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| **Biotinidase** |
| **Synonyms** |  |  |
| **Clinical Indication** |  | Biotinidase deficiency is an autosomal recessive metabolic disorder in which the body is not able to process the nutrient biotin properly.Biotin, sometimes called vitamin H, is an important water-soluble nutrient that aids in the metabolism of fats, carbohydrates and proteins. Biotin deficiency can result in behavioural disorders, lack of coordination, learning disabilities and seizure. Biotin supplementation can alleviate and sometimes totally arrest such symptoms. Symptoms of a biotinidase deficiency can appear several days after birth. Available if requested by Consultant Paediatrician or if agreed with Consultant Biochemist. |
| **Part of Profile / See Also** |  |  |
| **Request Form** |  | Combined Pathology manual Blood form or ICE request |
| **Availability / Frequency of Analysis** |  | Analysed by the Biochemical Genetics Laboratory, Addenbrookes Hospital, if specific criteria met. |
| **Turnaround Time** |  | 2 weeks |
| **Patient Preparation** |  |  |
| **Sample Requirements** |  |  |
|  | **Specimen Type** |  | Plasma |
|  | **Volume** |  | 0.5 ml minimum |
|  | **Container** |  |  Or  Paediatric green top (lithium-heparin)Or  Paediatric orange top (lithium heparin)Samples must be transported to laboratory immediately |
| **Reference Range & Units** |  | Provided on the referral laboratory report. |
| **Interferences** |  |  |
| **Interpretation & Clinical** **Decision Value (if applicable)** |  | Provided on the referral laboratory report. |
| **References** |  |  |
| **Test code** |  | BIOT |
| **Lab Handling** |  | Centrifuge and separate into 2 aliquots as soon as possible after collection and freeze at -20°C. |

