

Deepak Babu

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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 GHI, 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.**

