# CLINICAL ATLAS OF HUMAN CHROMOSOMES (A WILEY MEDICAL PUBLICATION) pdf

## 1: OMIM Entry - # - CHROMOSOME 18p DELETION SYNDROME

*Clinical Atlas of Human Chromosomes (A Wiley medical publication) (English and French Edition) [Jean Degrouchy, Catherine Turleau, Jean de Grouchy] on www.enganchecubano.com *FREE* shipping on qualifying offers.*

Reference List Chromosomes are the structures, in the nuclei of our cells, which are composed of helical, double-stranded DNA and associated proteins. The DNA molecules encode our human and individual genetic heritage. Two types of genetic injury which are readily caused by ionizing radiation at very low doses and low dose-rates are chromosomal deletions and translocations. Recent evidence links a great variety of chromosomal deletions and translocations with devastating birth defects and mental handicaps. Nonetheless, pressure to "forgive" more nuclear pollution and thus "forgive" more involuntary exposures to ionizing radiation is reviving in a big way. This CNR paper describes the evidence which links chromosomal deletions and translocations with mental handicap and structural defects of the heart, kidneys, digestive tract, skeleton, and genitalia, and it also describes the limits of technology which have delayed this evidence for so long. The essay is a non-technical introduction to just a small part of the story of chromosomal injuries, for it omits any consideration of health consequences such as cancer, schizophrenia, and metabolic diseases for instance, diabetes, hyper-lipidemia, cystic fibrosis. My next book, in Chernobyl Accident: Radiation Consequences for This and Future Generations Russian Language , will provide detailed evidence and analysis of the under-estimated health effects which can arise from radiation-induced chromosome damage. The information also has implications far beyond nuclear pollution, to the extent that chemicals and viruses and possibly other types of radiation may induce permanent chromosome injuries too. The fact that ionizing radiation can break chromosomes has been "answered" between and the present day by questioning the health effects see the three boxes [1] , [2] , [3] in this essay. With respect to this and many other pollutants, the "prove harm" proponents see nothing wrong about establishing "permissible" levels of involuntary exposure, despite an ocean of ignorance regarding the potential, miserable consequences some of which are identified in this essay. Chromosome damage confined to a segment of DNA representing a single gene. Breakage of a chromosome, followed by permanent loss of part of a chromosome carrying some or many entire genes, or just part of one gene. Breakage of one or more chromosomes, followed by permanent removal of some or many genes and partial genes from their normal place in the DNA chain; these relocated DNA segments can end up in an abnormal place within the same DNA chain or within the DNA of an entirely different chromosome. All three types of permanent chromosomal injury are now called "genetic mutations," and types B and C are also called "structural chromosome aberrations. Anytime during childhood and adulthood. When genetic mutations occur before conception inherited or during early gestation not inherited , the health consequences can be virtually identical. Distinctions are poorly defined between "genetic diseases," "irregularly inherited disorders," "constitutional diseases," "chromosomal disorders," "congenital diseases," and "birth defects" or "anomalies. Indeed, a large share of all bio-medical research in recent years has been devoted to the genetic basis of disease and health, and existing results await coherent assembly and analysis. Part of the explosion is generated by the Human Genome Project, in which the U. Department of Energy is extremely active. Although the existence of chromosomes has been known for over a century, very little progress was made for a long time. Chromosomes are not visible, unless you "catch" a cell which is preparing to divide. Then the very long, string-like chromosomes "condense" by folding themselves into enormously shorter and thicker objects. Ordinary stains used in biology showed their existence, but the objects appeared entangled with each other, and no one was even able to establish the correct number of human chromosomes per cell-nucleus during the pre-cytogenetic era. In , Hsu developed a simple but enormously powerful technical advance in chromosome studies. When cells are bathed in a solution with salt-concentration lower than their own salt-concentration, the cells swell. The chromosomes in cells preparing to divide become so well separated that quite a few details of individual, separated chromosomes can be noted, when the division is halted by a chemical inhibitor and the cells are flattened on a glass slide. The year was also the year in which Watson and Crick announced the structure of the gene and DNA helix. The required technologies for that kind of very sophisticated analysis had become available before the availability of techniques which would permit us merely to count the number of structures which carry the genes. The father contributes 23 chromosomes and the mother also contributes 23 chromosomes to the fertilized ovum, from which the 46 are replicated in the cell-nuclei of all the descendant cells when everything goes well. There are 22 matched pairs called autosomes, grouped by letters A-G for instance, paternal and maternal B-5 chromosomes. Each chromosome has a region somewhere along its length called the centromere, which divides the chromosome into a shorter arm called the p-arm and a longer arm q-arm. Each pair of undamaged autosomes provides the cell with two copies of each gene on the autosome a full set of this genetic information from the father and a full set from the mother. In , we were yet to learn that there can be severe consequences for the children who have either more than two complete copies or fewer than two complete copies of the genetic information on both arms of each chromosome. But in , our ignorance on this matter began to retreat. Here we will summarize some insights which were gained in the pre-banding era. In Part 2 , we described two types of structural chromosome aberrations deletions and translocations. There are also numerical chromosome aberrations. When an extra copy of one complete chromosome is present in cells, so that the cells contain 47 instead of 46 countable or "free" chromosomes, the condition is called a trisomy. When one complete copy of a chromosome is missing, so that cells have 45 instead of 46 countable chromosomes, the condition is called a monosomy. Of course, neither condition could be verified until the normal number of chromosomes was discovered in  We can call this an all-cell, full-chromosome trisomy The rest of the cases arise from two other types of trisomy Although we will explain them here, with the pre-banding era, these other types were not discovered as early as all-cell, full-chromosome trisomy. Mosaicism When an infant has 47 chromosomes in every cell, it means that the numerical aberration was present in the fertilized ovum. Thus, the aberration occurred during gestation, in-utero. Individuals with two types of cells some with 46 chromosomes, some with 47 are mosaics, and they have a some-cell, full-chromosome trisomy. We mean the translocation see Part 2. Although a thorough explanation would require more space than we have here, Figures 1 and 2 may convey a sense of the problem. Figure 1 depicts a normal E chromosome and a normal B-5 chromosome. Figure 2 depicts their possible status after a translocation. When a chromosome carries a mixture of information belonging to more than one chromosome, its name is set by the information around its centromere depicted by the black area. In Figure 2, the chromosome on the left is called the E, and B-5 is on the right. The condition of partial trisomy arises for a child as follows. Suppose that both the mother and father transmit normal E- 18 chromosomes to their child. But suppose that in the transmission of B-5 chromosomes, one parent transmits the damaged B-5 chromosome from Figure 2. It carries translocated genes belonging to the q-arm of the E chromosome. The child, whose numerical count of separate chromosomes is the normal 46, will nonetheless have three copies of part of the genetic information on the q-arm of the E chromosome. The child will have a partial trisomy This structural aberration can be described as an all-cell 18q trisomy. Translocations and Partial Monosomies In the example above, the child simultaneously has a partial monosomy because the child has received a B-5 chromosome which lacks part of its p-arm and lacks the genetic information which was on it. Even though the other parent sends the child a normal B-5 with a complete p-arm, the child will have only one copy instead of the normal two copies of some genetic information belonging to the p-arm. This condition can be described as an all-cell 5p monosomy. Because the chromosome-count will be the normal 46 per cell, this is not a numerical aberration. Emphasis belongs on the fact that the effect of a partial monosomy is no different from an inherited deletion see Part 2. The net effect is an all-cell deficit of chromosomal information. The deficit may or may not be limited to genes which code for specific enzymes. The deficit will often include A some segments of DNA which have presently unknown but presumably important functions, and B some chromosomal proteins whose functions are presumably important too. Since then, the frequency has been estimated between 1 case per 4, and 1 case per 10, live births. The clinical features of trisomy include percentage of cases: And also in , Edwards described the first patient with a full-chromosome trisomy The frequency is now estimated at 1 case per 8, live births. Box 1, A Contrast in Warnings: These were "permissible" doses of 0. Does it make you feel at all uneasy that this spectacular field of human cytogenetics is now in its infancy, after all the decisions had been made which led to the setting of FRC guidelines for radiation exposure of populations? Unofficially, there was talk of increasing the permissible dose. Back then, the AEC had ambitious plans to build "a plutonium economy," to license to large nuclear power plants by the year AEC , p. Later, with the advance of technology, it was discovered that both trisomy and trisomy like trisomy also can occur as mosaics in-utero and as partial trisomies inherited as a result of translocations. The Linking of Cause with Consequence No one is claiming that all individuals who have the health effects listed above are cases of trisomy, , or  Many additional genetic causes of these health problems have been discovered, and it is possible that some cases arise without any genetic injury at all. Then how can anyone be sure that a trisomy causes the problems of trisomic individuals? Whenever a variety of causes might produce the same health effect, two types of study can establish causation. In a prospective cohort study, you start with a suspected cause and then you measure the occurrence of presumed consequences. You measure the health of one group which has trisomic cells a presumed cause of the listed health effects and another group which does not have trisomic cells, and you discover which group has the higher rate of the health effects. In a retrospective case-control study, you start with presumed health consequences and then you measure the occurrence of a suspected cause. You measure the rate of trisomic cells in one group which has the health effects a presumed consequence of trisomy and in another group which does not have these health effects, and you find out which group has the greater frequency of trisomic cells a presumed cause. There is no doubt that the frequency of 47 chromosomes is higher among the persons who have the health effects listed above. Indeed, the rate of full-chromosome trisomy among persons who lack such health effects is so low that we are unaware of a single known case. The First Discoveries of Deletion Syndromes All-cell, full-chromosome trisomies were open to study in the early era, since this was a matter of just counting chromosomes. But the opposite possibility namely, a deficit of certain chromosomal information was not nearly so easily studied, since only arm-lengths and centromere positions were available to identify such losses. Nonetheless, by progress was underway when Lejeune and co-workers described the first three cases of the 5p partial monosomy or deletion syndrome. It was called Cri du Chat Syndrome because infants with it have a peculiar, cat-like mewing cry. The disorder is severe. Besides the cry, clinical features in most cases include profound mental handicap, small head, low-set ears, and growth failure. Box 2, A Contrast in Warnings: In , what sort of warning did it issue concerning radiation-induced chromosome damage? First, it grouped "small deletions" with single-gene mutations, and then stated:

## 2: Radiation-Inducible Chromosome Injuries: Some Recent Evidence on MAJOR Health Consequences

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 Abstract The duplication of chromosome 3q is a rare disorder with varying chromosomal breakpoints and consequently symptoms. Even rarer is the unbalanced outcome from a parental inv 3 resulting in duplicated 3q and a deletion of 3p. We report a case of an infant male with a  This patient also has an accompanying 1. The duplicated segment in this patient encompasses the known critical region of 3q Taken together, our data has refined the location and extent of the chromosome 3 imbalance, which will aid in better understanding the molecular underpinning of the 3q syndrome. Microarray technologies are more commonly becoming the tool of choice to accurately determine the underlying genetic cause and resulting phenotype [ 1 ]. The duplication of chromosome 3q is a rare genetic disorder resulting in mental retardation, seizures, broad nose, cardiac, renal, and genital malformations [ 2 ]. In contrast, deletions of chromosome 3p are associated with intrauterine and postnatal growth retardation with delayed bone maturation, severe psychomotor retardation, dysmorphism including ptosis, a narrow nose, flat nasal bridge, clinodactyly, heart and kidney defects, and impaired vision [ 2 , 4 ]. The size of the deletion appears to correlate with severity of the phenotype such that patients with a large deletion exhibit severe malformations and mental retardation [ 5 ]. Previously reported cases of patients carrying a duplication at 3qter as well as a large deletion at pter-3p25 have a fatal outcome [ 2 ]. We report a case here in which the patient has a much smaller 3p deletion in combination with the 3qter duplication, and discuss whether the 3p deletion size affects patient phenotype and outcome. Case Report A one-month-old male presented with a large ventricular septal defect VSD , large posterior and anterior fontanelle, dysmorphic features, single palmar crease, under-developed testes and mild seizures. The baby was the product of a normal first pregnancy and was delivered at 41 weeks, 6 days with a birth weight of 4. Labour was complicated by foetal distress, and delivery was by caesarean section and admitted to the Newborn Intensive Care Unit NICU on day 2 for respiratory distress. Ultrasound analysis revealed a thin corpus callosum and a consequent MRI of the brain and spine revealed a small right germinal matrix haemorrhage and mild craniofacial disproportion and mild micrognathia. There was also a separate choroidal fissure cyst on the left 9 mm in maximal dimension. The corpus callosum was thin, but present. At 14 months of age, the proband had gained weight despite feeding difficultes and was breast feeding; post surgery, the child exhibited normal biventricular function with no residual VSD and no audible murmurs; he was tachypnoeic with a respiratory rate of 60, but his chest was clear. He had a thickened filum, and surgery was suggested in infancy to prevent later problems with foot development. He also had reduced antigravity movement in his upper and lower limbs with reduced central tone, but increased tone in his limbs. Cytogenetic and Microarray Analysis Metaphase chromosomes were prepared from stimulated peripheral blood cells according to standard methods and karyotyping was performed by G-band metaphase analysis. This analysis showed a large duplication of material on the short arm of chromosome 3, which appeared 3q-like Figure 1. Karyotype and chromosome 3 ideogram of the proband. Panel A shows the karyotype of the proband, 46,XY,rec 3 dup 3q inv 3 p Panel B shows the normal and derivative chromosomes 3, together with an associated ideogram. Panel C is a summary ideogram of the regions of chromosome 3 that are duplicated and deleted in the proband. This analysis confirmed the copy number change as a  Schematic of the deleted chromosome 3 region in the proband. Panel A shows an ideogram of chromosome 3, together with the location of the deletion indicated in red. Panel B shows the genes that are localised within the deleted region, those reported in the OMIM database http: The images presented here are taken from the UCSC genome browser http: Parental analysis confirmed that these

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chromosome 3 changes arose as an unbalanced product of a meiotic recombination in the mother who has a pericentric inversion of one homologue of chromosome 3 between p26 and q23 Figure 3. Karyotype and chromosome 3 ideogram of maternal chromosomes. Panel A shows the karyotype of the mother 46,XX,inv 3 p Panel B shows the normal and structurally rearranged chromosomes 3, together with an associated ideogram. The deletions in these patients range in length from kb to  In the main, the associated clinical phenotypes of these patients do not match those identified in the proband who carries a smaller distal 3p deletion, as well as a duplication of 3q. Discussion The duplicated segment in the proband described here encompasses the known critical region of 3q Previously reported patients with a smaller 3p deletion at 3p Evidence to suggest that genes in the 3p The patient had a high birth weight and broad nose, which is not consistent with the deletion phenotype. Microarray analysis confirmed that this patient has a smaller accompanying 1. The size of the deletion appears to have minimal impact on the phenotype of this patient and this is consistent with previously reported cases. Conclusion Taken together, our data has refined the location and extent of chromosome 3 imbalances. Molecular karyotyping has led to a better understanding of the molecular underpinning and phenotypic outcome in the proband reported here and should be considered in future cases to aid a prognosis. A full list of centres who contributed to the generation of the data is available from http: Van Den Berg et al.

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