



# SZABO SCANDIC

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## Produktinformation



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# PRODUCT INFORMATION



## NR5A1 (C247S, C412S mutant; human, recombinant)

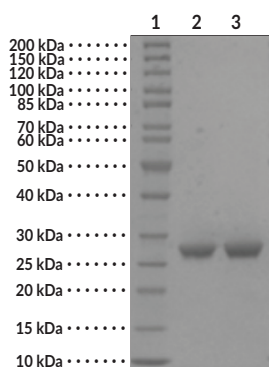
Item No. 41072

### Overview and Properties

<b>Synonyms:</b>	Ad4BP, Adrenal 4-binding Protein, Fushi Tarazu Factor Homolog 1, Nuclear Receptor Subfamily 5 Group A Member 1, SF-1, Steroidogenic Factor 1
<b>Source:</b>	Recombinant human N-terminal His-tagged NR5A1 (C247S, C412S mutant) expressed in <i>E. coli</i>
<b>Amino Acids:</b>	218-461
<b>Uniprot No.:</b>	Q13285
<b>Molecular Weight:</b>	31.01 kDa
<b>Storage:</b>	-80°C (as supplied); avoid repetitive freeze/thaw cycles
<b>Stability:</b>	≥1 year
<b>Supplied in:</b>	20 mM Tris-HCl, pH 8.5, with 1 mM TCEP and 150 mM sodium chloride
<b>Protein Concentration:</b>	<i>batch specific</i> mg/ml

Information represents the product specifications. Batch specific analytical results are provided on each certificate of analysis.

### Image



Lane 1: MW Markers

Lane 2: NR5A1 (2 µg, reduced)

Lane 3: NR5A1 (2 µg, non-reduced)

**SDS-PAGE Analysis of NR5A1.** This protein has a calculated molecular weight of 31.01 kDa.

**WARNING**  
THIS PRODUCT IS FOR RESEARCH ONLY - NOT FOR HUMAN OR VETERINARY DIAGNOSTIC OR THERAPEUTIC USE.

**SAFETY DATA**  
This material should be considered hazardous until further information becomes available. Do not ingest, inhale, get in eyes, on skin, or on clothing. Wash thoroughly after handling. Before use, the user must review the complete Safety Data Sheet, which has been sent via email to your institution.

**WARRANTY AND LIMITATION OF REMEDY**  
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# PRODUCT INFORMATION



## Description

Nuclear receptor subfamily 5 group A member 1 (NR5A1) is a nuclear receptor and member of fushi tarazu factor 1 subfamily of nuclear proteins.<sup>1,2</sup> NR5A1 monomers are composed of a hinge region, corepressor or coactivator binding site, a DNA-binding domain, and a hydrophobic ligand-binding domain, which can associate with different types of lipids, such as phosphatidylcholine, phosphatidylethanolamine, or phosphatidylglycerol, to activate steroidogenic gene expression.<sup>1-3</sup> Additional post-translational modifications, including phosphorylation, acetylation, and SUMOylation, are necessary for coactivator or corepressor binding and transcriptional activity or repression, respectively.<sup>1</sup> NR5A1 is critical in embryonic and pubescent sexual organ development and is expressed in the testes, ovaries, hypothalamus, and spleen, as well as pituitary and adrenal glands.<sup>2,4</sup> Mutations in NR5A1 are associated with low testosterone levels, androgyny, male infertility, malformed testes, ovarian dysfunction, and adrenal insufficiency.<sup>4-6</sup> Cayman's NR5A1 (C247S, C412S mutant; human, recombinant) protein consists of 243 amino acids and has a calculated molecular weight of 31.01 kDa.

## References

1. Urs, A.N., Dammer, E., Kelly, S., *et al.* Steroidogenic factor-1 is a sphingolipid binding protein. *Mol. Cell. Endocrinol.* **265-266**, 174-178 (2007).
2. Wang, W., Zhang, C., Marimuthu, A., *et al.* The crystal structures of human steroidogenic factor-1 and liver receptor homologue-1. *Proc. Natl. Acad. Sci. USA* **102(21)**, 7505-7510 (2005).
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4. Fabbri-Scaliet, H., de Sousa, L.M., Maciel-Guerra, A.T., *et al.* Mutation update for the NR5A1 gene involved in DSD and infertility. *Hum. Mutat.* **41(1)**, 58-68 (2020).
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6. Lourenço, D., Brauner, R., Lin, L., *et al.* Mutations in NR5A1 associated with ovarian insufficiency. *N. Engl. J. Med.* **360(12)**, 1200-1210 (2009).

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