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"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

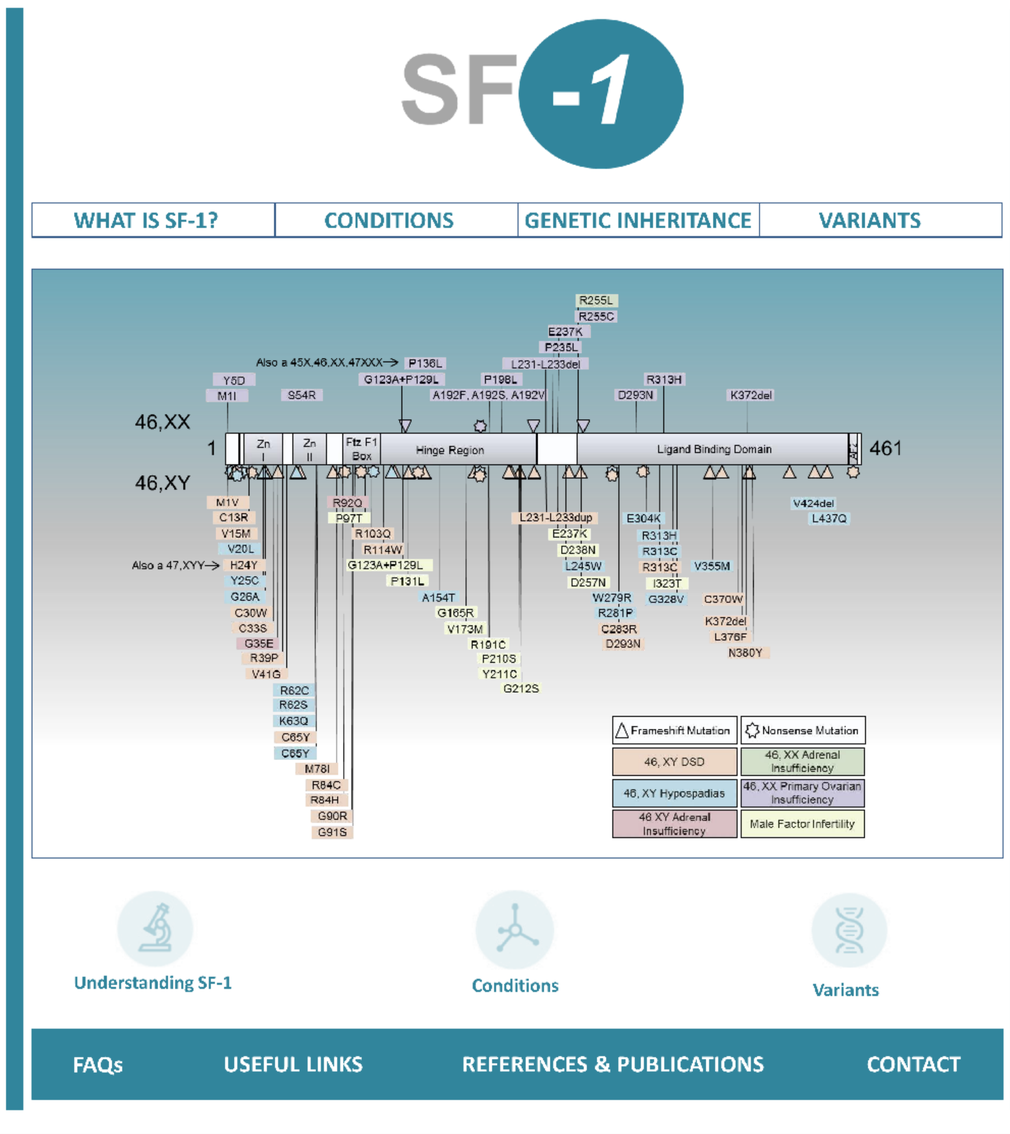
A brief history of the discovery of SF-1 and how it has been shown to be a key regulator of endocrine function.

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

Frequently Asked Questions

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



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

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

Key references: Suntharalingham JP, Buonocore F, Duncan AJ & Achermann JC. DAX-1(*NR0B1*) and steroidogenic factor-1 (SF-1, *NR5A1*) in human disease. *Best Pract Res Clin Endocrinol Metab*. 2015;29:607-619. Figure from Lin L. et al. Heterozygous missense mutations in steroidogenic factor 1 (SF1/Ad4BP, *NR5A1*) are associated with 46,XY disorders of sex development with normal adrenal function. *J Clin Endocrinol Metab*. 2007;92:991-999. Modified with permission of The Endocrine Society; Copyright © 2007.

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