
EuroDSD – Disorders of Sex Development

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EuroDSD is an acronym for a collaborative project of clinical and basic scientists working in the field of disorders of sex development (DSD). EuroDSD is currently supported by the European Commission under the 7th European Framework Programme (FP7) and receives funding from May 2008 until April 2011 within the topic of the natural course and pathophysiology of rare diseases. EuroDSD has 13 partners from 6 European countries; combining 6 clinical centres with 6 research institutions plus a private administrative partner. Research in DSD is desperately needed to provide a basis for diagnostic pathways, medical decision making, and distinct treatment advice.

EuroDSD aims at linking a European patient-based data collection and analysis tool with research on development of novel diagnostic strategies to identify currently unknown causes of DSD. It will design and validate a DSD GeneChip to offer the opportunity for rapid genetic analysis in DSD. A selected group of patients will be characterized by genome analysis with CGH array technology. EuroDSD will study steroid analysis by GC/MS and LC/MS/MS in urine and plasma to identify unique steroid metabolomes that will be a “fingerprint” to diagnosis. A special focus is on elucidation of androgen action as a major impact of genital differentiation. Selected mutations of the androgen receptor are investigated to correlate in-vitro function with long-term post-pubertal outcome. Also, the molecular functions of the androgen receptor during embryology are elucidated due to identification of relevant co-regulators of androgen action. Sex-specific methylation target genes will be identified in patients with androgen insensitivity syndrome. The EuroDSD project is complemented by the elaboration of a DSD e-learning webportal, which is targeted within the ESPE e-learning activities.

The EuroDSD project offers a unique networking opportunity for European-based research on rare disorders of sex development, and we hope that this consortium can lead to a sustainable association of clinicians and basic scientists propagating research in this field.