

Full name:	Cutis Laxa / Geroderma osteodysplasticum - UGent
Laboratory:	<u>Centrum Medische Genetica - UZ Gent</u>
Created:	11 Jun 2019 - 10:26
Changed:	14 Dec 2022 - 14:40

Related Diseases

- Autosomal dominant cutis laxa
- Autosomal recessive cutis laxa type 1
- Autosomal recessive cutis laxa type 2, classic type
- Autosomal recessive cutis laxa type 2B
- Geroderma osteodysplastica

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ALDH18A1</u>	100.00	1	
<u>ATP6V0A2</u>	100.00	1	
<u>ATP6V1A</u>	100.00	1	
<u>ATP6V1E1</u>	100.00	1	
<u>ATP7A</u>	100.00	1	
<u>COG7</u>	100.00	1	
<u>EFEMP1</u>	100.00	1	
<u>EFEMP2</u>	100.00	1	
<u>ELN</u>	100.00	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>FBLN5</u>	100.00	1	
<u>GORAB</u>	100.00	1	
<u>LOX</u>	100.00	1	
<u>LTBP1</u>	100.00	1	
<u>LTBP4</u>	100.00	1	
<u>NAA10</u>	100.00	1	
<u>PYCR1</u>	100.00	1	
<u>RIN2</u>	100.00	1	
<u>TALDO1</u>	100.00	1	