

Title: Primary Autosomal Recessive Microcephalies and Seckel Syndrome Spectrum Disorders *GeneReview* Tables 19 and 20

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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

Table 19. Published Pathogenic *N/N* Allelic Variants

DNA Nucleotide Change	Protein Amino Acid Change
(not available)	p.Gln1222Arg [Dauber et al 2012]
(not available)	p.Asn1709Ser [Dauber et al 2012]

See [Quick Reference](#) for an explanation of nomenclature. *GeneReviews* follows the standard naming conventions of the Human Genome Variation Society (www.hgvs.org).

Table 20. Published Pathogenic *N/N* Allelic Variants

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(not available) [Dauber et al 2012]	p.Gln1222Arg

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References

Dauber A, Lafranchi SH, Maliga Z, Lui JC, Moon JE, McDeed C, Henke K, Zonana, J, Kingman GA, Pers TH, Baron J, Rosenfeld RG, Hirschhorn JN, Harris MP, Hwa V. Novel microcephalic primordial dwarfism disorder associated with variants in the centrosomal protein ninein. *J Clin Endocrinol Metab.* 2012;97:E2140-51.