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## Genetic analysis of 20 facioscapulohumeral muscular dystrophy (FSHD) probands by southurn blot analysis and investigation of genotype/phenotype correlation

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Facioscapulo humeral muscular dystrophy (FSHD) is a dominantly inherited disease that is characterized with involvement and weakness of the facial and scapular muscles. It's one of the most common forms of myopathies with prevalence 1:7500-1:15000. FSHD is caused by genetic and epigenetic factors and can be classified into two subgroups: FSHD1 and FSHD2. FSHD1-accounts for around 95% of the cases- is associated with contraction of D4Z4 macrosatellites at the subtelomeric region of chromosome 4. Normal individuals carry 11-100 repeats whereas FSHD1 patients have 1-10 D4Z4 repeats. 20 Iranian FSHD probands were recruited. All cases consented to participate after being informed about the project. Their clinical presentations were recorded precisely. DNAs were extracted using salting out method and digested by EcoRI and EcoRI/BlnI enzymes. The southern blot was optimized for DIG-labelled probe P13E11. The average age at onset of patients was 15-/+6.5 (range 1-49 years). 12 out of 20 patients were familial and remaining cases were sporadic. Respectively, four (20%), six (30%) and ten (50%) of patents presented mild, severe and moderate type of the disease. Results of Southern blot showed 19 of 20 (95%) patients shared less than nine D4Z4 repeats. One patient presented more than 11 D4Z4 repeats. Our findings showed a correlation between genotype and phenotype. More severe type of the disease war related to less number of the D4Z4 repeats.

### Biography

Afagh Alavi has completed her MSc, PhD and Postdoctoral studies from University of Tehran. She has been working as an Assistant Profesor in the University of Social Welfare and Rehabilitation Sciences. She has published more than 13 papers in reputed journals.

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