TURNER SYNDROME KARYOTYPES

The "Classic" TS Karyotype is 45,x (monosomy x), but there are many other possibilities.

Turner Syndrome Karyotypes		
Non-Mosiac		
Chr 45	**Chromosome 46**	**Chr 47**
X	None*	
None*	46,X,i(Xq) or isochromosome Xq	
None*	46,X,del(Xp) or X chromosome partial deletion	
Mosiac		
Chr 45	**Chromosome 46**	**Chr 47**
X	XX	
X	XY	
X	X,i(Xq)	
None*	46,X,del(Xp)	
None*	46,XX	
X		47,XXX
X	46,X,r(X) or ring X chromosome	
X	46,X,+mar or marker chromosome	
Rare karyotypes with 3 cell lines		

Typical Karyotype			
Female XX		**Male XY**	
Chr 45	**Chr 46**	**Chr 45**	**Chr 46**
X	X	X	Y

* **Monosomy** refers to a condition where every cell in the body is missing one X chromosome, meaning the person has only one X chromosome.

Mosaic indicates that only some cells in the body are missing an X chromosome, with other cells having a normal pair, resulting in a mixture of cell types within the individual; essentially, monosomy is a complete absence of one X chromosome in all cells, whereas mosaicism means only some cells lack the X chromosome while others have a normal pair.

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There are multiple possible karyotypes associated with Turner syndrome, including mosaic karyotypes where some cells have a normal XX karyotype and others have a 45,X karyotype, as well as variations with structural abnormalities on the X chromosome, meaning there are several different Turner Syndrome karyotypes that can occur depending on the specific chromosomal deletion or mutation involved.

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Turner Syndrome Society of the United States®

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