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Invitation to Participate in a Research Study for Early Disease Diagnosis

We are working to develop a **newborn screening** assay panel for **10 biomarkers** covering **nearly 21 disorders**. We are seeking dried blood spots from individuals diagnosed with the following disorders:

1. Adenylosuccinase deficiency (*ADSL*)
2. AICA-ribosiduria due to ATIC deficiency (ATIC)
3. Aromatic L-amino acid decarboxylase deficiency (*DDC*)
4. GABA-transaminase deficiency (*ABAT*)
5. Molybdenum cofactor deficiency A (*MOCS1*)
6. Molybdenum cofactor deficiency B (*MOCS2*)
7. Molybdenum cofactor deficiency C (*GPHN*)
8. Sulfite oxidase deficiency (*SUOX*)
9. Ornithine transcarbamylase deficiency (*OTC*)
10. Hereditary orotic aciduria (*UMPS*)
11. Transaldolase deficiency (*TALDO1*)
12. Transketolase deficiency (*TKT*)
13. Ribose 5-phosphate isomerase deficiency (*RPIA*)
14. Sedoheptulokinase deficiency (*SHPK*)
15. Bachmann-Bupp syndrome (*ODC1*)
16. Synder-Robinson syndrome (*SMS*)
17. Pyridoxine-dependent epilepsy (*ALDH7A1*)
18. Zellweger syndrome and peroxisome biogenesis disorders
19. Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE), thymidine phosphorylase deficiency (*TYMP*)
20. Cerebrotendinous xanthomatosis, CTX (*CYP27A1*)

Key points about the study:

- We need volunteer participants to provide samples for validating the assay for disease screening.
- Your involvement would entail a simple finger prick procedure to obtain a dried blood spot sample.
- Conveniently, we can send you a home collection kit.

Families could also request that newborn blood spots for family members diagnosed with these disorders from the state labs be sent to us. We can provide the forms required to do this.

We are also interested in obtaining samples from parents or siblings of those diagnosed with the disorders to strengthen the assay further.

If you are open to participating, please contact **Abbhi Rajagopal** at **abbhirami.rajagopal@bcm.edu**. We will connect with you to answer any potential questions and obtain your informed consent for this study.

Your contribution will significantly enhance our understanding and potentially expedite the diagnosis of these disorders.

This study is approved by the Baylor College of Medicine Institutional Review Board