

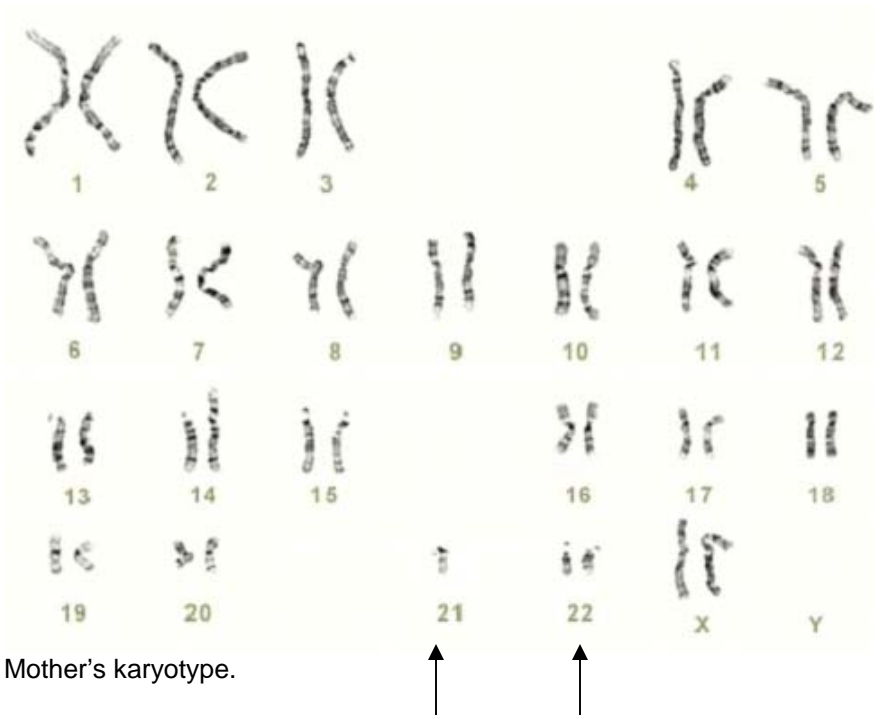
HOLY C.O.W.!

IT'S...

Clinical Question of the Week #17
 October 20th, 2008 through October 27th, 2008

Please e-mail your answers to Kuo, Tim, Wendy, and Kevin (klian@mednet.ucla.edu; tprovias@mednet.ucla.edu; wsimon@mednet.ucla.edu; kbreger@mednet.ucla.edu) by 0800 on Monday, October 27th, 2008. The resident or intern with the most correct answers at the end of each month will receive a prize!

Case: A 37-year-old woman presents for evaluation in family planning. She is healthy and does not drink, smoke, or use illicit drugs. She was adopted and her family history is unknown. She has been married to her husband for seven years and they have a three year old son. Her husband is healthy and has a family history notable for hypertension, but no other known disease. She notes that her son has progressed slowly in his development, with delayed onset of crawling and walking. He said his first word, "mommy" at eighteen months of age, but now has several words he uses to communicate. His height and weight are in the 30th percentile range. Physical examination of the mother reveals no abnormalities. Recent progress note from the son's pediatrician notes the developmental delay as described and also notes dysmorphic features on examination. A karyotype is obtained on the mother, which is shown below. Likewise, genetic analysis is performed also on the son.



Mother's karyotype.

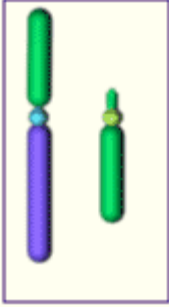


Diagram of a subset of chromosomes inherited by the son from the mother.

Questions:

1. Name the condition that the child has.
2. What is the genetic defect, and how does it occur?
3. Name two other causes of the child's genetic defect.
4. If the mother has another child, what are her chances of having a normal baby? A miscarriage?