Deepak Babu

Date of Birth: [redacted]

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CURRENT POSITION

March 2015 – Present
Post-Doctoral Researcher
Department of Health sciences, Laboratory of Human Genetics
University of Eastern Piedmont, Novara, Italy

I am part of a research group interested in the molecular elucidation of human diseases with a focus on growth and kidney disorders. I am involved in the study of how mutations correlate with diseases and the identification of new candidate genes that could be disease causative by integrating genetics, molecular and cell biological approaches.

EDUCATION

2012 – 2015
Ph.D in Molecular Medicine, University of Eastern Piedmont, Italy
Thesis Title: *Genetic analysis of GH1, GLI2 and SHOX genes in patients with growth impairments.*

Advisor: Prof. Mara Giordano

2008 - 2009
MSc Biotechnology, University of Abertay Dundee, Scotland, UK
Thesis Title: *Cloning of full length human ATM with the aim of tagging it with Fluorescent protein to study the expression.*

Advisor: Prof. Nikolai Zhelev

2004 – 2008
B.Tech Biotechnology, Anna University, Chennai, India

WORK EXPERIENCE

March 2010 – July 2010
Research Assistant, St.George's University of London, England
Department of Cellular and Molecular Medicine,
Laboratory of Dr.Jonathan Kerr

November 2009 – March 2010
Research Assistant (Voluntary), University of Abertay Dundee
Laboratory of Dr.Yusuf Deeni

PUBLICATIONS

Babu D, Mellone S, Fusco I, Petri A, Walker GE, Bellone S, Prodam F, Momigliano-Richiardi P, Bona G, Giordano M. (2014). Novel mutations in the growth hormone gene (GH1) uncover putative splicing regulatory elements. Endocrinology; 155:1786-1792

Prodam F, Savastio S, Genoni G, **Babu D**, Giordano M, Ricotti R, Aimaretti G, Bona G, Bellone S. (2014). Effects of growth hormone (GH) therapy withdrawal on glucose metabolism in not confirmed GH deficient adolescents at final height. PLoS One 9:e87157.

Fusco, I., **Babu, D.**, Mellone, S., Barizzone, N., Prodam, F., Fanelli, A., Muniswamy, R., Petri, A., Bellone, S., Bona, G., *et al.* (2015). Variations in the high-mobility group-A2 gene (HMGA2) are associated with idiopathic short stature. Pediatr Res; 79(2):258-61

De Rienzo, F., Mellone, S., Bellone, S., **Babu, D.**, Fusco, I., Prodam, F., Petri, A., Muniswamy, R., De Luca, F., Salerno, M., *et al.* (2015). Frequency of genetic defects in combined pituitary hormone deficiency: a systematic review and analysis of a multicentre Italian cohort. Clin Endocrinol (Oxf); 83(6):849-60.

Musetti C, **Babu D**, Fusco I, Mellone S, Zonta A, Quaglia M, Cantaluppi V, Stratta P, Giordano M. (2016). Testing for the cytosine insertion in the VNTR of the MUC1 gene in a cohort of Italian patients with autosomal dominant tubulointerstitial kidney disease. J Nephrol: 29(3):451-5

Giordano M, Muratore V, **Babu D**, Meazza C and Bozzola M. (2016). A 18p11.23-p11.31 microduplication in a boy with psychomotor delay, cerebellar vermis hypoplasia, chorioretinal coloboma,deafness and GH deficiency. Mol Cytogenet. 2016 Dec 3;9:89.

PRESENTATIONS

The frequency of *SHOX* mutations in patients with mild short stature is comparable to the frequency detected in severe Idiopathic Short Stature. Poster session presented at European Human Genetic Congress; 2014 May 31 – June 3; Milan, Italy.

Is autosomal dominant tubule-interstitial kidney disease caused by MUC1 mutation associated with epithelial tumors? Report on two Italian families. Poster session presented at Italian Human Genetic Congress; 2015 21 – 24 October; Rimini, Italy.

Expanding the spectrum of *SHOX* mutations in Idiopathic Short Stature patients through a custom array CGH. Poster session presented at European Human Genetic Congress; 2016 May 21 – May 24; Barcelona, Spain.

Functional analysis of *SHOX* promoter variants, identified in patients with idiopathic short stature (ISS). Poster session presented at Italian Human Genetic Congress; 2016 23 – 26 October; Torino, Italy.

OTHER UNIVERSITY ACTIVITIES

- Tutoring and assisting students with disabilities (Oct 2013 - Sep 2014.
- Assisting international students: providing welcoming and supporting atmosphere to the new international students (Oct 2014 – Sep 2015).
- Conducted molecular biology laboratory practical sessions for undergraduate students.

KEY TECHNICAL SKILLS

Molecular Biology: DNA/RNA extraction and purification, PCR, reverse transcription PCR, real time PCR, site-directed mutagenesis, vector design, transformation, plasmid preparation, electrophoresis. Sanger sequencing, SNaPshot minisequencing, Array Comparative Genomic Hybridisation (Agilent), Multiplex ligation-dependent probe amplification assay (MLPA, MRC Holland), Western blot

Cell biology: Mammalian cell culture, Transfection, Luciferase assay, *in-vitro* splicing assay,

I authorise the use of my personal data in compliance with Italian Legislative Decree 196/03 and subsequent amendments.

NOVARA
17/05/2017