

Patient Stories

1) Young patients identified in NBS who can't access an accepted medical food/formula or treatment due to inadequate coverage policies of state or private payer

Patient P is a 22 year old affected with Citrullinemia. Since Medicaid will not provide metabolic formula after the age of 21 he is currently receiving both ProPree and Cyclinex-2 from the Ross Patient Assistance Program and his family is purchasing his L-Arginine out of pocket (around \$270/month). P is neurologically delayed and cannot work or he will lose his Medicaid coverage. He receives his only income from social security.

P's parents are concerned about his health if the Ross Assistance Program should deny coverage and how he would get his L-Arginine if something should happen to them. They feel that P is already in a dangerous situation with his diet since he relies solely on the assistance program and his parents' finances.

Patient Y is a 14 year old female affected with PKU. She has health insurance that will cover her formula but the co-pay is so large that her family cannot afford it. She has instead applied to the Ross Assistance Program which will cover her Phenex-2 but still must pay the large co-pay required for her PhenylAde MTE Amino Acid Blend since this formula is not made by Ross.

Y's parents are concerned that the Ross coverage will not last and have seen the side effects when Y is not in metabolic control. Y has significant behavioral problems and difficulties focusing when her phe levels are not within therapeutic range that affect her school work and performance as well as severe eczema all over her body. Her parents worry that if she no longer has coverage from the assistance program she may have to decrease her formula to an amount that her family can afford, thus increasing her phe levels and increasing her behavioral problems.

Patient D is a 14 year old male affected with PKU. He was in good metabolic control as a young child but began to have variations in his levels due to difficulty in taking his formula. This difficulty eventually led to D stopping his formula altogether at the age of 12. D came back to clinic and tried a new formula which he liked and would take, but was much more expensive.

D continued to take his formula until his order was refused due to a \$1,600 that had accumulated over 3 months. Since his family could not afford such a large co-pay, D discontinued formula for almost a year. D's parents have been sending small payments to slowly decrease their balance but have not paid enough to allow D to receive more formula. The possibility of switching to a lower-cost formula in the future was discussed, but rejected as his current formula is the only one D will take.

D's growth has been retarded due to his lack of formula and his parents report that he is fidgety, short-tempered and unable to sit still when his levels are not under control. D is now waiting for the payments to be processed and will hopefully have paid enough of the balance to receive formula in the next few months.

2) Older patient who can't get covered after age of majority

Patient M is a 25 year old female affected with PKU. She is not currently taking metabolic formula nor adhering to the low protein diet recommended because she cannot afford these items (in addition to working full-time, she also provides care for her father and assists him financially).

Over the past few years M has had difficulty obtaining and maintaining health insurance due to her low income and ulcerative colitis, and she does not qualify for Medicare or Medicaid. She finally obtained insurance coverage in May and came to clinic for the first time in several years. During her visit, M reported that since she has gone off-diet she has been experiencing severe, frequent headaches, problems maintaining focus, mood swings, and fatigue. M was also very concerned about the outcome if she became pregnant while her diet was not controlled. She expressed a desire to go back to taking her formula and adhering to her diet, but did not have a way to afford it. As M's insurance will not cover the formula she needs, clinic staff gave M materials and documentation to apply for NORD assistance in purchasing her formula and are waiting to hear if she has been accepted.

3) Examples of parents who have had to fight for coverage in either a state with good or poor coverage policies

Patient L is a 28 year old male affected with PKU. He has been off diet since he was 18 years of age and was no longer covered by his parents' insurance. L is unable to afford formula and low-protein foods, and his insurance has repeatedly denied coverage for his formula. He would like to return to his low-protein diet, especially since he has noticed the side effects of poor metabolic control: L reported that since he has been off diet he has frequent headaches and lethargy as well as an inability to stay focused that has interfered with his work. As his insurance will not cover the formula, L is considering a patient assistance program.

Patient F is a 37 year old female affected with PKU. F was off diet for several years, but returned to clinic and went back on diet when planning her pregnancy. F quickly became pregnant, then discovered that her insurance was only providing payment for some formula charges and giving no reimbursement for others. Clinic staff sent letters of medical necessity detailing the need for F's treatment during pregnancy and after several months of negotiation the insurance company paid a percentage of the outstanding charges. This greatly reduced the amount F owed, but she was still left with a bill of over \$4000 after her pregnancy. Because she could not afford to pay the bill in full, F had to discontinue her formula. F would like to have another child, but as she cannot obtain formula she has not been able to begin planning for pregnancy.

4) Examples of a patient that was harmed due to inability to access food/formulas or treatments

Patient C is a 31 year old female affected with PKU. She is mother of a 3 year old and would like to have another child. She is very conscientious of the dangers of high phe during pregnancy and had terminated a pregnancy previously because her levels

were not in control during the first trimester. Currently, her co-pay for her formula is so expensive that she is not currently able to afford it. C has been participating in research for the last 6 months and was able to obtain formula through the study. Now that the study is over, she is without formula and very concerned that she may have become pregnant at the end of the study. C is extremely anxious about what will happen if she has become pregnant since she cannot afford her co-pay and does not qualify for assistance in purchasing her formula.