



Technical Assistance from the New York State Newborn Screening Program

Michele Caggana, Sc.D., FACMG June 23, 2017 November 14, 2017

Workshop

- July 19 21, 2017 in Albany
- Didactic training from experts
- Laboratory component: MS/MS and molecular; didactic and hands-on
- Tour of Applied Genomics Technology Core facility
- Follow-up component: visit to local Center; review of cases; talking ot provider about molecular
- Materials to take home



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Additional Assistance

- Technical Assistance; visits
- ALD, Pompe, MPS1 education
- 1st tier (MS/MS) for Pompe, ALD and MPS1
- 2nd tier (Sanger) for Pompe and MPS1
- 3rd tier (Sanger) for ALD
- Coming soon: Next generation sequencing for Pompe, ALD and MPS1



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Questions from the Crowd

- Cost: Depends on what you need, reagents? Volume? Instrumentation? Staff levels?
- Number of people: 3 trained on each condition
- When reported: Start DNA if initial very low or if late in week; wait for result and report at referral (unless very low); ~ 2 days



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Questions? More Information

michele.caggana@health.ny.gov 518-473-3854

I am happy to discuss availability for testing services via contractual agreements



