did (3). We associated her increased mobility and independence with cognitive gains occurring at the same time, as well as improvements using her AAC devices, from low tech home-made velcro letters, icons, picture books, photo social stories to GoTalk 4, 16, and Vantage Lite. Once we had initiated all these things and met with success, the school did incorporate them.
- We had an app on her iPad that helped with pincher grasp. She had to pinch the crabs to win the game. We also had flash cards that we used at home. We also had puzzles with letters, numbers, shapes and colors.
- Everything you can think of: magnet letters, pop beads, stringing beads, alphabet toys of every kind. I tried everything I could find.
- Every sensory trick written (it felt like) shaving cream writing, bean writing, play doh, slime, etc.
- We had legos, coloring books, puzzles, cognitively appropriate toys and did whatever we could to turn play into learning.
- Foam letters on the bath; a see-through Perspex board where we did shape matching (card on one side with shapes to be matched on the other side); many counting systems, with not too much luck in that field
- We read every night, make up stories and, if we watch TV, I will ask him questions about what we watched. Now that he reads I ask him to read signs when we are out in the car.
- We did anything and everything to help her. we did use shaving cream. Foam letters, magnetic numbers and letters. Stamp pad and stamps. Pegs and puzzles. And, so many other things.
- Reading to her, magnet letters on refrig, counting crackers, watching Sesame Street and a math program. We had sandpaper letters that he would trace with his fingers. We had alphabet BINGO games, and an aquadoodle to make drawing more fun.
- Buckets to dump things, Interactive toys (knobs, dials, gears, lights, etc.), Music
- Puzzles, electronic toys that played music or spoke words, and he also had his own computer with a trackball mouse and a touch screen that he played educational games on. We used picture symbols to help with communication and behavior. We encouraged him to use his communication device by programming it with people's names he knew, sports teams, places that he liked to visit, etc. He loved to look at books and be read to.
- Lot of reading books, magnetic letters on the fridge, lot and lots of iPad activities, interactive toys, Magna Doodle

During this time, many families also indicated that they had outside therapy, including but not limited to: hippotherapy, OT, PT, ST, music, family play therapy, adaptive sports, PROMPT, ABA and water therapy.

Parents reported that they would not have done anything different at this time. Those who did said they would have liked to see more: communication skills, reading, speech therapy, technological skills, improve their social skills with their peers.

One thing to look into is creating a Circle of Friends. This is created during kindergarten or first grade and is especially important if your child will be in an inclusive program. First you should pitch the program to the administrators and the teachers, indicating that you'd like to improve your child's social skills by having his or her typical peers engage in assisting with every day life at school. You need to ask the teacher to hand pick a number of students who show empathy towards your child (maybe a few that don't and need to learn empathy). Lastly, you need to get permission from the parents of these children. Ask to call an informal meeting with the stu-



dents and their parents (bring cookies or goodies), the teacher, principal and any other administrator that you feel should attend. First tell them all about your child: what he or she likes, dislikes and can do and cannot do. Tell them that you would like to improve your child's social skills while at school and could really use their help. You can tell them that they will assist your child on the playground, at lunch time, at snack time, recess, etc. (they will not accompany your child to the bathroom). This does not take the place of an aide. They do not need to invite them to their home (but wouldn't that be nice). Let them know that at any time that the student feels uncomfortable or doesn't want to continue that it is okay. A plan is created and the students who are interested then become your child's circle of friends. Parents who have used this program report improved social interactions.

Middle School

Middle school years can be very trying. There is a split between children who were in full inclusion programs, partial inclusion programs, and special day programs with pull outs for therapy and segregated program. Many of the children did not change schools or programs from elementary to middle school. Some children had full time aides, where others shared an aide with 2 or 3 other children. Most schools had resource specialists that assisted the instructors especially in the full inclusion classrooms. During this time, outside therapies seem to come to an end or are reduced tremendously.

Reported goals:

Cognitive: Reading (safety signs), academic standards, regular school goals, identifying coins, using addition, subtraction, multiplication and division, using a calculator to compute answers, retelling stories, and art

Gross Motor: Dancing/Movement therapy, swimming, independence on stairs, curbs, sitting, pulling out and pushing in chairs, catch a ball, kick a ball, throw a ball, walk a short distance, run a short distance, use walker or wheelchair to get from classroom to classroom independently, motor planning, trunk & extremities strengthening, jumping, both feet, one foot

Fine Motor: Sit at a desk independently and use pen, pencil, crayons or markers to create shapes, numbers and letters, work on puzzles, correct use of utensils, drinking from a cup, learning sign language, learning how to use a computer correctly, telling time on a digital clock

Self Help/Care: Safety skills, learning to cross the street, use of street lights, stranger safety, equipment safety, learning manners in a public setting and appropriate behaviors, learning how to control behaviors, create behavior plan, trips into the community to practice safety skills, dressing oneself, housekeeping skills, vacuuming, sweeping, etc., basic hygiene and self care, creating and maintaining friendships

Speech and Language: Increasing vocabulary and string more words together to form sentences, understanding opposites, improve sign language, learn to use communication devices appropriately, increasing and improving on sounds to help communicate, follow a two or three step direction, increase breathe support to assist with word production, continue working with oral motor exercises, make eye contact with peers and say "hello," ask/answer "Wh" questions

Extracurricular activities: Church, family activities, plays, school dances, music therapy, hippotherapy, swimming, sports teams (Challenger/Miracle League baseball, VIP soccer), Girl Scouts, Boy Scouts, Camp Fire, 4H Club

Words of wisdom from some of our wonderful parents:

Be positive and grateful!

Fight for equipment that your child needs to perform and learn

Go with your gut on program placement. If you feel that your child would be more successful in a self contained environment or inclusion environment let the school know

Fight for the 1:1 aide that my child needed



High School

As you can imagine, as your child ages and goes into high school, goals and expectations change. By this time, most kids have gotten to a fairly consistent cognitive level, although still learning and understanding. At this stage in a child's life, it is equally important to continue to work on practical daily living and social skills but to also create a successful environment to improve your child's self worth. Most children at this age will be in a self contained classroom. They may or may not have a job on campus. You may want them to have a job on campus to interact with all the students and staff (i.e. deliver the mail, pick up recyclables, etc). If your high school has a Best Buddies program, consider having your child participate. This is a great way for your child to engage with his or her peers and some might become life-long friends.

In high school your child will most likely go out into the community weekly to learn and improve his or her safety skills, communication skills, directional skills, money handling skills and job skills. He or she should also learn how to manage time and be responsible for executing a daily schedule at school. Although he or she may not be receiving therapies, all goals should include a communication and functional aspect to the goal.

Reported goals:

Cognitive: Increase use of computer to assist with math and language production, construct sentences to communicate effectively, improve independent skills, improve routine skills such as coming into class, hanging up coat and backpack, placing lunch in refrigerator, etc., correctly identify coins and paper money and assign their value, learn calendar skills—dates, days of week, month, etc., telephone skills and etiquette, laundry sorting skills

Gross and Fine Motor: Learn to use large and small tools necessary to clean up both outside and inside, maintain balance while bouncing on a therapy ball, improve upper body strength, continue to work on writing skills, continue to work on dressing skills, buttoning, tying shoes, zipping up pants/jackets

Social Skills: Improve self help skills, learn appropriate job related skills, improve daily living skills, engage in social activities and games with peers, follow instructions, use calendar to keep track of activities, learn to cook or prepare own snacks, including clean up work station and table, go shopping and learn how to use money (dollar over)

Speech and Language Skills: Accurately identify new words, work on increasing speech by lengthening of sentences, use pictures and signs to improve communication with others, continue to work on speaking more clearly and appropriately, improve sentence structure with the use of a communication device, ask appropriate questions and answer questions correctly, work on Oral Motor activities to improve tone and coordination, use social stories to decrease inappropriate behaviors, initiating conversations, using appropriate eye contact and saying "hello" to both familiar and non-familiar individuals, work on appropriate lip closure during mealtime, work on appropriate posture while eating

Extracurricular Activities: Special Olympics, school dances or dance groups, music groups, church groups, hippotherapy, teen afterschool programs

Transition

In the United States and many other countries around the world a young adult can stay in school until he or she turns 22. In most cases, when a young adult turns 19 or 20 he or she is encouraged to leave high school and go into Transition. This 2-4 year program is designed to get them ready for adulthood and adult programs. This program is still the responsibility of the public school system. Most goals are strictly daily living skills based and communication based. The young adult will be given (or should be given) a wide range of possibilities to see what he or she thrives at. Some young adults will have volunteer jobs in markets, clothing stores, animal shelters or daycares. Some will learn social and self help skills. Others will learn how to do jobs around campus. There are reported cases of young adults not going through a true transition program. They continue into high school until they age out at 22.



Homeschooling

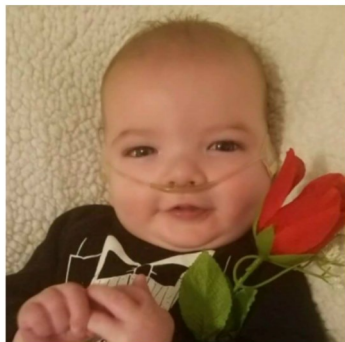
There has been an increase in parents deciding to keep their child at home and schooling them. Some of the reasons listed:

- Safety of child in an environment where they cannot communicate
- Child is fragile and is medically compromised
- Parents want to make sure that child is getting an education and not just being babysat
- Child/Adult not being properly supervised in the public school

If you are interested in homeschooling your child, the 5p– Society can connect you with parents who are currently homeschooling or have homeschooled their child for information and support. Some places to look into information and curriculum for homeschooling are through your church, parent groups, and homeschooling support groups on the internet.

Parent report the advantages to homeschooling a child:

- Teaching the child at his or her own pace
- Ability to continue therapy sessions during school hours
- Following age appropriate curriculum and teaching the child the way he or she can learn (auditory, visual, tactile, sensory) with great response.
- Using real life to teach the child practical skills



Raising an Adult with 5p-

The oldest living adult that the 5p- Society has in its database is 68-years-old (in 2018). Approximately one-fourth of the database is now an adult, 21 years and older. We greatly appreciate the very honest and candid responses we received from families all over the world who have an adult with the syndrome. We will review the topics that generate the most questions and the responses that have been given to assist those of you who are just starting this journey or are getting close to adulthood.

Medical Issues to consider

Overall health: The majority of the parents surveyed stated that their child was in good overall health with very few colds.

Weight: Responses by parents regarding their adult child's weight varied. Several parents reported that after their child turned 30, there seemed to be a thyroid issue with symptoms including gaining weight, becoming irritable and being tired. Many were then put on thyroid medication. This seemed to assist the weight and behavior issues. Other parents reported weight loss. Choking, swallowing, not tolerating foods, inability to chew properly, loss of teeth, Barrett's Esophagus, GERD and other underlying issues caused the weight loss. These individuals were given higher caloric foods and/or supplemental drinks. More females than males had the thyroid issue and more males than females were underweight.

Seizures: Many parents reported that their adult child started to have seizures as they got older. Seizure medications were used to assist in controlling them.

Tremors: A handful of parents reported that their child has hand tremors when attending to a task that requires a lot of concentration. One parent talked with a doctor about the tremors and felt that it had a lot to do with low muscle tone and the need to strengthen the back muscles.

Diabetes: Only a few parents reported that their adult child had elevated levels of AC1 for pre-diabetic considerations, but none of the parents reported that their adult child had diabetes.

Arthritis: A few parents reported that their adult child had arthritis type symptoms in the knee and hip joints.

Walking: Several parents reported that as their adult child has gotten older the harder it is for him or her to walk unassisted. Some are afraid of falling due to a loss of depth perception (off a curb, etc). Some adult children do not pay attention to their surroundings and will fall or stumble over cracks in the ground, uneven sidewalks or have poor motor planning and cannot maneuver properly around obstacles.

Vision: Those individuals who already have vision loss tend to get worse as they age.

Hearing: Some parents reported that their child had an increased hearing loss as they aged.

Constipation: Unfortunately, the constipation issues that our younger children have continue into adulthood. Parents reported that they continue to give over the counter medications to assist with the constipation.

Hormonal Changes: Very few parents talked about the hormonal fluctuations in their adult child. One parent did report that her child had a hysterectomy due to increase in behavioral issues and hormonal changes.

Social Interactions

Daily routines: The majority of adults attend either an Adult Day Program or Sheltered Workshop. At these programs they continue to work on their social and daily living skills: learning how to communicate with others, cooperation, volunteering, improving social interactions, following directions, learning new skills, and improving their vocabulary. Many of the volunteer opportunities are with Meals on Wheels, homeless shelters, animal shelters and day care centers.



Some of the centers will have classes where the adults will learn how to cook, how to shop using money, learn to garden, create artwork, learn carpentry, improve typing skills, improve their safety knowledge, perform karaoke, or perform in a play. They might have Wii tournaments, or have yoga classes (fitness). Several of the programs do go out into the community. They might go to the movies, go shopping, go to the park, or go walking/hiking. There are programs that are located on farms and the adults will learn to care for farm animals as well as grow food.

Most programs run from 8 AM to 2 PM or 9 AM to 3PM. Transportation to the programs is either provided by the program itself, via public transportation, state funded door to door pick up or by parent/guardian. The majority of the programs are state funded programs.

Other programs that adults are involved in: religious programs; outdoor camps (typically in the summer months); peer mentoring, Special Olympics or other sports programs.

Another large group of adults with the syndrome stay at home during the day with their parent or guardian. The adults will engage in daily living skills including daily chores, assisting with meal prep, going to the store, going to the library, running errands, engaging in therapy, and also playing with their favorite things. Parents or guardians will make sure their adult child is getting enough exercise through walks, gym memberships, or classes. Some parents report that their adult child may have medical issues, be severely affected by the syndrome or have difficulty with behaviors when in a day program. Parents also make sure their adult child gets plenty of social interaction through various means.

A handful of adults work. Most work with a job coach; however, a few are able to work independently. Because of Social Security laws and income requirements, those who work make pennies on the dollar (below minimum wage). Each country has its own rules on how much a disabled individual can work; please make sure you check or know the income rules before your adult becomes employed.

It is important to remember that children and adults with the syndrome require a routine. When you find the right routine you will find the child or adult more compliant. However, if you have a sudden change in routine you will find the child or adult might engage in unwanted behaviors. Some adults will also be sensory seeking if they do not get enough interactions and may begin to self harm, withdraw or engage in unwanted behaviors.

Living Options

- Lives at home with parents in own room
- Lives at home with parents but has a space designed especially for him or her
- Lives with a family member
- Lives in group home with 4 or less individuals of same sex
- Lives in group home with 8 or less individuals of same sex
- Lives in group home facility or community with over 10 individuals, mixed sex and some with same sex
- Lives on own or in a group home owned by parents where adult pays rent and parents hire support staff
- Lives on own with support staff to assist with meal planning, budgeting and keeping living area clean

Your decision on keeping your adult child with you or having him or her move to his or her own home is one that is not easy to make. Some adults are ready to take the leap and assert their independence. Some are quite content with their current living situation. You have to do what is right for you and your family. The biggest concern is what happens when you are no longer around to care for your adult and he or she only knows how to live with you. Even having them go to a week long stay over camp will provide them with a few days where they can learn some independence away from parents/guardians.



Social Security, Money, and Planning for the Future

Guardianship or Conservatorship:

When a child with the syndrome (or any genetic disability) turns 18 he or she is considered an adult and your rights as a parent are severed, according to the US government. What does this really mean . . . As the parent/guardian of your now adult you will need to apply for either guardianship or conservatorship of your child's estate. This way you can be in control of his or her monies, medical decisions, living decisions, and relationships. Each state has different rules when it comes to applying to be your child's guardian/conservator. Some states require you hire an attorney, other states will walk you through the process. In most cases, there will be a court hearing where a judge will grant you to be the guardian or conservator. You will receive a legal document that you can then use as needed for your adult child.

Special Needs Trusts:

These are designed to have a plan set for your adult child when you are gone. A special needs trust can be part of your own family trust. Consult with an attorney who can assist you in designing the trust. Make sure you talk with family members before adding them into the trust to make sure everyone is okay with the future plans. One parent surveyed explained that "everything is spelled out in her trust from what she eats, likes to do, medications/vitamins, things she cannot do and needs help with, etc." Many parents reported that they periodically updated the trusts to keep the information current.

Social Security:

At the age of 18 your adult will also be able to apply for Social Security benefits that not only include a monthly living allowance but also medical care through Medicaid/Medicare. Cri du Chat Syndrome is now listed as one of the covered conditions. Make sure you reference:

SSI Compassionate Allowance Conditions – DI 23022.375 Cri du Chat Syndrome

Contact the Social Security Administration at least 2 months before your child turns 18 to schedule an appointment. Bring all documents that support your child has a syndrome to the appointment along with your child. Social Security will send you a notification of what you need to bring.

Even though your child is sitting with you at the appointment, don't assume that they will grant the Social Security benefits immediately. They have protocols. They will schedule an appointment for you to meet with a psychologist to confirm that your child is unfit to handle his or her own affairs. It will all seem unnecessary. Keep in mind when you visit the Social Security office that your child cannot have any more than \$2,000 in his or her name. This includes, savings accounts, and savings bonds, and more. If there is more than \$2,000 in his or her name remove it and put it into your own account. Your income does not count towards your child's eligibility. Parents reported that their adult child receives between \$635 to \$755 a month, with amounts increasing if they live on their own, or if one parent is retired. The amount your child receives may be different from another child — there are several factors that will assist SSA in determining the exact amount.

You will need to make sure you do not let your child's income exceed the \$2,000.00 limit or the Social Security Administration will take it away from you and make you pay back the overage. For most families the adult will contribute to the family budget. You as the beneficiary of your adult child's account can withdraw the money and utilize it for rent, food, utilities, personal items, gifts, clothing, medical, dental, vision co-pays or other items that insurance does not cover. Keep receipts and records as you may be asked to present them. You will be asked to prepare an annual statement to let SSA know how the monies are being spent. A representative may call you annually to ask if anything has changed (living arrangements, etc.). Every couple of years a representative from Social Security will contact you and ask you a series of questions to confirm that your child is still disabled. While this seems unnecessary, please be prepared and ready for them to ask these questions.



The ABLE Act (Achieving Better Living Experience Act):

The Stephen Beck Jr. Achieving a Better Life Experience (ABLE) Act (Public Law 113-295) was signed into law on December 19, 2014. The ABLE Act amends the Internal Revenue Service Code of 1986 to create tax-free savings accounts for individuals with disabilities. The funds in the ABLE account do not count toward the \$2,000 cap on assets that is required to remain eligible for critical government supports.

An ABLE account may fund a variety of essential expenses for individuals with disabilities including medical and dental care, education, community-based supports, employment training, assistive technology, housing, and transportation. The federal ABLE Act authorizes the states to develop their own ABLE programs, and many states have moved quickly to pass ABLE laws and are in various stages of developing their ABLE programs. (<https://www.ndss.org/advocate/national-advocacy-public-policy/achieving-a-better-life-act-experienceable-act/>)

Not every state is participating in the ABLE Act. Please visit <https://www.ndss.org/advocate/national-advocacy-public-policy/achieving-a-better-life-act-experienceable-act/state-able-programs/> to see if your state is listed, if the program is enabled or still waiting to be launched.

There are specific rules with the ABLE Act. The best part is that monies that are included do not count against your \$2,000 limit that the SSA has set. Please note that all monies may be required to be returned back to the government after the passing of your adult whose name is on the ABLE Account.

Always Learning and Growing:

Don't let anyone tell you that your adult with the syndrome will stop learning. Keeping in mind that many of our adults are over 30 years of age and technology was just beginning, many of our adults know how to use computers, iPads, communication devices, and smart phones. Many adults learn how to complete chores including laundry sorting, washing dishes, making beds, managing their items, taking care of their animals (feed them and walk them), bathing independently and controlling their behaviors. Some are able to complete more difficult tasks such as cooking with assistance, managing a budget, working independently, putting on make-up and operating a pod-like coffee maker.

Even parents who have adults who are severely affected by the syndrome report that they continue to learn, recognize, and understand the world around them.

Safety Concerns:

A common concern among the parents of adults with the syndrome is in regard to safety. For the most part our children and adults are very outgoing, friendly, happy and curious. They like to go up to strangers and say hello, touch little babies and kids and give hugs "just because" to anyone. We as parents continually have to teach our kids about boundaries; what is appropriate and not appropriate. Fist pumps, high fives and waves are constantly being encouraged at home, at programs, and in the community. Safety with strangers is only one of the safety concerns. Another is safety out in the community. Learning how to cross the street correctly, being mindful of their surroundings, and walking without walking into something or somebody are a few of the safety concerns. Most parents report that they make sure that their adult is being monitored at all times, providing just enough space so that they feel some sort of independence.



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Resources and References

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You will find C5 throughout the book, guiding you along the way. C5 (for *Chromosome 5*) , was created by our friends in Australia and adapted as the mascot for the syndrome and awareness week.

