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PRACTICAL PEARL: MAKING SENSE OF EXPANDED NEWBORN SCREENING

INTRODUCTION	• In January 2018, a pilot Newborn Screening (NBS) program was launched, testing for 4 additional genetic diseases. • SMA is a potentially severe progressive muscle disease. Patients who screen positive for SMA may be candidates for life-altering therapy that needs to be started immediately after diagnosis is confirmed. Testing can inform prognosis since there are multiple types / severities. Babies who screen positive may have milder forms and not need urgent treatment. Affected newborns may have muscle weakness. • Pompe disease is a potentially severe muscle disease that affects heart and skeletal muscle. Available treatment is enzyme replacement therapy which should be started as soon as diagnosis is confirmed. Testing can inform prognosis since there are multiple types / severities including adult onset. Affected newborns may have hypotonia and cardiomegaly. • MPS1 is a progressive multisystemic disorder affecting the brain, heart, skeleton, and other systems due to accumulation of glycosaminoglycans in the lysosomes. Severity is variable. Treatment includes enzyme replacement therapy and hematopoietic stem cell transplant (HSCT). • XL adrenoleukodystrophy is a peroxisomal disorder that affects boys who can develop leukodystrophy and adrenal insufficiency in childhood, some of whom will become candidates for HSCT. Affected male newborns may be asymptomatic.
INITIAL EVALUATION AND MANAGEMENT BY PRIMARY CARE	• The NBS lab will contact you with abnormal results and advise you to contact a metabolic center (Baystate Genetics). You may be asked to send a repeat specimen to the lab. •
HOW TO REFER	• (413) 794-KIDS or 413 794 8890. • Page Dr Abbott or Dr Villalba
WHAT TO EXPECT FROM BAYSTATE CHILDREN'S HOSPITAL VISIT	• We are available to urgently coordinate evaluation and next steps for these patients. Baystate Genetics will confirm diagnosis and prognosis, provide genetic counseling, treat patients with enzyme replacement therapies, and refer to Boston in cases needing HSCT or intrathecal SMA treatment.

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