RESUME

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Demographic Profile:

Date of Birth	3rd Nov, 1977
Sex	Male
Marital Status	Married
Nationality	Indian

Educational Synopsis:

Sep, 2005- Till date- working on the project entitled "Molecular cloning and characterization of major copper binding protein (50kDa) from Indian Childhood Cirrhosis patients".

Jan, 2001- Sep, 2005- worked on "Effect of copper toxicity in isolated human peripheral blood mononuclear cells and it's attenuation by zinc" and "Ectopic expression of alkaline phosphatase in proximal tubular brush border membrane of human renal cell carcinoma".

Jan, 2002- Sep, 2005- Ph.D in Biochemistry from Department of Biochemistry Postgraduate Institute of Medical Education & Research Chandigarh, 160012. Research Topic: "Identification and Characterization of wide Spectrum of mutations in Wilson's disease gene (ATP7B): Genetic and Diagnostic Implications". Thesis submitted on 12th Sep, 2005.

Jan, 2001- 31 Dec, 2001 Worked as Junior Research Fellow in Department of Biochemistry Postgraduate Institute of Medical Education & Research Chandigarh, 160012, in the project entitled "Characterization of mutations in Wilson's disease gene and their consequences: DNA based presymptomatic diagnosis" sponsored by Department Of Biotechnology, Govt. of India, New Delhi, India.

Dec, 1999- Jan, 2001 worked as Technical Assistant with Genetix Biotech (Asia), Pvt Ltd, New Delhi, India.

July, 1997- Sep, 1999: M.Sc. in Biotechnology from Kurukshetra University Kurukshetra, Haryana, (India) with 71.5% marks in 1999.

1994-1997: B.Sc. in Biology from Chaudhary Charan Singh University, Meerut (India) with 65% marks in 1997.

Merit and Awards:

Awarded Dr. Rhada Krishnan scholarship in M.Sc (Biotechnology) by Kurukshetra University Kurukshetra, Haryana, (India) from 1998-1999.

Qualified the examination conducted by Postgraduate Institute of Medical Education & Research Chandigarh, 160012 for Ph.D (2001), which was held at national level.

Awarded Senior Research Fellowship by Indian Council of Medical Research, Govt. of India, New Delhi, India from Feb-2004 to 2007.

STATEMENT OF RESEARCH SUMMARIES:

I embarked upon my research career because of a great interest in molecular basis of diseases. During my Ph.D. program (Jan, 2002 – Sep, 2005) I focused on the identification and characterization of wide spectrum of mutations in Wilson's disease gene (ATP7B). Initially I characterized 22 mutations (18 novel) and 6 polymorphisms (3 novel). During this time I determined the alleles distribution and haplotype association with specific mutation by using dinucleotide markers in Wilson disease patients and also developed PCR-RFLP based molecular diagnostic tool for diagnosis of asymptomatic Indian WD patients as well as assessment of carrier status of WD families.

Simultaneously I have also worked on antagonist behavior of copper and zinc on human peripheral blood mononuclear cells (PBMCs) and evaluate that zinc play an important role in prevention of copper toxicity in peripheral blood mononuclear cells. In another study I found that expression of alkaline phosphatase (ALP) in tumor renal BBM was markedly reduced as compared to normal kidney. Lipid composition in reference to phospholipids, glycolipids and cholesterol in tumor renal BBM was altered to that of normal renal BBM, indicating an alteration in membrane fluidity of tumor renal BBM. However, the exact role of these potentially and functionally significant molecular abnormalities associated with RCC remains to be identified. An understanding of the reason underlying decreased expression/ activity of ALP in renal tumor BBM may provide insights into the process of carcinogenesis and normal function of the enzyme alongwith a new tumor marker. I have also elucidated destabilization of DNA associated with alterations in configuration and subsequently massive DNA fragmentation in response to copper and H_2O_2.

PAST ACCOMPLISHMENTS:

All my research works are published in reputed International journals. My doctoral research gave me an exquisite exposure to genetic, biochemical, molecular biology techniques. During my Ph.D. program, I participated in Journal clubs and

Seminars held at the department of Biochemistry, at Postgraduate Institute of Medical Education and Research (PGIMER), Chandigarh, India. I have presented many papers on RNAi, Riboswitchs, ribozymes, RNA repair, RNA editing, Chimeraplasty etc. I have also trained the students undertaking Masters Degree course in Biochemistry, at the department of Biochemistry, PGIMER and in Biotechnology from other institutes/Universities. The postgraduate students include, Mr. Rashim Pal Singh (co-author on one of the articles in the journal Mol Cell Biochem, (**Mol Cell Biochem 282(1-2):13-21, 2006.**), Ms. Sheetal Kumar (co-author on one of the articles in the journal **Indian J Exp Biol, 44, 377-380, 2006**) and Ms. Simranjit Kaur. The Indian Council of Medical Research, Govt. of India, New Delhi, India awarded me Senior Research Fellowship from Feb-2004 to Feb-2007.

Meetings Attended:

Vivek Chander Shekher memorial, 2001 (VCSMYS 01), Genomics organized by Association of Basic Medical scientists, PGIMER, Chandigarh, 5th April, 2001.

Scientific symposium "proteomics" organized by Association of Basic Medical scientists, PGIMER, Chandigarh, Oct 5th, 2002.

Attended: CME on "Recent advances in Genetic disorders related to various disabilities in Indian population" organized by Department of psychiatry, Government medical college and hospital, Chandigarh.

Expertise in:

Molecular Biology:

* DNA isolation from whole blood and tissues.
* Plasmid DNA isolation.
* RNA isolation from whole blood and tissues.
* Electrophoresis: Agarose gel Electrophoresis, Polyacrylamide gel Electrophoresis: Non-denaturating, denaturating and SDS-PAGE
* EtBr and silver staining for DNA
* Silver and Coomassie staining for proteins
* Polymerase Chain Reaction: DNA amplification, RT-PCR, Seminested PCR, Real time PCR, ARMS-PCR and Multiplex PCR.
* Single strand conformation polymorphism.
* Dcode mutation analysis system
* Rapid translation system
* Protein truncation test
* Dot-Blot hybridization
* Heteroduplex analysis
* Haplotype analysis
* Restriction fragment length polymorphism.
* Random amplification of polymorphic DNA.
* Southern and Western blotting

* Gel purification of DNA
* DNA Sequencing
* Probe labeling (end labeling, random labeling)
* Transformation
* Transfection
* Competent cells preparation
* Cloning
  Topo TA cloning
  Homopolylinker Tailing

Biochemistry:

* Isolation and purification of brush border membrane
* Protein purification
* Protein estimation by Lowry's method, Bradford method
* Copper ATPase assay
* Lipid peroxidation in Plasma
* SOD and catalase
* Glutathione Peroxidase, Total and reduce glutathione
* Metallothionein estimation by silver saturation method.
* Serum ceruloplasmin estimation
* Serum copper, urinary copper and hepatic copper estimation on Atomic Absorption Spectrophotometer
* Fluorescence spectrophotoetry

Cell Culture Techniques

* Cell line maintenance (LLCPK-1A and Hep-G2)
* Peripheral mononuclear cells isolation from blood
* Isolation and culture of renal cells
* Trypan Blue assay
* Cell counting
* Microbial culture

Bio-Informatics:

* Analysis and annotation of DNA sequences using annotation software such as BLAST and Gene Runner version 3.2 software.
* Arlequin, HAPLO program for haplotying analysis
* Lab Image, Kapelan for Gel analysis and gel documentation system.
* Statistical analysis by using SPSS (Version 13.0) and Excel.
* Usage of all PC base programs like MS word, Excel, Adobe Photo shop, ACD photo enhancer, Corel draw, Power point etc.

Publications:
Articles published-

1. **Kumar S,** Thapa BR, Kaur G, Prasad R. Identification and molecular characterization of 18 novel mutations in the *ATP7B* gene from Indian Wilson disease patients: genotype. **Clin Genet** 67(5): 443-445, 2005.
2. **Kumar S,** Thapa BR, Kaur G, Prasad R. Familial gene analysis for Wilson disease from North-West Indian patients. **Annals of human Biology (In Press).**
3. **Kumar S,** Thapa BR, Kaur G, Prasad R. Analysis of most common mutations R778G, R778L, I1102T and H1069Q in Indian Wilson disease patients: correlation between genotype/phenotype/copper ATPase activity. **Mol Cell Biochem (In press).**
4. Prasad R, **Kumar S,** Kaur G, Thapa BR. Two novel mutations (2976insA, 4311insA) of *ATP7B* in a Wilson Disease patient coexisting with Glucose-6-Phosphate Dehydrogenase deficiency. **J Gastroenterol Hepatol** 20(4): 661-663, 2005.
5. Prasad R, Lambe S, Kaler P, Pathania S, **Kumar S,** Attri S, Singh SK. Ectopic expression of alkaline phosphatase in proximal tubular brush border membrane of human renal cell carcinoma. **Biochim Biophys Acta (Molecular Basis of Diseases**) 1471(3):240-245, 2005.
6. Singh RP, **Kumar S**, Nada R, Prasad R. Evaluation of copper toxicity in isolated human peripheral blood mononuclear cells and it's attenuation by zinc: ex-vivo. **Mol Cell Biochem 282(1-2):13-21, 2006.**
7. Prasad R, Kumar RS, **Kumar S.** Hydrogen peroxide commences copper induced DNA damage isolated from human blood: in vitro study. **Indian J Exp Biol 44, 377-380, 2006.**

Articles Communicated:

1. **Kumar S,** Thapa BR, Kaur G, Prasad R. Haplotype analysis in Indian Wilson disease patients. (**European J of Human Genetics, Jan, 2006)**

Abstracts:

1. Kaur G, **Kumar S**, Thapa BR, Prasad R. . Haplotype analysis in Indian Wilson disease patients. **Am J Hum Genetics, 2005. (Accepted)**
2. Kaur G, Thapa BR, Prasad R, **Kumar S**. Mutational analysis of ATP7B and genotype-phenotype correlation in Indian Wilson disease patients**. Am J Hum Genetics** PSA: 75(4) 1758, 2004.

3. Prasad R, Kaur G, Thapa BR, **Kumar S**. Analysis of most common mutations H1069Q, R778L and R778G in Wilson disease patients from North West India. **Am J Hum Genetics** PSA: 75(4), 2577, 2004.

4. Prasad R., **Kumar S**, Kaur G, Thapa BR, Jahagidar S, Verma S. Antioxidant in Wilson disease: Role of copper. 29th Annual Conference of Human Genetics (ISHG 2004), Bangalore. India. Pp –29.

5. Prasad R, Kaur G, Thapa BR, **Kumar S**,. Wide spectrum of mutations in CuATP7B gene and their consequences in north-west Indian Wilson disease patients.29th Annual Conference of Human Genetics (ISHG 2004), Bangalore. India. Pp –40.

6. Prasad R, Kaur G, **Kumar S**, Thapa BR,. Assessment of antioxidant status in Wilson's disease children: Effect of vitamin E and Zn supplementation. **Am J Hum Genetics** PSA: 71 (4), 415, 2002.

Referees:

1. **Prof. Rajendra Prasad**
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I hereby solemnly declare that all the statements made in the above curriculum vitae are true and correct to the best of my knowledge and belief.

Place: Chandigarh, India

SANDEEP KUMAR