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Mutations in mental retardation

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The genetic and molecular basis of mental retardation, which may affect up to 3% of the human population, is unclear. In the June 28 Science, Vervoort *et al.* report that mutations in the *AGTR2* gene, encoding the angiotensin II receptor may affect cognitive function in people with X-linked mental retardation (XLMR; *Science* 2002, **296:**2401-2403). Linkage analysis of XLMR families suggested that the Xq23-25 region might contain more than one gene related to the disease. Vervoort *et al.* studied translocation breakpoints in a female patient with mild retardation (with an IQ of 44); they examined expression of several candidate genes in the breakpoint region and found that the *AGTR2* gene was silenced. They then screened affected males in several XLMR families and found one with a mutation in the *AGTR2* gene that results in a truncated protein. Additional retardation patients with were also found *AGTR2* mutations in the extracellular and intracellular domains of the receptor. The functional role of the angiotensin-renin system in brain function thus merits further investigation.

References

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2. *Science*, [http://www.sciencemag.org]