

CaHASE: CAH in Adulthood

Congenital Adrenal Hyperplasia (CAH) is one of the most common inherited diseases. As an inherited disease it affects individuals for life. Despite this, most of the current knowledge on CAH is based on the disease in childhood. The studies published on adults have generated some conflicting data with reports of normal and abnormal; bone mineral density, height and weight which may be due to the necessity for steroid replacement.

An audit of adults with CAH in the UK conducted by The Society for Endocrinology revealed a lack of a consensus on the best treatment of adults with CAH (*Ross RJM, UK audit of Adult Congenital Adrenal Hyperplasia Care. J. Endocrinol 164 suppl: S38.*) CAH has therefore been identified as an area that requires further research into the day to day management of adults with CAH.

The Society for Endocrinology with a grant from the Clinical Endocrinology Trust set up CaHASE (or Congenital Adrenal Hyperplasia Adult Study Executive) in 2003. It specifically looks at congenital adrenal hyperplasia in adulthood. Nineteen specialist Endocrinology centres around the UK are currently collaborating in CaHASE, a multi-centre prospective research study. By working together the 19 centres aim to study a large number of patients (more than any one centre could alone) to obtain much needed information on the effect of CAH in adults.

The aim of the research is deliberately broad to gather information on medical treatment, fertility, genetic analysis and the quality of life experienced by adults with all types of CAH (classical and non-classical, salt-wasting and non-salt-wasting and all genotypes). For individuals who agree to participate the study involves a single visit to gather data on past and present treatment, height, weight, blood pressure, blood hormone levels, genetic analysis of CAH genes and assessment of quality of life.

An exciting aspect of CaHASE is the opportunity to link the symptoms experienced by individuals with CAH and the faulty CAH causing gene that was inherited. There are a number of common gene mutations which arise in CAH. By investigating the gene mutation involved it may become clear that certain CAH genotypes are associated with specific symptoms. In time, this could lead to suggesting different treatments for different individuals.

CaHASE is also interested in how having CAH affects individual. Therefore as part of the study participants are asked to fill in questionnaires that help to assess quality of life but they are also asked to comment on any issues that they would like to highlight about having CAH.

Eighty adults with CAH have already taken part in the study and another 120 are needed. The results should be available by the end of next year. The information collected from the study should help to improve treatment for adults with CAH.

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