A fragile X and female retardation

An X chromosome with a fragile, long arm is known to cause the second most common form of mental retardation. But unlike the most common form, Down’s syndrome, “fragile X” retardation is inherited. Down’s syndrome is caused by a non-heritable fault in the mechanics of sperm or egg production (SN: 8/18/79, p. 117), with the result that the offspring receives three copies of chromosome 21 instead of the usual two. Like other so-called “sex-linked” or X chromosome-associated abnormalities, fragile X mental retardation was believed to affect only males.

A report in the Sept. 18 NEW ENGLAND JOURNAL OF MEDICINE now shows that females, too, are susceptible, although their retardation is milder than that of males. In fact, the report, from Gillian Turner and her team at the Prince of Wales Children’s Hospital, Randwick, New South Wales, Australia, suggests that mildly retarded, physically normal girls should be screened to see if their retardation is due to a fragile X.

There are several reasons why only males were believed to be susceptible to fragile X retardation. More males than females are institutionalized for mental retardation, males have been found to possess the fragile X, and mothers of males with the fragile X also possess it but are usually of normal intelligence. But when Turner and her co-workers identified the fragile X in a girl with mild mental retardation, they began to look for the fragile X among girls attending schools for the mildly retarded.

Of the 128 girls the researchers investigated, 72 had no physical abnormalities and thus were candidates to possess the fragile X. Of these 72, five girls (seven percent) carried the fragile X. As further proof that the retardation of these five girls was due to the abnormal X, the researchers found retarded males in four of the families; chromosomal analysis revealed the presence of the fragile X in two of the boys. Moreover, the fragile X chromosome was detected in other, female relatives of all the five girls: All five of the mothers possessed the abnormal chromosome, as did the mildly retarded sister of one of the girls and 18 of the girls’ other female relatives, six of whom were mildly retarded.

Turner and her colleagues thus conclude that “the presence of the marker X should be suspected in mildly retarded but physically normal girls,” and that the identification of the chromosome in such girls is important, not only because they are at risk of passing the fragile X on to offspring, but because their identification can lead to the identification of other female carriers of the fragile X in their families. In other words, even if a female with a fragile X is not retarded, she faces a one-in-two risk of having a son with moderate mental retardation and a slight but definite risk of having a daughter with mild retardation.

If all males with a fragile X are retarded, why is it that some females are retarded and others are not? A possible explanation is provided in an accompanying editorial by Park S. Gerald of Children’s Hospital Medical Center in Boston. Because a male has only one X chromosome, he will experience retardation if his one X chromosome is a fragile X. But a female has two X chromosomes, and there is evidence that one of the X’s expresses itself in some cells in her body, while the other X expresses itself in other cells. Thus, if a female has one fragile X chromosome, and it happens to express itself in her brain cells, she will probably be retarded. But if the abnormal chromosome expresses itself in cells other than brain cells, then the female will probably be normal.

Jean Piaget: 1896-1980

Jean Piaget, who revolutionized the study of intellect much as Freud revolutionized study of the emotions, died last week in Geneva at the age of 84. He was “haunted,” he once said, “by the idea of discovering a sort of embryology of intelligence.” Piaget the psychologist came to the fore in helping to formulate the methodology of Piaget the child psychologist. In his attempt to see the world through children’s eyes, he rigorously studied their level, playing games with them for hours at a time while carefully questioning and observing. His own three children were the subjects of much of this research. Their incorrect answers to his questions fascinated Piaget the most, and eventually helped him form his theory of four stages of mental development.

The stages are based on the cognitive tasks accomplished in each. By applying separate criteria to periods ofintellectual growth, Piaget was able to chart the development of a child’s mind from that of a primitive to the level at which abstract thought takes place. He theorized that children in all cultures go through these stages in the same order and at roughly the same time. He believed, however, that intellectual development also depends on the child’s interaction with the environment.

These beliefs placed him squarely in the middle of the “nature versus nurture” controversy. In addition, cognitive development is now seen by many as a series of changes that is somewhat smoother than the discrete stages of his theory. The broad concept of Piaget’s work, however, is still widely accepted and he continues to be acclaimed as one of the most creative scientific thinkers of the century.

Piaget was the author of more than 50 books and monographs, and in 1955 established a research center in Geneva.

Mondays may be hazardous to your health

Mondays carry a heavy load of negative associations — back to work, back to school, wash day. To that burden, Canadian researchers now add the possibility of death from a sudden heart attack.

University of Manitoba scientists reviewed the records of nearly 4,000 Canadian men whose medical histories have been followed since 1948. Of the 63 men who died suddenly with no previous history of heart problems, 22, or one-third, died on a Monday. Moreover, they found that 75 percent of the deaths that occurred at work were on a Monday. In contrast, non-fatal myocardial infarctions and deaths of men with previous clinical evidence of heart disease, as well as deaths from cancer, were spread evenly throughout the week.

“Reintroductions to occupational stress, activity, or pollutants after a weekend respite may be factors precipitating the arrhythmias that are the presumed basis for sudden death,” the scientists write in the Sept. 19 JOURNAL OF THE AMERICAN MEDICAL ASSOCIATION. They also suggest psychological stress caused by return to work and natural fluctuations in people’s seven-day biological clocks as other possible causes for the Monday clusters.

But because blue Mondays can’t be eliminated without creating blue Tuesdays, Simon Rabkin, one of the authors of the study, has an alternate suggestion: “If further investigation shows us psychological stress is involved, what we’ll have to do is get people educated so they can modify their reactions to their jobs,” he says.