

Table S1: Questions of the online survey

| <b>OTC Deficiency Patient Population Snapshot</b><br>Please only include alive patients into the survey   |
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| <b>1. Where is your medical centre?</b><br><br>UK<br>Turkey<br>France   |
| <b>2. What is your clinical centre's name?</b>  |
| <b>3. How many OTC deficiency cases are followed up in your clinical centre?</b><br>Total number:<br>Male:<br>Female asymptomatic:<br>Female symptomatic: |
| <b>4. Please give the numbers of your patients in each age group</b><br>0-6 years:<br>6-12 years:<br>12-18 years:<br>>18 years:                           |
| <b>5. What are the three most common initial symptoms for these patients?</b>   |
| <b>6. How many of these patients had their initial symptom in the first month of life?</b>  |
| <b>7. How many of these patients have family history?</b>   |
| <b>8. How many of these patients were diagnosed with enzyme activity?</b>   |
| <b>9. How many of these patients were diagnosed with mutation analysis?</b>   |
| <b>10. How many of these patients are on a protein restricted diet?</b>   |
| <b>11. How many of these patients are on nitrogen scavengers?</b><br>Only Sodium benzoate:<br>Only Sodium or glycerol phenylbutyrate:<br>Both:            |
| <b>12. How many of these patients had liver transplantation?</b>  |