



# International Congress on Friedreich's ataxia

11<sup>th</sup> April, 2015

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## International Congress on

“DNA Structure in Health & Disease”

12 - 13 April, 2015

All India Institute of Medical Sciences  
Ansari Nagar, New Delhi-110029 (INDIA)



### Abstract Submission

(The abstract along with desired information should accompany the registration form)

#### **ABSTRACT SUBMISSION GUIDELINES**

(Abstract Submission is extended to 30th March, 2015)

### Instructions:

1. Abstract should be submitted in MS Word format by E-mail attachment.
2. Abstract should NOT EXCEED 300 WORDS.
3. Abstract should possess title (all capital), author's name (family name first), affiliation and text. Underline the presenting Author's name.
4. Abstract will be accepted ONLY after receiving the Registration Fee.
5. Please send the Draft and registration form by Speed Post to the Organizing Secretary.
6. You will be intimated regarding the **confirmation of the Abstract and /Registration by email.**
7. Please follow the format of "sample Abstract", given below.

Please send abstract as E-mail attachment to: [dshd2015@gmail.com](mailto:dshd2015@gmail.com)

<b>Title</b>	:	<b>Arial in Capital 12 Font</b>
<b>Author(s)</b>	:	<b>Arial 10 Font, Underline the presenting Author's name</b>
<b>Address</b>	:	<b>Arial Font 10 with email . email in italics, Font 10</b>
<b>Abstract</b>	:	<b>Arial , Single space , Font 10 .</b>

**Sample Abstract:**

**IDENTIFICATION AND QUANTIFICATION OF DIFFERENTIALLY EXPRESSED PROTEINS IN PLASMA OF SPINOCEREBELLAR ATAXIA TYPE 12**

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Spinocerebellar ataxia 12 (SCA12) is a unique dominant type of ataxia characterized by early and prominent action tremors, memory deficit, neuropathy, dysarthria etc. The expansion of DNA triplet (CAG) repeats in 5'UTR of *PPP2R2B* gene appears to be the cause for the pathogenesis of the neurodegenerative disorder, SCA12. The objective of the current study was to identify the aberrantly expressed plasma proteins for their potential application in therapy or diagnosis/prognosis of SCA12. Sixty-two clinically suspected patients were assessed using International co-operative ataxia rating scale (ICARS) and genetic confirmation was done using PCR followed by DNA sequencing. Twenty patients who were genetically confirmed were included in the study. 2D-DIGE analysis of plasma proteins of SCA12 patients revealed 14 differentially expressed protein spots, which were confirmed as nine proteins by LC-MS/MS. The 6 downregulated and 3 upregulated proteins are known to have physiological role in transport (thyroxin and retinol to brain), lipid metabolism, memory, scavenging of free haemoglobin etc. Altered expression of some of the proteins of interest, transthyretin, haptoglobin, apolipoprotein C-II, apolipoprotein C-III are indicative of clinical manifestations such as neuropathy, cognitive impairment and altered lipid metabolism in SCA12.