

Reference protein:

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1 MDYSYDEDLD ELCPVCGDKV SGYHYGLLTC ESCKGFFKRT VQNNKHYTCT ESQSCKIDKT
61 QRKRCPFQRF QKCLTVGMRL EAVRADRMRG GRNKFGPMYK RDRALKQQKK AQIRANGFKL
121 ETGPPMGVPP PPPPAPDYVL PPSLHGPEPK GLAAGPPAGP LGDFGAPALP MAVPGAHGPL
181 AGYLYPAFFG RAIKSEYPEP YASPPQGPL YGYPEPFSGG PNVPELILQL LQLEPDEDQV
241 RARILGCLQE PTKSRPDQPA AFGLLCRMAD QTFISIVDWA RRCMVFKELE VADQMTLLQN
301 CWSELLVFDH IYRQVQHGE GSILLVTGQE VELTTVATQA GSSLHSLVLR AQELVLQLLA
361 LQLDRQEFVC LKFIILFSLD LKFLNNHILV KDAQEKANAA LLDYTLCHYP HCGDKFQQLL
421 LCLVEVRALS MQAKEYLYHK HLGNEPRNN LLIEMLOAKQ T*
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Protein predicted from variant coding sequence:

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1 MDYSYDEDLD ELCPVCGDKV SGYHYGLLTC ESCKGFFKRT VQNNKHYTCT ESQSCKIDKT
61 QRKRCPFQRF QKCLTVGMRL EAVRADRMRG GRNKFGPMYK RDRALKQQKK AQIRANGFKL
121 ETGPPMGVPP PPPPAPDYVL PPSLHGPEPK GLAAGPPAGP LGDFGAPALP MAVPGAHGPL
181 AGYLYPAFFG RAIKSEYPEP YASPPQGPL YGYPEPFSGG PNVPELILQL LQLEPDEDQV
241 RARILGCLQE PTKSRPDQPA AFGLLCRMAD QTFISIVDWA RRCMVFKELE VADQMTLLQN
301 CWSELLVFDH IYRQVQHGE GSILLVTGQE VELTTVATQA GSSLHSLVLR AQELVLQLAC
361 AAAGPAGVCL PQVHHPLQPG FEVPE*
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Affected transcript:

NM_004959.4:c.1074dupG

Affected protein:

NM_004959.4(NR5A1_i001):p.(Leu359A)fs*28

Supplemental Figure 1. Prediction of the mutated protein using Name checker by Mutalyzer 2.0.beta-26 (<http://www.lovd.nl/mutalyzer/>).

Supplemental Table 1. **Features lost in the mutated protein (p.Leu359Alafs*28), obtained by using Mutation Taster analysis.**

START (AA)	END (AA)	FEATURE DETAILS	CONSEQUENCE
230	461	Region Important for dimerization.	lost
342	360	HELIX	lost
365	376	HELIX	lost
381	383	HELIX	lost
387	408	HELIX	lost
411	413	STRAND	lost
415	442	HELIX	lost
433	433	MUTAGEN A>F: Strongly reduced transactivation	lost
436	436	BINDING Lipid headgroup	lost
436	436	MUTAGEN Y>F: Loss of transactivation; when associated with A440	lost
440	440	BINDING Lipid headgroup.	lost
440	440	MUTAGEN K>A: Loss of transactivation; when associated with F436	lost
451	456	HELIX	lost