

KEY PRIORITIES FOR FUTURE RESEARCH AND PATIENT ADVOCACY



Optimise Current Therapy

- Long-term outcome studies under current Standards of Care
- Improve quality & taste of supplements/medical foods
- Clinical studies to further define approach with supplements (amino acids, vitamins)
- Define impact of CBS deficiency on quality of life (QoL) to understand avenues for improvement



Optimise Diagnostic Process

- Improve Newborn Screening tools & programs
- Increase detection rate of symptomatic and asymptomatic patients
- Improve understanding of the frequency of the disease and overall awareness of CBS deficiency



Advance New Treatment Modalities

- Safe oral therapies to ease/avoid dietary restrictions ie chaperones
- Enzyme replacement therapy with CBS enzyme or enzymes designed to remove Met or Hcy
- Gene therapies including gene transfer as potentially one-time curative approach