Specific information metabolic diseases

(Version January 2026)

Website links:
Amount of patient material to submit
Turnaround times mutational scanning and prices

Adrenal hypoplasia, congenital

Adrenal Hypoplasia Congenital

OMIM: # 300200

Gene	Method
NR0B1 (DAX1)	Sequence analysis of the entire coding region (exon 1 to 2) including intron/exon junctions

Procedure:
Sequence analysis of the NR0B1 gene (DAX1).

Detection ratio:
~33% % (8/24)

Gene	Gene product	Locus	Inheritance	OMIM number	Reference sequence
NR0B1	DAX1	Xp21.2	X-linked recessive	300473	NM_000475.4; NT_011757.12

Cystinuria

Cystinuria

OMIM: # 220100

Gene	Method
SLC3A1	Sequence analysis of the entire coding region (exon 1 to 10) including intron/exon junctions
SLC7A9	Sequence analysis of the entire coding region (exon 2 to 13) including intron/exon junctions

Procedure:
Sequence analysis of the genes SLC3A1 en SLC7A9

Gene	Gene product	Locus	Inheritance	OMIM number	Reference sequence
SLC3A1	SLC3A1-protein	2p21	Autosomal dominant and autosomal recessive	104614	NM_00341.3
SLC7A9	SLC7A9-protein	19q13.11	Autosomal dominant and autosomal recessive	604144	NM_001126335.1 / NC_000019.9