VMP, llc
Expanding Genetic Horizons

Voice: 404.793.7800
Fax: 866.744.5665
www.vmpgenetics.com

CASE STUDY – PEDIATRIC HOSPITAL (0036)

THE NEED

Request from the metabolic service in a children's hospital and two other hospitals for which the service provides coverage. The metropolitan area has a population of approximately 7.7 million people. A solo biochemical geneticist of a busy metabolic practice requested various types of coverage, including days (8am-5pm) or nights (5pm-8am), a full 24 hours (from 8am-8am), a full-week (24 hours/day from Monday 8am through Monday 8am), as well as through some weekends (24 hours/day, Friday 5pm to Monday 8am). The coverage request varied from month to month. The center is a major metabolic center and a referral center for the state's newborn screening program.

THE SERVICE

Physician support coverage varied throughout the contract to meet fluctuating schedule needs of the hospital genetics service. Clinical geneticists at the hospital rotated in the monthly schedule to take periodic first metabolic call with back-up from the center's biochemical geneticist or the VMP Physician Support Service. The on-call geneticist decided on a case-by-case basis whether or not back-up assistance was required. Most cases were discussed by phone with follow-up by email. Less commonly, the consult was very focused and conducted entirely by email. All information, including laboratory and other tests results, were provided by the geneticist-on-call to the VMP consultant, as needed. The center provided to VMP access to all management protocols used by the metabolic service. A written summary of the consultation was provided after every Physician Support encounter.

Consultation requests have included:

1. The acute and chronic management of diagnosed patients with a wide range of inborn errors, including amino acid disorders (phenylketonuria, maple syrup urine disease), urea cycle defects, organic acidemia, glycogen storage diseases, fatty acid oxidation defects and carnitine transporter defects, mitochondrial diseases (pyruvate dehydrogenase deficiency, MELAS), TANGO II metabolic encephalopathy, creatine synthesis defects, and congenital disorders of glycosylation;
2. Referrals from the newborn screening program (high carnitine, low carnitine, high phenylalanine, high C3, high C5DC). Of note, due to inadequate staffing, the center has periodically been closed to newborn screening referrals;
3. Clinical work-up of (undiagnosed) patients with:
    a. Unexplained symptoms (e.g., failure to thrive, autism, cholestasis, liver failure, rhabdomyolysis, seizures (neonatal, childhood), hypotonia, neuropathy, cardiomyopathy, acute fatty liver of pregnancy), and/or
    b. Biochemical derangements (e.g., hypoglycemia, metabolic acidosis, lactic acidemia/acidosis, respiratory alkalosis, hyperammonemia,

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homocysteinemia, low carnitine, low methionine, high liver transaminases, renal tubular acidosis).

4. In addition, there was a need for expert metabolic nutrition assistance when the on-site dietitian was out due to illness or on vacation. VMP arranged for a seasoned metabolic dietitian to provide remote assistance and teaching to the on-site novice dietitian around the dietary management of several metabolic diseases.

THE OUTCOME

Physician support is ongoing for over 5 years.