

Nature and distribution of unique AR mutations that cause disease

Loss of function disease	Type of mutation	N-terminal domain ¹	DNA-binding domain ²	Hinge region ³	Ligand-binding domain ⁴	Splice site	Intron
CAIS	Single base substitution	8	26		119	15	1
	Premature termination	33	4	1	18		
	Complete gene deletion	4					
	Partial gene deletion	9	8		4*		
	Deletion (1-4 bases)	19	4		10*		
	Insertion	11	2*		3*	1	
	Duplication Indel	2	3*		1* 1*		
PAIS	Single base substitution	9	20	3	93	2	1*
	Multiple base substitution		1		1		
	Premature termination	2					
	Deletion (1-4 bases)	2			2		3
MAIS	Single base substitution	22	4*		15		
	Partial gene deletion				1 ?		
	Deletion (1-4 bases)				1		
Gain of function disease						UTR	
Prostate cancer⁵	Single base substitution	42	7	3	52		2
	Premature termination mutations	1			4		
	Deletion (1-4 bases)	2	2*	1*	1	1*	
	Insertion	1*				1	1
Breast cancer	Single base substitution		2			1	
Larynx cancer⁵	Deletion (30 bases)	1					
Liver cancer⁵	Single base substitution	4*			1*		
Testicular cancer⁵	Single base substitution	3*					

¹ aa 1-534; ² aa 559-624; ³ aa between DBD and LBD 625-663; ⁴ aa 664-919; ⁵ somatic mutation;

*Mutations not reported in 2004