

# Resources



1. PMID: 38245004

[Ciliopathy due to POC1A deficiency: clinical and metabolic features, and cellular modeling - PubMed \(nih.gov\)](#)

Perge K, Capel E, Villanueva C, Gautheron J, Diallo S, Auclair M, Rondeau S, Morichon R, Brioude F, Jéru I, Rossi M, Nicolino M, Vigouroux C. *Eur J Endocrinol.* 2024 Feb 1;190(2):151-164. doi: 10.1093/ejendo/lvae009. PMID: 38245004

2. PMID: 37873427

[Mesenchymal-specific Alms1 knockout in mice recapitulates key metabolic features of Alström Syndrome - PubMed \(nih.gov\)](#)

McKay EJ, Luijten I, Weng X, Martinez de Morentin PB, De Frutos González E, Gao Z, Kolonin MG, Heisler LK, Semple RK. *bioRxiv* [Preprint]. 2023 Oct 13:2023.10.12.562074. doi: 10.1101/2023.10.12.562074. PMID: 37873427 **Free PMC article.** Preprint.

3. PMID: 35234134

[Biallelic POC1A variants cause syndromic severe insulin resistance with muscle cramps - PubMed \(nih.gov\)](#)

Mericq V, Huang-Doran I, Al-Naqeb D, Basaure J, Castiglioni C, de Bruin C, Hendriks Y, Bertini E, Alkuraya FS, Losekoot M, Al-Rubeaan K, Semple RK, Wit JM. *Eur J Endocrinol.* 2022 Mar 23;186(5):543-552. doi: 10.1530/EJE-21-0609. PMID: 35234134 **Free PMC article.**

4. PMID: 33566311

[Alström syndrome: an ultra-rare monogenic disorder as a model for insulin resistance, type 2 diabetes mellitus and obesity - PubMed \(nih.gov\)](#)

Dassie F, Favaretto F, Bettini S, Parolin M, Valenti M, Reschke F, Danne T, Vettor R, Milan G, Maffei P. *Endocrine.* 2021 Mar;71(3):618-625. doi: 10.1007/s12020-021-02643-y. Epub 2021 Feb 10. PMID: 33566311 Review.

5. PMID: 32994277

[Relative Adipose Tissue Failure in Alström Syndrome Drives Obesity-Induced Insulin Resistance - PubMed \(nih.gov\)](#)

Geberhiwot T, Baig S, Obringer C, Girard D, Dawson C, Manolopoulos K, Messaddeq N, Bel Lassen P, Clement K, Tomlinson JW, Steeds RP, Dollfus H, Petrovsky N, Marion V. *Diabetes.* 2021 Feb;70(2):364-376. doi: 10.2337/db20-0647. Epub 2020 Sep 29. PMID: 32994277 **Free PMC**

6. PMID: 26336158

[Truncation of POC1A associated with short stature and extreme insulin resistance - PubMed \(nih.gov\)](#)

Chen JH, Segni M, Payne F, Huang-Doran I, Sleigh A, Adams C; UK10K Consortium; Savage DB, O'Rahilly S, Semple RK, Barroso I. *J Mol Endocrinol.* 2015 Oct;55(2):147-58. doi: 10.1530/JME-15-0090. PMID: 26336158 **Free PMC article.**

## Resources



7. PMID: 21270239

[Genetic defects in human pericentrin are associated with severe insulin resistance and diabetes - PubMed \(nih.gov\)](#)

Huang-Doran I, Bicknell LS, Finucane FM, Rocha N, Porter KM, Tung YC, Szekeres F, Krook A, Nolan JJ, O'Driscoll M, Bober M, O'Rahilly S, Jackson AP, Semple RK; Majewski Osteodysplastic Primordial Dwarfism Study Group. *Diabetes*. 2011 Mar;60(3):925-35. doi: 10.2337/db10-1334. Epub 2011 Jan 26. PMID: 21270239 **Free PMC article.**