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Diagnostik & molekulare Diagnostik



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### Zuschläge

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- Gefahrgutzuschlag
- Expressversand

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# QDPR (m): 293T Lysate: sc-122863

## BACKGROUND

QDPR (quinoid dihydropteridine reductase), also known as DHPR (dihydropteridine reductase) or PKU2, is a member of the short-chain dehydrogenases/reductase (SDR) family of enzymes. Functioning as a homodimer, QDPR plays an important role in the recycling of tetrahydrobiopterin (BH4), an essential cofactor for the hydroxylation of the aromatic amino acids (tryptophan, tyrosine and phenylalanine). More specifically, QDPR catalyzes the regeneration of BH4 from quinonoid dihydrobiopterin (qBH2), the product generated from the hydroxylation reactions. Mutations in the gene encoding QDPR can lead to phenylketonuria II (also called PK2 or dihydropteridine reductase deficiency), a disorder resulting from the depletion of Dopamine, epinephrine and Serotonin due to defective recycling of BH4. Symptoms of PK2 include hyperphenylalaninemia, axial hypotonia, truncal hypertonía, microcephaly and abnormal thermogenesis.

## REFERENCES

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2. MacDonald, M.E., et al. 1987. Physical and genetic localization of quinonoid dihydropteridine reductase gene (QDPR) on short arm of chromosome 4. *Somat. Cell Mol. Genet.* 13: 569-574.
3. Dianzani, I., et al. 1993. Two new mutations in the dihydropteridine reductase gene in patients with tetrahydrobiopterin deficiency. *J. Med. Genet.* 30: 465-469.
4. Dianzani, I., et al. 1998. Dihydropteridine reductase deficiency: physical structure of the QDPR gene, identification of two new mutations and genotype-phenotype correlations. *Hum. Mutat.* 12: 267-273.
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7. Online Mendelian Inheritance in Man, OMIM™. 2002. Johns Hopkins University, Baltimore, MD. MIM Number: 261630. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>
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9. Farrugia, R., et al. 2007. Molecular genetics of tetrahydrobiopterin (BH4) deficiency in the Maltese population. *Mol. Genet. Metab.* 90: 277-283.

## CHROMOSOMAL LOCATION

Genetic locus: *Qdpr* (mouse) mapping to 5 B3.

## RESEARCH USE

For research use only, not for use in diagnostic procedures.

## PRODUCT

QDPR (m): 293T Lysate represents a lysate of mouse QDPR transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

## APPLICATIONS

QDPR (m): 293T Lysate is suitable as a Western Blotting positive control for mouse reactive QDPR antibodies. Recommended use: 10-20 µl per lane.

Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

## STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. Non-hazardous. No MSDS required.

## PROTOCOLS

See our web site at [www.scbt.com](http://www.scbt.com) for detailed protocols and support products.