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Full name:	Hot spot mutation among Jewish (4 genes, 7 mutations) - UZA
Quality coverage:	c.854A>C (p.Glu285Ala) and c.693C>A (p.Tyr231*) in ASPA; c.1274_1277dupTATC, c.1421+1G>C and c.805G>A (p.Gly269Ser) in HEXA; c.2204+6T>C in ELP1 (IKBKAP); c.456+4A>T in FANCC
Created:	05 Jul 2019 - 14:33
Changed:	21 Aug 2019 - 15:41

Related Diseases

- Familial dysautonomia
- Fanconi anemia
- Severe Canavan disease
- Tay-Sachs disease, B1 variant

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
ASPA			
ELP1			
FANCC			
HEXA			