

## **Impact of CaHASE,**

Congenital Adrenal Hyperplasia (CAH) is one of the commonest inherited diseases, affecting 1:14,200 live births. In its severest form it is life-threatening and requires life-long treatment. With the advancement in hormone treatment over the past 50 years, individuals now survive in to adulthood. Most of the published literature on CAH is centred on CAH in childhood. CaHASE was established to address this issue and look specifically at clinical outcomes, quality of life and sexual function in *adults* with CAH.

The impact of CaHASE has been acknowledged in a recent independent review published in the *Journal of Clinical Endocrinology & Metabolism* (December 2011) and presented at Endo 2011. The review identified the results of CaHASE published in November 2010 as one of the eleven most significant papers published in the field of adrenal endocrinology in the past year (Carey. 2011. Adrenal disease update 2011. JCEM, 96:3583–3591).

The significance of CaHASE is also highlighted by the number of publications, invited talks and presentations at conferences and patient support events and the reported improvement in patient services (listed below). The project has been invited to join the Euro-DSD registry and as a direct result of the project, the Society is preparing clinical guidance on the management of the adult patient with congenital adrenal hyperplasia. Further publications on steroid equivalence and genetic analysis are underway.

### ***International Impact***

- 2012 Oral ENDO 2012
- 2012 Professor Ross, Chair of CaHASE invited speaker at ICE/ECE 2012, Florence
- 2012 Poster ICE/ECE 2012, Florence
- 2012 Oral genetics ICE/ECE 2012, Florence
- 2011 Dr Mooij presented novel mutation analysis results at ESPE, Glasgow
- 2010 CaHASE published results in American Journal– JCEM
- 2010 Professor Ross, Chair of CaHASE invited speaker at ICE, Japan
- 2009 Dr Krone presented genetic results at LWPES/ESPE, New York, USA
- 2009 Professor Arlt , Oral communication at Endo, Washington, USA
- 2009 Dr Krone presented genetic results at Endo, Washington, USA

### ***National impact***

- 2012 Oral BES 2012
- 2012 Poster BES genetics
- 2011 Dr Steve Ball. Annual North East Endocrine Research and Audit meeting

- 2011 Dr Steve Ball Northern Paediatric Endocrine group meeting
- 2010 Professor Ross, Chair of CaHASE, invited speaker at SfE BES 2010
- 2009 Dr Doherty presented fertility aspects at SfE BES
- 2009 Dr Thang presented bone and body composition aspects at SfE BES
- 2009 Dr Krone presented quality of life aspects at SfE BES

***Interaction with Patient support groups.***

- 2011 Professor Ross invited speaker at UK patient support group annual meeting, Manchester
- 2008 Article in patient support group (living with CAH) newsletter
- 2007 Professor Ross invited speaker at UK patient support group annual meeting, Stevenage
- 2006 Professor Ross invited speaker at USA patient support group (CARES)

**Effect on clinical practice at participating centres,**

Centres report finding patients lost to follow up and improved transition from paediatric to adult endocrine care. Specifically;

- Oxford - identified patients lost to follow-up and set up a designated CAH clinic in 2007-2008 after having comments from CAH patients that they would like consistency of care. This clinic is held jointly with a consultant in Clinical Genetics. They have managed to bring back the majority of patients who had been lost to follow-up and also to expand further the number of patients they see. Based on data of the last 8 months, their DNA (did not attend) rate is down to 3% from approximately 20% before the set up of the clinic
- Edinburgh - presented the CaHASE data locally, resulting in increased collaboration with local paediatric team to improve transitional care. They report wider effects to include most of the rest of Scotland.

**Publications**

- Han TS, Stimson RH, Rees DA, Krone N, Willis DS, Conway GS, Arlt W, Walker BR, Ross RJ and the United Kingdom Congenital adrenal Hyperplasia Adult Study Executive (CaHASE). Glucocorticoid treatment regimen and health outcomes in adults with congenital adrenal hyperplasia. Paper submitted to Clinical Endocrinology,.
- Han TS, Stimson RH, Rees A, Krone N, Willis DS, Wild SH, Conway GS, Arlt W, Walker BR, Ross RJ and the United Kingdom Congenital adrenal Hyperplasia Adult Study Executive (CaHASE) 2012. Glucocorticoid dose in adults with CAH; association with

clinical and biochemical variables – analysis of the United Kingdom Congenital adrenal Hyperplasia Adult Study Executive (CaHASE) cohort. ENDO 2012 Oral.

- Han TS, Stimson RH, Rees A, Krone N, Willis DS, Wild SH, Conway GS, Arlt W, Walker BR, Ross RJ and the United Kingdom Congenital adrenal Hyperplasia Adult Study Executive (CaHASE) 2012. Relationship of current glucocorticoid dose with metabolic outcomes in CAH – analysis of the United Kingdom Congenital adrenal Hyperplasia Adult Study Executive (CaHASE) cohort ICE/ECE 2012; P 53.
- Ross RJ. CAH and adulthood. Invited talk at ICE/ECE 2012.
- Krone N, Rose IT, Crosby C, Willis DS, Wild SH, Doherty EJ, Hahner S, Parajes S, Han TS, Carroll PV, Conway GS, Walker BR, MacDonald F, Ross RJ, Arlt W and the United Kingdom Congenital adrenal Hyperplasia Adult Study Executive (CaHASE) 2012 (submitted) Genotype-phenotype correlation in 153 adult patients with congenital adrenal hyperplasia due to 21-hydroxylase deficiency – analysis of the United Kingdom Congenital adrenal Hyperplasia Adult Study Executive (CaHASE) cohort. BES 2012; P306.
- Christiaan F. Mooij, Silvia Parajes, Ian T. Rose, Angela Taylor, Taner Bayraktaroglu, John Wass, John Connell, David Ray, Wiebke Arlt, Nils Krone (2011) In vitro analysis of five CYP11B1 gene mutations detected in patients with classic and nonclassic 11 $\beta$ -hydroxylase deficiency. 50th Annual ESPE Meeting.
- Ross, RJ (2010) Health Status of Adult Patients with Congenital Adrenal Hyperplasia (CAH) and the Potential for New Therapies. International Congress of Endocrinology Japan, 2010
- Wiebke Arlt, Debbie S. Willis, Sarah H. Wild, Nils Krone, Emma J. Doherty, Stefanie Hahner, Thang S. Han, Paul V. Carroll, Gerry S. Conway, D. Aled Rees, Roland H. Stimson, Brian R. Walker, John M. C. Connell, Richard J. Ross and the United Kingdom Congenital Adrenal Hyperplasia Adult Study Executive (CaHASE). 2010 Health Status of Adults with Congenital Adrenal Hyperplasia: A Cohort Study of 203 Patients. J of Clin Endocrinol & Metab Vol. 95, No. 11 5110-5121.
- R J Ross, D S Willis, S H Wild, N Krone, E J Doherty, T S Han, P V Carroll, G S Conway, D A Rees, R H Stimson, B R Walker, J C Connell, W Arlt and the United Kingdom Congenital adrenal Hyperplasia Adult Study Executive (CaHASE). The adult with CAH: results of the UK congenital adrenal hyperplasia, adult study executive (CaHASE). Endocrine Abstracts SfE/BES 2010 21 CM3.3
- Wiebke Arlt, Debbie S Willis, Emma J Doherty, Nils Krone, Thang S Han, Roland H Stimson, Paul V Carroll, Gerry S Conway, D Aled Rees, Brian R Walker BR, Sarah H Wild, Richard J Ross and the United Kingdom Congenital adrenal Hyperplasia Adult Study Executive (CaHASE). Current Management and Morbidity of Adult Patients with

Congenital Adrenal Hyperplasia - Analysis of the United Kingdom Congenital adrenal Hyperplasia Adult Study Executive (CaHASE) cohort. Endo 2009 OR40–5

- Krone N, Rose IT, Crosby C, Willis DS, Wild SH, Doherty EJ, Hahner S, Han TS, Carrol PV, Conway GS, Walker BR, MacDonald F, Ross RJ, Arlt W and the United Kingdom Congenital adrenal Hyperplasia Adult Study Executive (CaHASE). Genotype-phenotype correlation in 153 adult patients with congenital adrenal hyperplasia due to 21-hydroxylase deficiency – analysis of the United Kingdom Congenital adrenal Hyperplasia Adult Study Executive (CaHASE) cohort. Endo 2009 P2-694.
- Krone N, Willis DS, Rose IT, Crosby C, Wild SH, Doherty EJ, Hahner S, Han TS, Stimson RH, Carrol PV, Conway GS, Rees DA, Walker BR, MacDonald F, Ross RJ, Arlt W and the United Kingdom Congenital adrenal Hyperplasia Adult Study Executive (CaHASE). Current Management, Morbidity and Genotype-Phenotype Correlation in Adults with Congenital Adrenal Hyperplasia - Analysis of the United Kingdom Congenital adrenal Hyperplasia Adult Study Executive (CaHASE) cohort. 2009 LWPES/ESPE 8th Joint Meeting Abstract Number: 550629
- Emma Doherty, Debbie Willis, Sarah Wild, Louise Breen, Richard Ross, Paul Carroll and the Congenital Adrenal Hyperplasia Adult Study Executive (2009). Fertility in Adults with Congenital Adrenal Hyperplasia: Congenital Adrenal Hyperplasia Adult Study Executive (CaHASE). Sfe/BES 2009, P10.
- Thang Han, Debbie Willis, Sarah Wild, Richard Ross, Gerry Conway and the Congenital Adrenal Hyperplasia Adult Study Executive (2009). Bone mineral density and body composition in adult men and women with congenital adrenal hyperplasia: Congenital Adrenal Hyperplasia Adult Study Executive (CaHASE). Sfe/BES 2009, P299.
- Nils Krone, Debbie Willis, Stefanie Hahner, Sarah Wild, Bruno Allolio, Richard J. Ross, Wiebke Arlt and the Congenital Adrenal Hyperplasia Adult Study Executive (2009). Quality of life (QoL) in adult patients with congenital adrenal hyperplasia – Results of the Congenital Adrenal Hyperplasia Adult Study Executive (CaHASE). Sfe/BES 2009, P88.