Patau K, Smith D W, Therman E, Inborn S L & Wagner H P. Multiple congenital anomaly caused by an extra autosome. *Lancet* **1**:790-3, 1960.

A syndrome of multiple developmental abnormalities is defined on the basis of a chromosome aberration, the presence of an extra D autosome in all cells. This paper relates the chromosomal abnormality to the process of nondisjunction and to the origin of congenital defects. [The *SCI*® indicates that this paper was cited 330 times in the period 1961-1977.]

Stanley L. Inhorn
State Laboratory of Hygiene
University of Wisconsin
Madison, WI 53706

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"The field of human cytogenetics had experienced a rebirth in the late 1950's as a result of technical improvements that permitted clear delineation of somatic cell chromosomes. Recent reports from Europe had linked numerical chromosome abnormalities with known development syndromes—Turner's syndrome, Klinefelter's syndrome, and mongolism or Down's syndrome.

"A junior faculty man at the University of Wisconsin, David Smith, was just beginning to work in the field of developmental abnormalities. He was naturally excited by these discoveries in cytogenetics and was determined to apply chromosome studies to the cases of multiple congenital malformations he was examining. Thus, Smith corresponded with Charles Ford, in Great Britain, about the possibility of sending him bone marrow specimens from these cases. Dr. Ford graciously agreed to collaborate.

"At the time, I was a new faculty person in the Department of Pathology, working with quantitative cytologic methods. Smith asked me if I could prepare the marrows in tissue culture media for shipment to Britain. This I assured him would present no problem. As we were preparing our first material, we received a communique from Ford that his results with specimens sent from other investigators were unsatisfactory, and so he advised against sending any samples.

"I related this discouraging turn of events to D. Murray Angevine, chairman of Pathology. Dr. Angevine asked why we needed to send specimens to Europe when we had a prominent cytogeneticist, Klaus Patau, in the department. Although I shared a laboratory suite with Patau, I had regarded him as a quantitative cytologist and was unaware of his extensive experience in plant and animal cytogenetics.

"Within a short period of time, fruitful collaboration began between Patau, his associate Eva Therman (later to become Mrs. Patau), myself. Smith and his fellows. Smith selected cases of multiple congenital malformations for the chromosome study. Case 5 was the patient reported in this Citation Classic. The reason that this paper is so commonly cited is that it was the first delineation of a newly described syndrome of chromosomal etiology. Turner's, Klinefelter's, and Down's syndromes were well known conditions waiting for an etiologic explanation. The syndrome described in Case 5 was heretofore unrecognized. Also mentioned in this paper were two cases of another chromosomal syndrome, the trisomy 18 syndrome, the first being our Case 11.

"How does one name a new syndrome? Patau argued that we should call it the D_1 trisomy syndrome, since cytogenetic techniques at that time did not permit distinction between chromosomes 13, 14, or 15 Since we had discovered two trisomy syndromes in our first 11 cases, he reasoned that trisomies would be described for all chromosomes, so that trisomies D_2 and D_3 would soon be found. Time and better techniques have proven that D_1 trisomy is really trisomy 13, and that trisomies 14 and 15 are always lethal, resulting in spontaneous abortion. 13 trisomy is often referred to as Patau's syndrome, an appropriate tribute to one of the pioneers in human cytogenetics."