

Gene Details – A2ML1

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| ID | 1 |
| Gene | A2ML1 |
| Summary | <p>This gene encodes a member of the alpha-macroglobulin superfamily. The encoded protein is thought to be an N-glycosylated monomeric protein that acts as an inhibitor of several proteases. It has been shown to form covalent interactions with proteases, and has been reported as the p170 antigen recognized by autoantibodies in the autoimmune disease paraneoplastic pemphigus (PNP; PMID:20805888). Mutations in these gene have also been associated with some cases of Noonan syndrome (NS; PMID:24939586) as well as some cases of otitis media (PMID:26121085). Alternative splicing results in multiple transcript variants encoding different isoforms. [provided by RefSeq, Aug 2015]</p> |
| Chromosome | 12 |
| Gene Type | protein_coding |
| Gene Start (bp) | 10191 |
| Gene End (bp) | 24554 |
| GC Content (%) | 53.68 |
| Transcript Count | 4 |
| Ensembl ID | ENSG00000166535 |
| UniProt ID | A8K2U0 |