Relating Symptoms with Protein Production Abnormality in Patients with Down Syndrome

Authors : Ruolan Zhou

Abstract : Trisomy of human chromosome 21 is the primary cause of Down Syndrome (DS), and this genetic disease has significantly burdened families and countries, causing great controversy. To address this problem, the research takes an approach in exploring the relationship between genetic abnormality and this disease's symptoms, adopting several techniques, including data analysis and enrichment analysis. It also explores open-source websites, such as NCBI, DAVID, SOURCE, STRING, as well as UCSC, to complement its result. This research has analyzed the variety of genes on human chromosome 21 with simple coding, and by using analysis, it has specified the protein-coding genes, their function, and their location. By using enrichment analysis, this paper has found the abundance of keratin production-related coding-proteins on human chromosome 21. By adopting past researches, this research has attempted to disclose the relationship between trisomy of human chromosome 21 and keratin production abnormality, which might be the reason for common diseases in patients with Down Syndrome. At last, by addressing the advantage and insufficiency of this research, the discussion has provided specific directions for future research.

Keywords : Down Syndrome, protein production, genome, enrichment analysis

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