“www.steroidogenicfactor-1.info”: An online database of variants in steroidogenic factor-1 (SF-1, NR5A1) and resource for families and professional healthcare providers

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Background
Steroidogenic factor 1 (SF-1), encoded by the gene NR5A1, is a member of the orphan nuclear receptor superfamily and an important regulator of gonadal and adrenal function. Pathogenic variants of SF-1 can disrupt the development and function of adrenal and reproductive systems leading to a spectrum of conditions including: 46,XY DSD, hypospadias, adrenal insufficiency, male factor infertility and primary ovarian insufficiency. Inheritance patterns can also be complicated (e.g. de novo dominant, sex-limited dominant, autosomal recessive). Current information available on SF-1 is not easily accessible for patients and families or healthcare professionals not working in the field.

Objective
To provide information on SF-1 for patients, families and healthcare providers through the website www.steroidogenicfactor-1.info to be launched in early 2016.

A preview of www.steroidogenicfactor-1.info

Welcome to steroidogenicfactor-1.info!
The new website designed to inform patients and allied health professionals about steroidogenic factor-1.

What is SF-1/Understanding SF-1
A brief history of the discovery of SF-1 and how it has been shown to be a key regulator of endocrine function.

SF-1 Variant Database
- Find the reported variants/mutations alongside clinical features and experimental evidence to support pathogenicity
- See phenotypic trends in relation to key protein domains
- Get allele frequencies of variants in control populations
- Population genetics

SF-1 Related Conditions
- Learn more about the range of phenotypes associated with disruption of SF-1 in humans
- Find out how these conditions manifest at different ages

Genetic Inheritance Patterns
- Information about how SF-1 related conditions can be inherited

Useful Links
Find links to:
- Patient support resources
- SF-1 publications and reviews
- Website contact information

Frequently Asked Questions
- Perspectives on SF-1 associated conditions for families and links to useful resources
- Specialised information for healthcare providers

Summary
- www.steroidogenicfactor-1.info will be a useful website relating to NR5A1 genetics and associated clinical phenotypes
- To date, there are more than 60 publications on the SF-1 Variant Database and these are increasing by approximately 10-15 per year
- This website will also provide information for patients and families about SF-1 and links to further resources


Available in early 2016