VMP, llc
Expanding Genetic Horizons

Voice: 404.793.7800
Fax: 866.744.5665
www.vmpgenetics.com

CASE STUDY – PEDIATRIC HOSPITAL (0178)

THE NEED

Request from the metabolic service in a university children's hospital serving a metropolitan area of approximately 700,000 people and a state of four million. The mildly busy service had no clinical geneticist at all at that time, and the hospital was busy trying to recruit several genetics faculty members. The center is the major metabolic center for the state and a referral center for the state's newborn screening program.

THE SERVICE

Physician support provided 24/7 coverage through the first 19 months of the contract and then adjusted downward to more regular case reviews and training. Given the absence of an accredited on-site geneticist, during the first 5-6 months, the VMP provided metabolic support to the ancillary health professionals in the metabolic clinic (nurse, physician assistant, metabolic dietitian), and various hospital attending physicians (e.g., Neonatology, Hospital Medicine, Emergency Medicine). After new faculty were hired, assistance was provided mostly to the junior clinical geneticists. More recently, the onsite biochemical geneticist left and VMP coverage was increased to 24/7.

About half the cases were discussed by phone with follow-up by email; the others were more focused and conducted entirely by email. The metabolic service had printed emergency protocols, and these were shared with VMP. A written summary of the consultation was provided after every Physician Support encounter.

Consultation requests have included:

1. The evaluation and management of acute, new-onset metabolic disorders, neonatal-onset hyperammonemia and encephalopathy (due to a urea cycle defect), acute hyperammonemia in a teenager (due to a urea cycle defect), metabolic ketoacidosis/hyperammonemia (due to an organic acidemia), neonatal lethargy (due to new-onset maple syrup urine disease), neonatal hyperbilirubinemia (due to galactosemia), neonatal-onset seizures and hypotonia (due to nonketotic hyperglycinemia);
2. The acute and chronic management of diagnosed patients with a wide range of inborn errors, including acid disorders aminoacidopathies, urea cycle disorders, organic acidemias, cobalamin defects, vitamin transporter defects, glycogen storage diseases, fatty acid oxidation defects, defects of pyruvate metabolism, respiratory chain defects, mucopolysaccharidosis);
3. Referrals from the newborn screening program (e.g., Pompe, mucopolysaccharidosis type I, citrullinemia, glutaric acidemia type I, VLCAD deficiency, CPT II deficiency, as well as high citrulline, low C0, high C0, elevated C3, high C4, high C5, high C5OH, high C14:1, high carnitine, ?Pompe, ?MPS I);
4. The clinical work-up of (undiagnosed) patients with:

VMP, llc
Expanding Genetic Horizons

Voice: 404.793.7800
Fax: 866.744.5665
www.vmpgenetics.com

a. Unexplained clinical phenotypes (e.g., liver failure, autism, failure to thrive, global delays, seizures, coma, loss of motor skills), and/or
b. Biochemical derangements (e.g., hypoglycemia, metabolic acidosis, lactic acidemia, hyperammonemia, carnitine deficiency, high transaminases).

In the 5th year of the contract, an hour of (remote) teaching per month for clinicians, trainees and support staff was added to the schedule.

THE OUTCOME

Physician support is ongoing for over 6 years.

Comments by faculty members:

"Thanks Mark. You always teach me so much."

"I just wanted to thank you again for guiding me through all of this. You are awesome."

"Thanks for the great talk yesterday! Only you can make it to so fundamental yet still interesting! I had one development fellow and 2 med students with me in the room… they all said no one ever explained the blood gas and its implication to this level !"

"I have been using VMP services since 2020. My experience has been beyond amazing. Dr. Korson's medical knowledge and expertise to provide guidance has helped me save many lives. During the last three years I have faced complex metabolic emergencies and the support I received from VMP has helped my patients to have a good outcome. I read extensively about metabolic disorders, patient's may not fit what has been published and treatment interventions may not be the same for everyone, VMP physicians Dr. Korson and Dr. Arnold have many years of experience and have helped me come up with management interventions unique to my patient's situation. Dr. Korson is a talented educator and not only helps you with recommendations but also helping you understand the mechanism, pathophysiology, recognize ultra-rare metabolic flags that may not be in books or easily available in medical literature. VMP has provided my institution with physician support, consultation, educational presentations, interpretation of challenging NBS. Their program has been lifesaving to our institution. I am thankful for all the support provided to me and understanding I am not alone in the unpredictable scenarios I have faced."