



NEWBORN SCREENING FOR ARGINASE 1 DEFICIENCY (ARG-I)

Technical Advisory Committee meeting
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Presenter



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MPHc

Follow-up Lead

Newborn Screening Program

Test for ARG-I

Adapted to Mass Screening

- Technology – tandem mass spectrometry (MS/MS)
- Uses one 1/8" hole punch from dried blood spot to test for 19 congenital disorders simultaneously
 - Amino acids
 - Acylcarnitines (fat transporters)
- In WA NBS Program since 2004

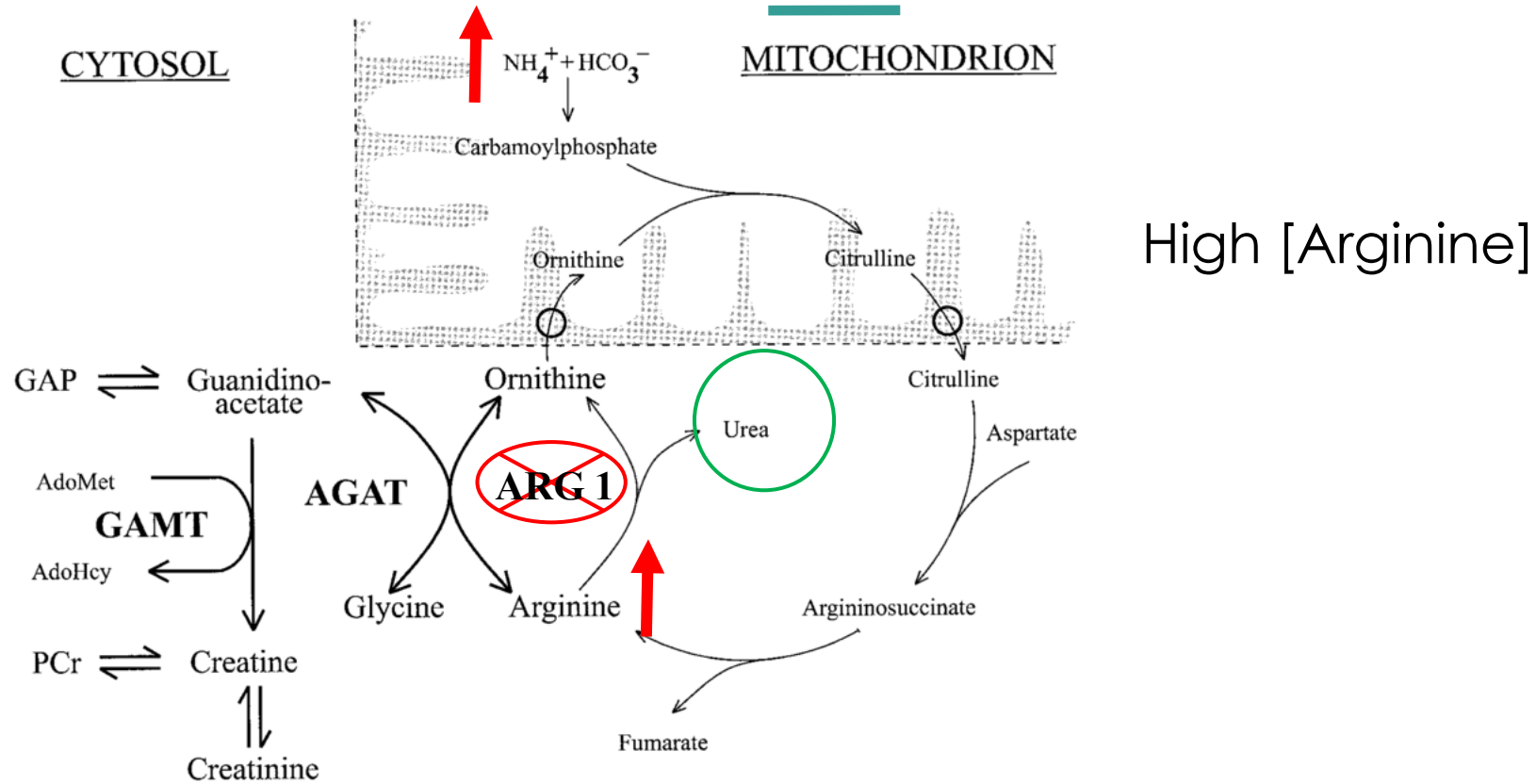


Test for ARG-I Adapted to Mass Screening

- Amino acid analysis
 - Primary target: high Arginine



Test for ARG-1 Adapted to Mass Screening



Newborn Screening – ARG-I

- US based Screening– 29 million babies
 - 22 cases of ARG-I (prevalence = 0.75:1,000,000 births)
 - 22 true positives (sensitivity = 100.0%)
 - 0 false negatives
 - False positive rate (US) 5.0/100,000 (specificity = 99.99%)
 - PPV = 99.99%

Questions?



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