PUBLICATION PROFILE

Academic visibility and professional authority

Clinical-translational human genetics | rare diseases | genomic diagnostics

This publication profile documents substantial scientific visibility in clinical and translational human genetics, with a publication record focused on rare genetic disorders, growth disorders, skeletal developmental disorders, and genomic diagnostics.

85	34	4.569	4.228
Original publications	h-index	Total citations	Citing articles
11	**16**	**58**	
First-author papers	Senior-author papers	Collaborative papers	

Selected highly cited work

Year	Journal	Paper / focus	Cites
2012	*The Lancet*	Exome sequencing in sporadic non-syndromic intellectual disability	809
2008	*Science*	PCNT mutations in primordial dwarfism	322
2006	*Am J Med Genet A*	Diagnostic yield in unexplained developmental delay / intellectual disability	293
2005	*Nature Genetics*	UBR1 deficiency and Johanson-Blizzard syndrome	202
2015	*Nature Cell Biology*	Functional genomics of ciliogenesis and ciliopathy genes	194

Thematic focus areas

- Growth disorders and short stature
- Clinical and functional variant interpretation
- Ciliopathies and skeletal developmental disorders
- NGS diagnostics and translational implementation

Relevance for expert witness work

This scientific visibility underlines robust expertise in assessing rare molecular findings, classifying complex phenotypes, and critically appraising diagnostic methods and the literature.