Agammaglobulinemia-microcephaly-craniosynostosis-severe dermatitis syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Agammaglobulinemia-microcephaly-craniosynostosis-severe dermatitis syndrome. ORPHA:83617

Agammaglobulinemia-microcephaly-craniosynostosis-severe dermatitis syndrome combines agammaglobulinemia with marked microcephaly, significant developmental delay, craniosynostosis, a severe dermatitis, cleft palate, narrowing of the choanae, and blepharophimosis. It has been described in three siblings, two males and one female, born to nonconsanguineous parents. Transmission is probably autosomal recessive. It has been suggested that this syndrome represents a new form of agammaglobulinemia due to a defect in early B-cell maturation.