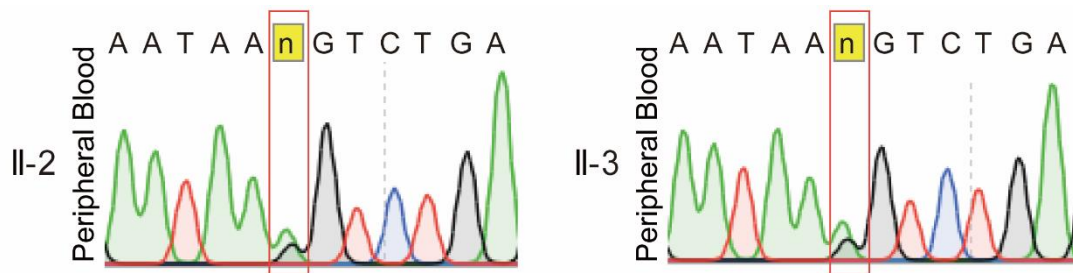


Scientific Letters

Hb H Disease Caused by Uniparental Disomy: First Report of the $\alpha^{T-Saudi}\alpha$ Mutation in the Chinese Population

Supplementary Data



Supplementary Figure 1. Sanger sequencing results for samples of peripheral blood cells (II-2 and II-3). The red box indicates the mutation site.

Supplementary Table 1. Summary of reported cases of hemoglobinopathies caused by UPD

Related gene	Disease	Genotype	Source	Techniques for identifying UPD	UPD pattern	mosaicism	Reference
α -globin	Abnormal hemoglobin (Hb Bronovo)	$\alpha^{\text{Bronovo}}\alpha/\alpha^{\text{Bronovo}}\alpha$	maternal	STR	isodisomy and heterodisomy	n.d	PMID: 32552204
	Hb Bart's hydroptic fetus	$--_{\text{SEA}}/--_{\text{SEA}}$	maternal	STR	isodisomy and heterodisomy	n.d	PMID: 18564363
	Hb Bart's hydroptic fetus	$--_{\text{SEA}}/--_{\text{SEA}}$	maternal	STR	isodisomy	n.d	PMID: 26574185
	Hb Bart's hydroptic fetus	$--_{\text{SEA}}/--_{\text{SEA}}$	maternal	CMA	n.d	n.d	PMID: 34434471
	Hb H disease	$\alpha^{\text{T-Saudi}}\alpha/\alpha^{\text{T-Saudi}}\alpha$	maternal	CMA	isodisomy and heterodisomy	no	This study
β -globin	β -thalassemia major	$\beta^{\text{CD15}}/\beta^{\text{CD15}}$	paternal	CMA	isodisomy	yes	PMID: 22875618
	β -thalassemia major	$\beta^{\text{CD17}}/\beta^{\text{CD17}}$	paternal	CMA	isodisomy	yes	PMID: 18413893
	β -thalassemia major	$\beta^{\text{CD39}}/\beta^{\text{CD39}}$	paternal	RFLP	isodisomy	n.d	PMID: 1351764
	β -thalassemia major	$\beta^{\text{CD76}}/\beta^{\text{CD76}}$	paternal	CMA	isodisomy	yes	
	β -thalassemia major	$\beta^{\text{CD39}}/\beta^{\text{CD39}}$	unknown	CMA	isodisomy	yes	PMID: 22983591
	β -thalassemia major	$\beta^{\text{CD39}}/\beta^{\text{CD39}}$	paternal	CMA	isodisomy	yes	
	Abnormal hemoglobin (sickle cell anemia)	$\beta^{\text{S}}/\beta^{\text{S}}$	paternal	STR	isodisomy	yes	PMID: 20595515

n.d: not detected. STR: short tandem repeat markers. CMA: chromosomal microarray analysis. RFLP: restriction fragment length polymorphism.

Supplementary Table 2. Summary of reported cases carrying $\alpha^{T-Saudi}\alpha$ mutation

Country	α -Genotype	Number	Sum	Reference
Israel	$\alpha^{T-Saudi}\alpha/\alpha^{T-Saudi}\alpha$	1	5	PMID: 1281602
	$\alpha^{T-Saudi}\alpha/\alpha^{Hb\ Taybe}$	1		PMID: 7942784
	$\alpha^{T-Saudi}\alpha/\alpha\alpha$	3		
Azerbaijan	$\alpha^{T-Saudi}\alpha/\alpha^{T-Saudi}\alpha$	5	5	PMID: 31755088
Greece	$\alpha^{T-Saudi}\alpha/-\alpha$	4	11	PMID: 10569721
	$\alpha^{T-Saudi}\alpha/\alpha\alpha$	1		PMID: 8136277
	$\alpha^{T-Saudi}\alpha/\alpha^{Agr}\alpha$	1		
	$\alpha^{T-Saudi}\alpha/\alpha^{T-Saudi}\alpha$	4		PMID: 8213764
	$\alpha^{T-Saudi}\alpha/\alpha\alpha^T$	1		
Nothern Cyprus	$\alpha^{T-Saudi}\alpha/\alpha^{T-Saudi}\alpha$	3	12	PMID: 1281602
	$\alpha^{T-Saudi}\alpha/\alpha\alpha$	4		PMID: 8136277
	$\alpha^{T-Saudi}\alpha/\alpha\alpha$	3		
	$\alpha^{T-Saudi}\alpha/\alpha^{Agr}\alpha$	2		
Iraq	$\alpha^{T-Saudi}\alpha/\alpha\alpha$	1	14	PMID: 19205971
	$\alpha^{T-Saudi}\alpha/\alpha\alpha$	8		PMID: 34121736
	$-\alpha^{3,7}/\alpha^{T-Saudi}\alpha$	4		
	$-\alpha^{3,7,4,2}/\alpha^{T-Saudi}\alpha$	1		
Saudi Arabia	$\alpha^{T-Saudi}\alpha/\alpha\alpha$	4	19	PMID: 19371220
	$\alpha^{T-Saudi}\alpha/\alpha^{T-Saudi}\alpha$	4		
	$-\alpha^{4,2}/\alpha^{T-Saudi}\alpha$	1		
	$-\alpha^{3,7}/\alpha^{T-Saudi}\alpha$	8		PMID: 17240829
	$\alpha^{T-Saudi}\alpha/\alpha^{\Delta G}\alpha$	1		
UAE	$\alpha^{T-Saudi}\alpha/\alpha\alpha$	1	23	PMID: 9691147
	$\alpha^{T-Saudi}\alpha/\alpha^{T-Saudi}\alpha$	1		
	$-\alpha^{3,7}/\alpha^{T-Saudi}\alpha$	9		PMID: 22074123
	$\alpha^{T-Saudi}\alpha/\alpha^{T-Saudi}\alpha$	3		
Tunisia	$\alpha^{T-Saudi}\alpha/\alpha^{T-Saudi}\alpha$	9	26	PMID: 1281602
	$\alpha^{T-Saudi}\alpha/\alpha\alpha$	1		PMID: 12603097
	$\alpha^{T-Saudi}\alpha/\alpha^{T-Saudi}\alpha$	5		PMID: 19147907
	$\alpha^{T-Saudi}\alpha/\alpha^{T-Saudi}\alpha$	6		PMID: 12484630
	$\alpha^{T-Saudi}\alpha/\alpha\alpha$	9		
Turkey	$\alpha^{T-Saudi}\alpha/\alpha^{T-Saudi}\alpha$	4	24	PMID: 1281602
	$\alpha^{T-Saudi}\alpha/\alpha\alpha$	5		PMID: 11480787
	$\alpha^{T-Saudi}\alpha/\alpha\alpha$	2		
	$\alpha^{T-Saudi}\alpha/\alpha^{T-Saudi}\alpha$	1		PMID: 26377141
	$\alpha^{T-Saudi}\alpha/\alpha\alpha$	4		
	$\alpha^{T-Saudi}\alpha/\alpha^{T-Saudi}\alpha$	1		PMID: 17486510
	$\alpha^{T-Saudi}\alpha/\alpha^{inti.CD***}\alpha$	1		PMID: 20332613
	$\alpha^{T-Saudi}\alpha/\alpha^{T-Saudi}\alpha$	1		
	$--MED/\alpha^{T-Saudi}\alpha$	1		
$\alpha^{T-Saudi}\alpha/\alpha\alpha$	3			

	$\alpha^{T-Saudi}/\alpha/\alpha$	1		PMID: 23419704
	$\alpha^{T-Saudi}/\alpha/\alpha$	1		PMID: 12028055
	..MED/ $\alpha^{T-Saudi}/\alpha$	1		
Kuwait	$\alpha^{T-Saudi}/\alpha/\alpha$	7	65	PMID: 7701914
	$\alpha^{T-Saudi}/\alpha/\alpha^{5nt}$	1		
	$-\alpha^{3.7}/\alpha^{T-Saudi}/\alpha$	4		
	$\alpha^{T-Saudi}/\alpha/\alpha^{T-Saudi}/\alpha$	13		
	$\alpha^{T-Saudi}/\alpha/\alpha^{T-Saudi}/\alpha$	17		
	$-\alpha^{3.7}/\alpha^{T-Saudi}/\alpha$	6		
	$\alpha^{T-Saudi}/\alpha/\alpha^{T-Saudi}/\alpha$	11		
	$-\alpha^{3.7}/\alpha^{T-Saudi}/\alpha$	6		PMID: 26076396
Oman	$-\alpha^{3.7}/\alpha^{T-Saudi}/\alpha$	1	75	PMID: 21410534
	$\alpha^{T-Saudi}/\alpha/\alpha^{\Delta 5nt}$	1		
	$\alpha^{T-Saudi}/\alpha/\alpha^{T-Saudi}/\alpha$	28		
	$-\alpha^{3.7}/\alpha^{T-Saudi}/\alpha$	2		PMID: 32019385
	$\alpha^{T-Saudi}/\alpha/\alpha^{codon 19}$	1		
	$\alpha^{T-Saudi}/\alpha/\alpha$	12		PMID: 20353348
	$\alpha^{T-Saudi}/\alpha/\alpha^{T-Saudi}/\alpha$	16		
	$\alpha^{T-Saudi}/\alpha/\alpha^{T-Saudi}/\alpha$	3		PMID: 23668236
	$\alpha^{T-Saudi}/\alpha/\alpha^{T-Saudi}/\alpha$	4		
	$-\alpha^{3.7}/\alpha^{T-Saudi}/\alpha$	1		PMID: 9691147
	$\alpha^{\Delta 5nt}/\alpha/\alpha^{T-Saudi}/\alpha$	1		
	$\alpha^{T-Saudi}/\alpha/\alpha^{\Delta G}$	1		PMID: 22074123
	$\alpha^{T-Saudi}/\alpha/\alpha$	1		
	$\alpha^{T-Saudi}/\alpha/\alpha^{T-Saudi}/\alpha$	1		PMID: 25370869
$-\alpha^{4.2}/\alpha^{T-Saudi}/\alpha$	1			
$-\alpha^{3.7}/\alpha^{T-Saudi}/\alpha$	1			
Iran	$\alpha^{T-Saudi}/\alpha/\alpha$	5	109	PMID: 24826792
	$\alpha^{T-Saudi}/\alpha/\alpha$	3		
	$\alpha^{T-Saudi}/\alpha/\alpha^{T-Saudi}/\alpha$	3		PMID: 19065332
	$\alpha^{T-Saudi}/\alpha/\alpha^{T-Saudi}/\alpha$	6		
	..MED/ $\alpha^{T-Saudi}/\alpha$	3		PMID: 21250880
	$-\alpha^{3.7}/\alpha^{T-Saudi}/\alpha$	1		
	$\alpha^{T-Saudi}/\alpha/\alpha^{T-Saudi}/\alpha$	7		PMID: 26523940
	..MED/ $\alpha^{T-Saudi}/\alpha$	2		
	$\alpha^{T-Saudi}/\alpha/\alpha$	21		PMID: 31273613
	$\alpha^{T-Saudi}/\alpha/\alpha$	4		
	$-\alpha^{3.7}/\alpha^{T-Saudi}/\alpha$	2		PMID: 28385057
	$\alpha^{T-Saudi}/\alpha/\alpha$	2		
	$\alpha^{T-Saudi}/\alpha/\alpha$	50		PMID: 19657838
Jordan	$\alpha^{T-Saudi}/\alpha/\alpha$	108	133	(1)
	$\alpha^{T-Saudi}/\alpha/\alpha^{T-Saudi}/\alpha$	20		
	..MED/ $\alpha^{T-Saudi}/\alpha$	5		
Bahrain	$\alpha^{T-Saudi}/\alpha/\alpha$	5	229	PMID: 26878087
	$\alpha^{T-Saudi}/\alpha/\alpha^{T-Saudi}/\alpha$	13		
	$\alpha^{T-Saudi}/\alpha/\alpha^{Hph-1}$	3		
	$-\alpha^{3.7}/\alpha^{T-Saudi}/\alpha$	6		PMID: 22401170

$\alpha^{T-Saudi}\alpha/\alpha^{PA-2}\alpha$	1	
$\alpha^{T-Saudi}\alpha/\alpha^{T-Saudi}\alpha$	32	
$\alpha^{T-Saudi}\alpha/\alpha^{Hph}\alpha$	29	
$-\alpha^{4.2}/\alpha^{T-Saudi}\alpha$	22	PMID: 30864492
$-\alpha^{3.7}/\alpha^{T-Saudi}\alpha$	46	
$\alpha^{T-Saudi}\alpha/\alpha\alpha$	28	
$\alpha^{T-Saudi}\alpha/\alpha\alpha$	6	
$\alpha^{T-Saudi}\alpha/\alpha^{T-Saudi}\alpha$	26	
$\alpha^{T-Saudi}\alpha/\alpha^{T-Turkish}\alpha$	1	(2)
$-\alpha^{3.7}/\alpha^{T-Saudi}\alpha$	11	

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