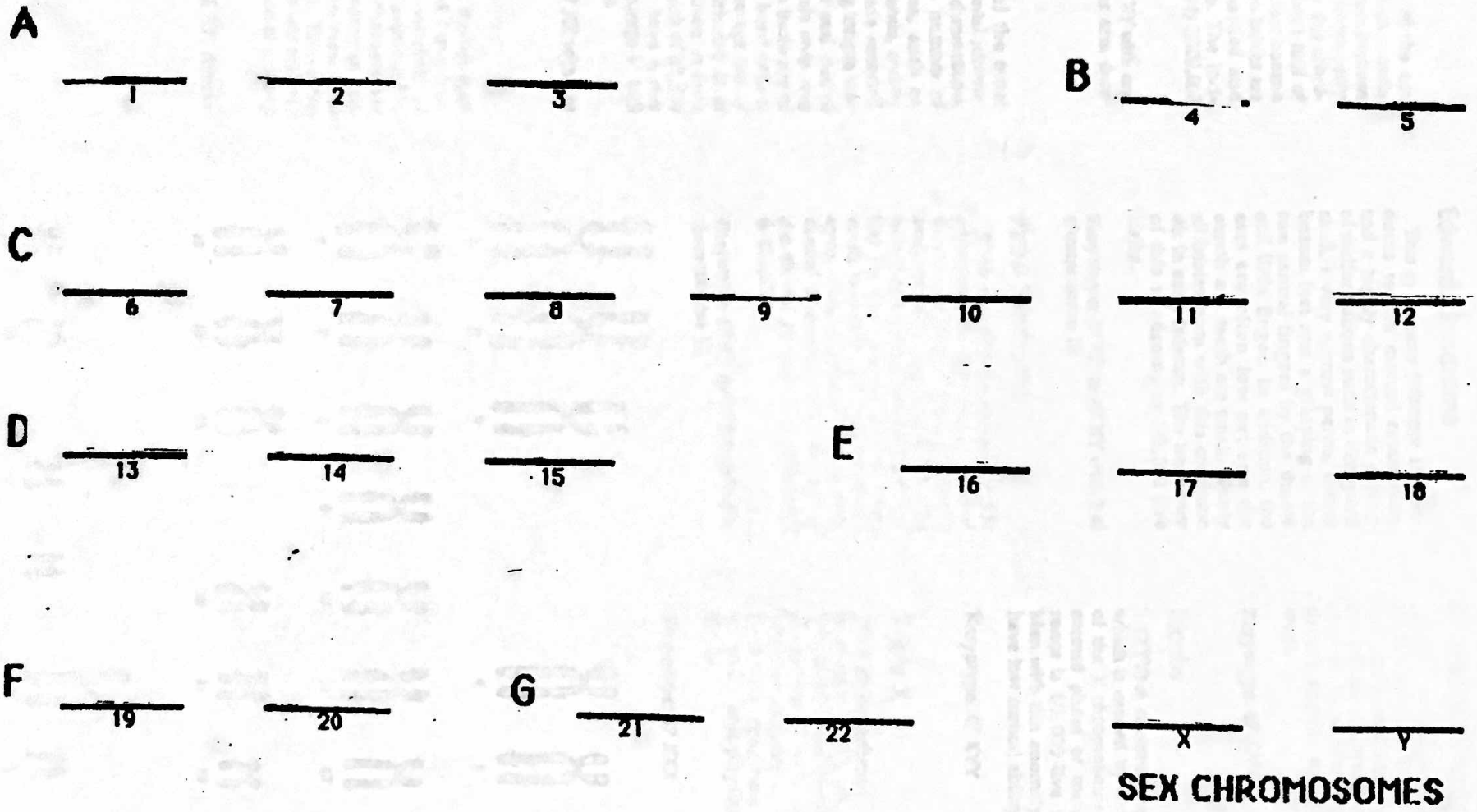
HUMAN KARYOTYPE

From Smear
Number _____



Number of Chromosomes

Sex of Subject

Type of Disorder

Cri-du-chat

Babies with the cry of the cat syndrome have a cry which sounds like that of a cat in distress; because the infant's larynx is improperly developed. The cause of this condition is a deletion of about half of the short arm of chromosome number five. Cri-du-chat babies are severely mentally retarded and have a small cranium. The incidence of this syndrome is 1/100,000 live births.

Karyotype: 46 XX or 46 XY with one chromosome #5 upper arm deletion.

Down Syndrome

Trisomy 21, one of the most common causes of mental retardation is due to an extra chromosome 21. This results in a number of characteristic features, such as short stature, broad hands, stubby fingers and toes, a wide rounded face, a large protruding tongue that makes speech difficult and mental retardation. Individuals with this syndrome have a high incidence of respiratory infections, heart defects and leukemia. The average risk of having a child with trisomy 21 is 1/750 live births. Mothers in their early twenties have a risk of 1/1,500 and women over 35 have a risk factor of 1/70, which jumps to 1/25 for women 45 or older.

Karyotype: 47 XY or 47 XX with 3 of chromosome 21

Down Syndrome

14-21 translocation. Rather than trisomy 21, this form of Down syndrome has a chromosone count of 46, but an extra chromosome 21 has become attached to chromosome 14. Signs and symptoms of this anomaly are identical to those associated with trisomy 21. The carrier of the translocation is normal but has a 50 percent chance of passing it on.

Karyotype: 46 XX or 46 XY, translocation 14/21

Edwards Syndrome

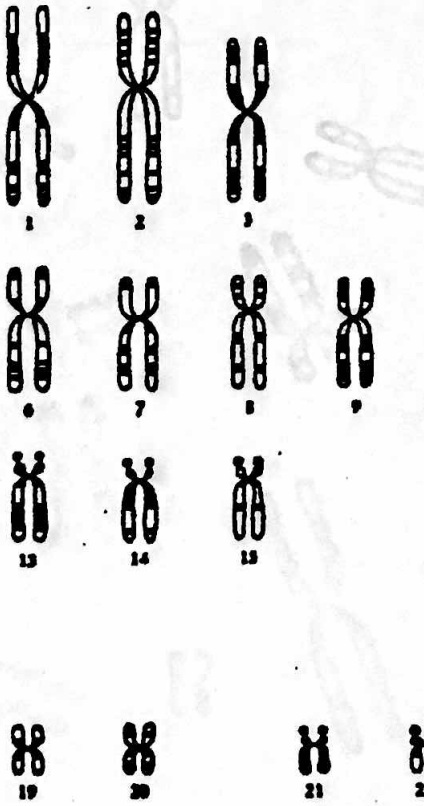
This syndrome (trisomy 18) produces severe mental retardation and a highly characteristic pattern of malformations such as elongated skull, a very narrow pelvis, rocker bottom feet and a grasping of the two central fingers by the thumb and little finger. In addition, the ears are often low set and the mouth and teeth are small. Nearly all babies born with this condition die in early infancy. The frequency of this syndrome is 1/5,000 live births.

Karyotype: 47 XX or 47 XY with 3 of chromosome 18

Patau Syndrome

This syndrome (trisomy 13) causes severely abnormal cerebral functions and virtually always leads to death in early infancy. This baby has very pronounced clefts of the lip and palate, broad nose, small cranium and nonfunctional eyes. Heart defects and severe mental retardation are also part of the clinical picture. The frequency is 1/15,000.

Karyotype: 47 XX or 47 XY with 3 of chromosome 13.



Klinefelter Syndrome

(XXY) A condition occuring in 1/1,000 male live births. Characteristics associated with this condition are tall stature, small testicles, and sterility. Most men with this syndrome appear normal in other ways.

Karyotype: 47 XXY

Jacobs

(XYY) A chromosome aberration which is caused by nondisjunction of the Y chromosome during the second phase of meiosis. Occurrence is 1/1,000 live male births. Men with this anomaly are tall and have low mental ability.

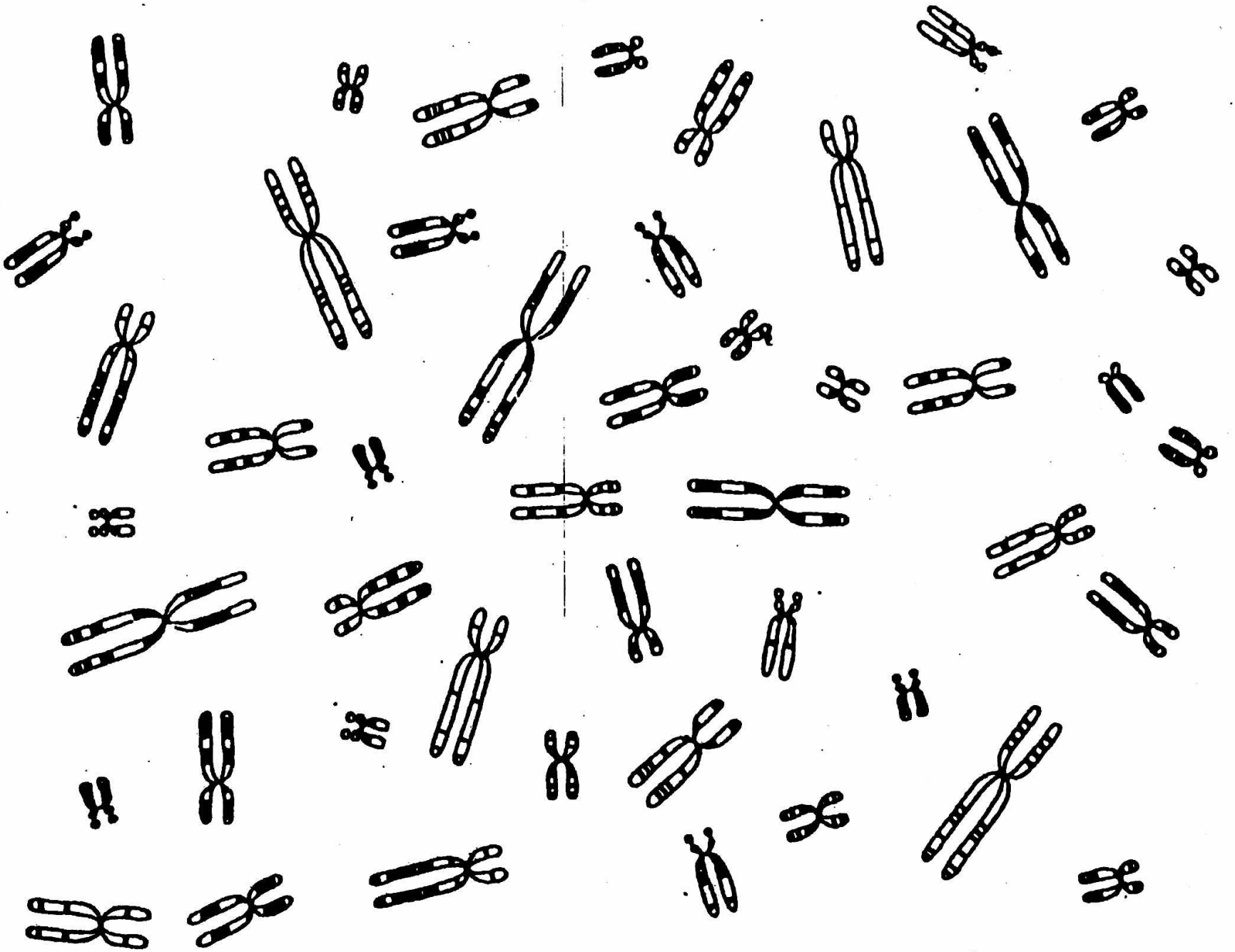
Karyotype: 47 XYY

Triple X

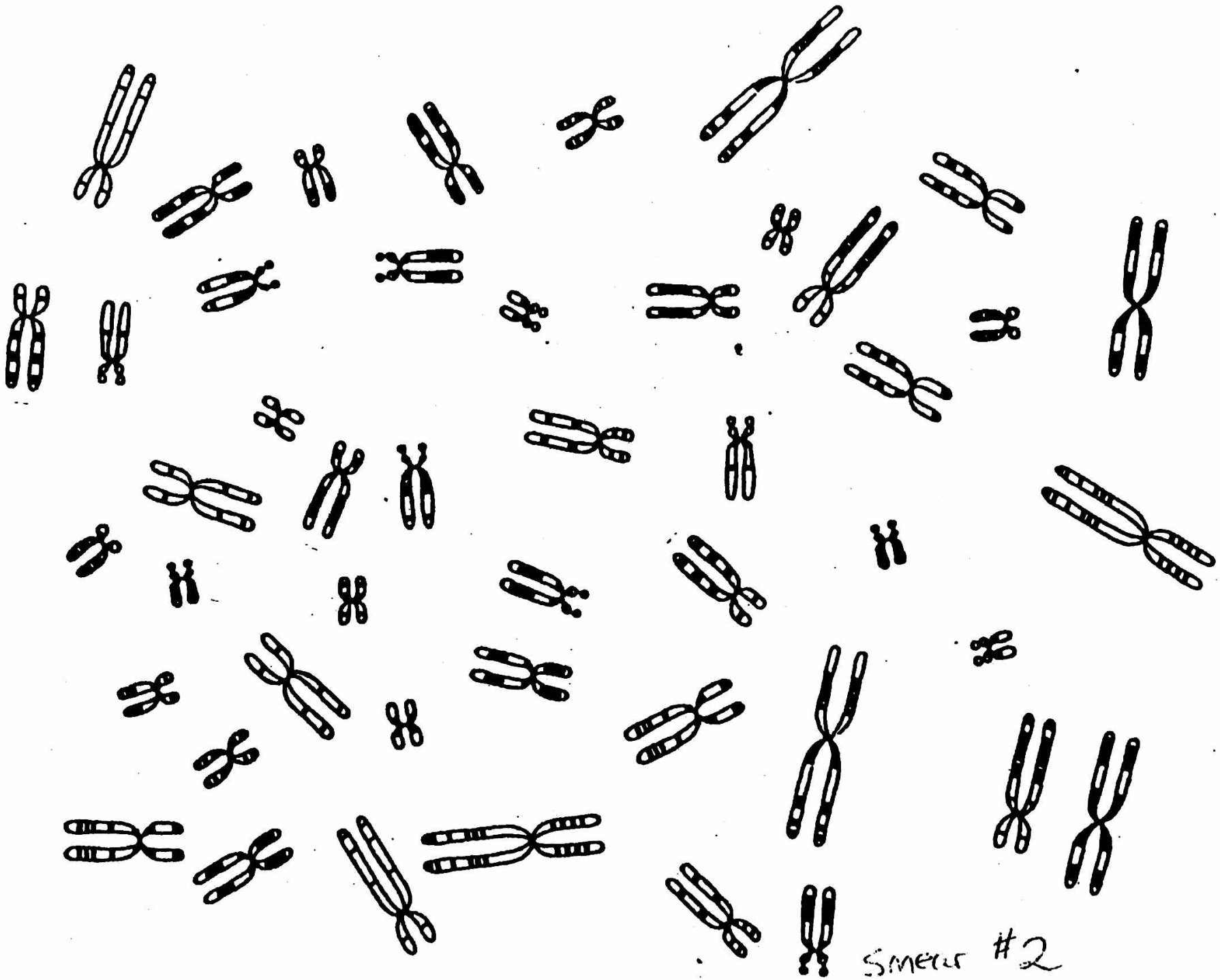
(XXX or superfemale) Occurs at a frequency of 1/1,000 female live births. No specific abnormalities are associated with this condition. The vast majority of women who have this condition are normal mentally and physically and are fertile.

Karyotype: 47 XXX

EXAMPLE OF CHROMOSOME 5 WITH UPPER ARM DELETION



Smear #1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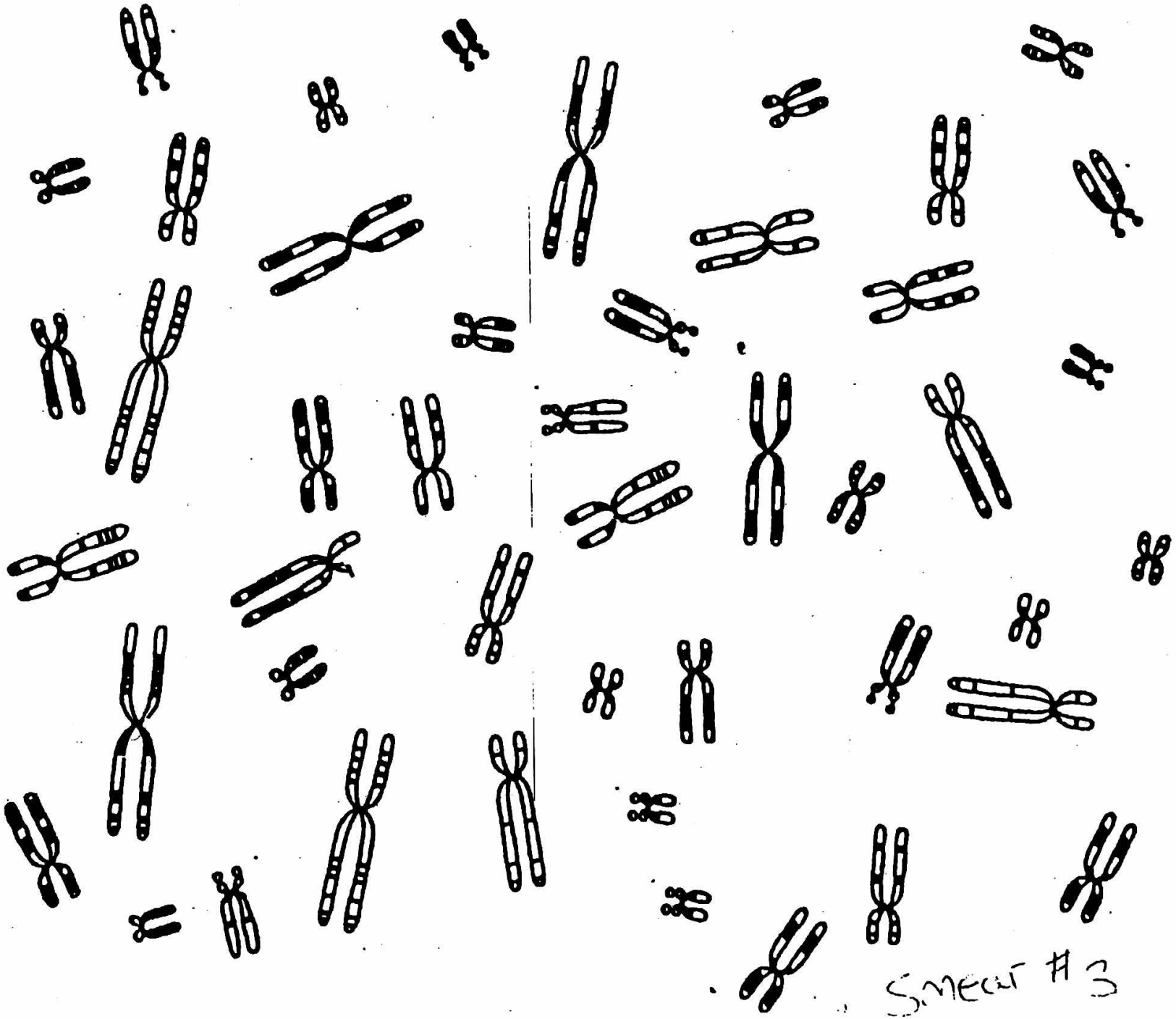
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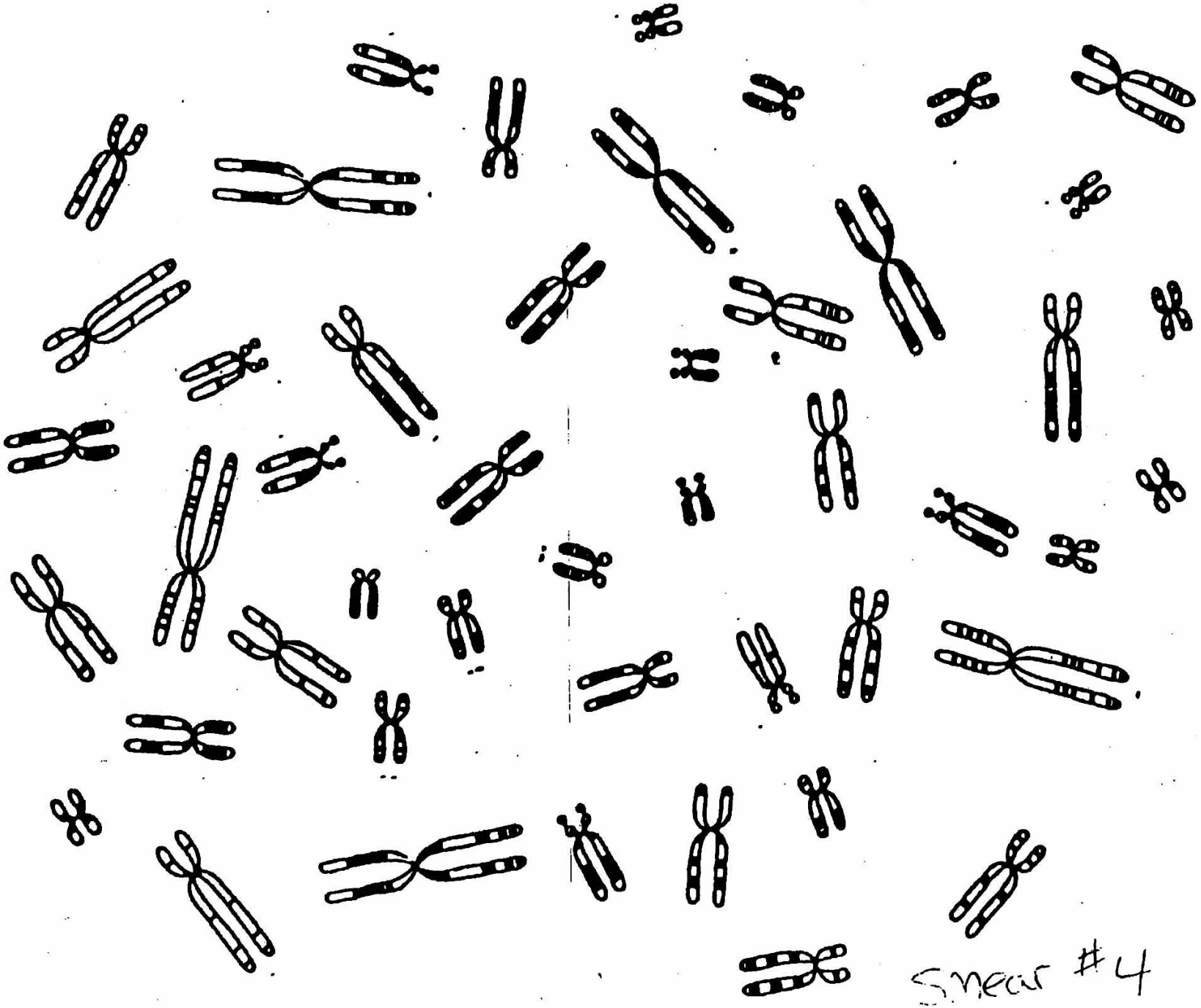
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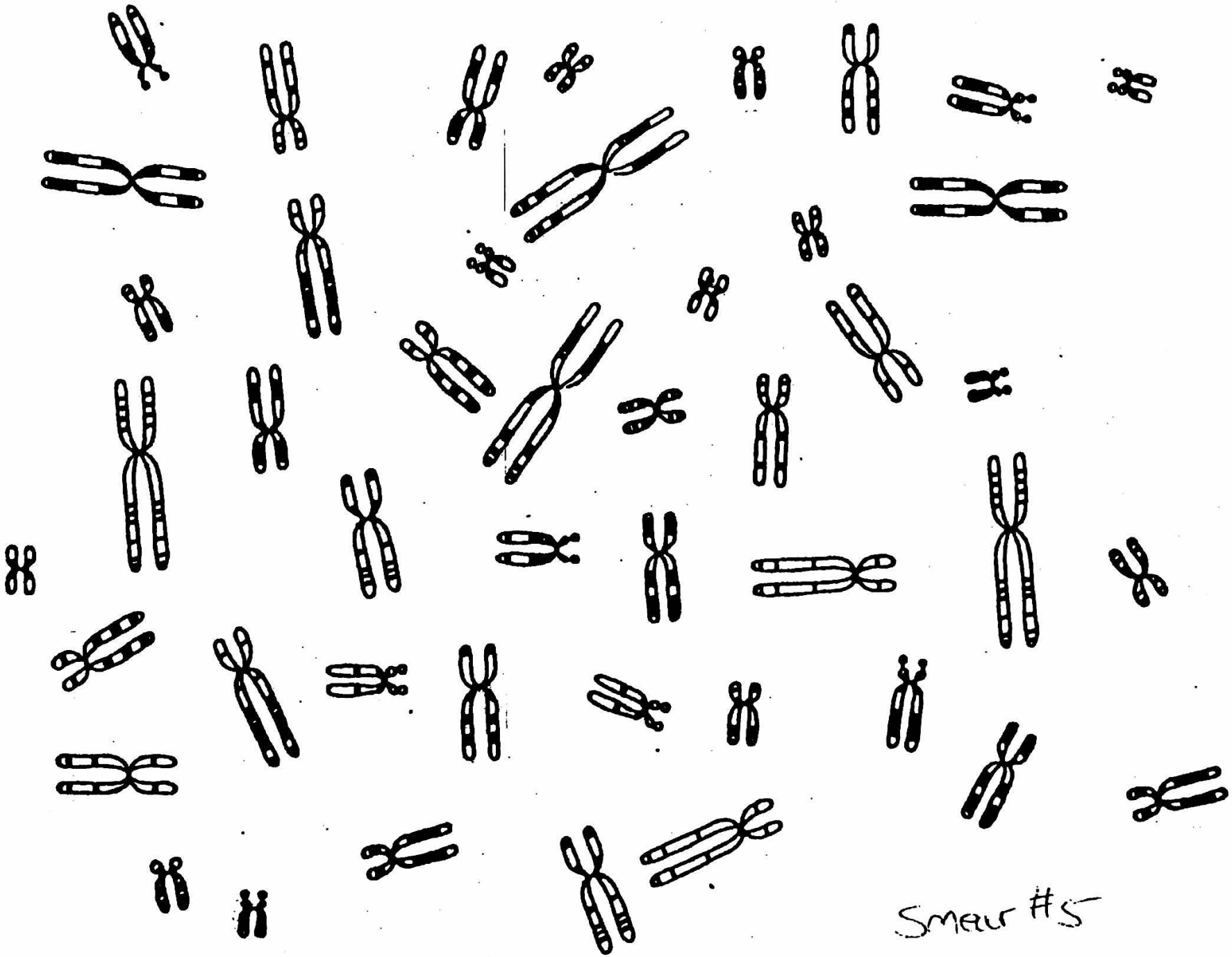
Smear #5