Karyogram for the Primary Diagnosis of Human Chromosomal Abnormalities

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During pregnancy, a genetic test or Non-Invasive Prenatal Testing (NIPT) is performed on the unborn child to detect potential chromosomal abnormalities. For the test, the wet lab checks which of the mother's amniotic fluid cells are to be examined with karyotypes. Using a microscope to correct the alignment, the alignment results are then used to diagnose if there is an extra or missing chromosome or the structure is abnormal.The objectives of this project were: 1.) to design code for machine learning to process images of chromosomes to create karyotypes, create schematic diagrams to guide design and write code, and then execute the design and implementation. A code was written in Pycharm, images were extracted, and the code was used for image analysis. A second code was written for error testing and correction, after which specimens were tested for performance. 2.) To study karyotype performance in intelligence models. All models were tested and the best model was Nadam + conv2D + Activation + Maxpooling2D + ImageDataGenerator + augmenting 93.06% test. By processing images of human chromosomes to diagnose genetic anomalies from the images, the alignment was reduced. From 100 test results, the mean accuracy was 93% and all 7 disorders were diagnosed: Patau syndrome, Edward's syndrome, Down's syndrome, Klinefelter's syndrome, Double Y's syndrome, Turner's syndrome, Triple X's syndrome. The validation accuracy was 93.06%. Keywords: karyotype, characterization of machine learning images, diagnosis of human genetic disease disorders.

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