Invitation for patients to be enrolled in study

"Prevalence of Aromatic L-Amino Acid Decarboxylase Deficiency in cerebral palsy patients with unknown aetiology" (PTC-MA-AADC-402)

Aromatic L-amino acid decarboxylase (AADC) deficiency:

- Is a rare genetic disorder of neurotransmitter (serotonin and dopamine) synthesis
- Patients can present with varying neurology, frequently with abnormal tone (hypo or hypertonia), movement disorders and developmental delay

In this study we are calling for patients with unexplained neurological symptoms to be tested with a finger prick blood test.

- Main **inclusion** criteria: unexplained neurological findings, all patients regardless of age
- Main **exclusion** criteria: acquired causes, e.g. HIE, brain infarcts
- *Patients are not required to have received a diagnosis of CP – this is a diagnosis frequently applied to patients with AADC-deficiency and may aid identification of patients suitable for testing*

For more information, including patient leaflets, or to discuss referrals,
contact Dr Marie Monaghan (Dept of Paediatric Neurology, Bristol Royal Hospital for Children) by email :
marie.monaghan2@nhs.net alternatively, marie.monaghan@uhbw.nhs.uk
Or by telephone **07849610083**

Study Principal Investigator (Bristol): Dr Sam Amin