

ANALYTE:
ATP6V0A2

NAME:	ATPase H ⁺ transporting V0 subunit a2
SYMBOL:	ATP6V0A2
VERSION OF ORPHANET:	2023-06-22 14:14:43
SYNONYMS:	ATP6N1D ATP6a2 J6B7 RTF Stv1 TJ6 TJ6M TJ6s V-ATPase subunit a2 V-type proton ATPase 116 kDa subunit a2 Vph1 a2 a2V regeneration and tolerance factor

XREF(S):	Orphanet Reactome SwissProt Ensembl Genatlas HGNC OMIM
CREATED:	13 May 2019 - 01:01
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- [Congenital disorders of glycosylation \(79 genes\)](#)
- [Congenital malformation \(gene panel - 1721 genes\)](#)
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- [Dermatogenetic panel, severe, rare and hereditary genodermatoses \(gene panel - 394 genes\)](#)
- [Early onset epileptic encephalopathy \(gene panel - 845 genes\)](#)
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- [Intellectual disability \(gene panel\)](#)
- [Intellectual disability \(virtual gene panel\)](#)
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- [Neurodevelopmental disorders \(1300 genes\)](#)
- [Neurodevelopmental disorders gene panel](#)
- [Primary immune deficiencies \(gene panel\)](#)
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- [Skeletal dysplasia \(gene panel\)](#)
- [Skeletal dysplasia \(gene panel\)](#)

Related Diseases

- Autosomal recessive cutis laxa type 2, classic type
- Wrinkly skin syndrome

Related Gene Panels

- Congenital disorders of glycosylation (79 genes) - KUL
- Congenital malformation (1721 genes) - ULB
- Congenital malformation gene panel - VUB
- Cutis Laxa / Geroderma osteodysplasticum - UGent
- Dermatogenetic / severe, rare and hereditary genodermatoses (394 genes) - ULB
- Early onset epileptic encephalopathy (845 genes) - ULB
- Epilepsy gene panel - VUB
- Intellectual disability & Epilepsy - UGent
- Intellectual disability (gene panel)
- Intellectual disability/Epilepsy (1091 genes) - ULG
- Malformations of cortical development (235 genes) - VUB
- Metabolic disorders (213 genes) - VUB
- Neurodevelopmental disorders (1300 genes) - ULB
- Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders (1162 genes) - VUB
- Primary immune deficiencies (444 genes) - KUL
- Skeletal dysplasia (394 genes) - VUB
- Skeletal dysplasia (genepanel) - UZA
- Skeletal dysplasia - UGent

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