

## **FACE publication list**

### **RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome**

J Clin Invest. 2015 Sep 1;125(9):3585-99. Epub 2015 Aug 17

Bögershausen N, Tsai IC, Pohl E, Kiper PÖ, Beleggia F, Percin EF, Keupp K, Matchan A, Milz E, Alanay Y, Kayserili H, Liu Y, Banka S, Kranz A, Zenker M, Wieczorek D, Elcioglu N, Prontera P, Lyonnet S, Meitinger T, Stewart AF, Donnai D, Strom TM, Boduroglu K, Yigit G, Li Y, Katsanis N, Wollnik B.

#### **DETAILS**

### **Acrofacial Dysostosis, Cincinnati Type, a Mandibulofacial Dysostosis Syndrome with Limb Anomalies, Is Caused by POLR1A Dysfunction**

Am J Hum Genet. 2015 May 7;96(5):765-74. doi: 10.1016/j.ajhg.2015.03.011. Epub 2015 Apr 23.

Weaver KN1, Watt KE2, Hufnagel RB3, Navajas Acedo J4, Linscott LL5, Sund KL3, Bender PL3, König R6, Lourenco CM7, Hehr U8, Hopkin RJ3, Lohmann DR9, Trainor PA2, Wieczorek D9, Saal HM3.

#### **DETAILS**

### **CRIM1 haploinsufficiency causes defects in eye development in human and mouse**

Hum Mol Genet. 2015 Jan 5. [Epub ahead of print]

Beleggia F, Li Y, Fan J, Elcioglu NH, Toker E, Wieland T, Maumenee IH, Akarsu NA, Meitinger T, Strom TM, Lang R, Wollnik B.

#### **DETAILS**

### **Birth prevalence and initial treatment of Robin sequence in Germany: a prospective epidemiologic study**

Orphanet Journal of Rare Diseases 2014, 9:9

Vatlach S, Maas C, Poets CF

#### **DETAILS**

### **Oto-facial syndrome and esophageal atresia, intellectual disability and zygomatic anomalies – expanding the phenotypes associated with EFTUD2 mutations.**

Orphanet J Rare Dis. 2013 Jul 24;8(1):110.

Voigt C, Mégarbané A, Neveling K, Czeschik JC, Albrecht B, Callewaert B, von Deimling F, Hehr A, Falkenberg Smeland M, König R, Kuechler A, Marcelis C, Puiu M, Reardon W, Riise Stensland HM, Schweiger B, Steehouwer M, Teller C, Martin M, Rahmann S, Hehr U, Brunner HG, Lüdecke HJ, Wieczorek D.

### **Human facial dysostoses.**

Clin Genet. 2013 Jun;83(6):499-510. doi: 10.1111/cge.12123. Epub 2013 Apr 8.

Wieczorek D.

### **Mutations in WNT1 cause different forms of bone fragility.**

Am J Hum Genet. 2013 Apr 4;92(4):565-74. doi: 10.1016/j.ajhg.2013.02.010. Epub 2013 Mar 14.

Keupp K, Beleggia F, Kayserili H, Barnes AM, Steiner M, Semler O, Fischer B, Yigit G, Janda CY, Becker J, Breer S, Altunoglu U, Grünhagen J, Krawitz P, Hecht J, Schinke T, Makareeva E, Lausch E, Cankaya T, Caparrós-Martín JA, Lapunzina P, Temtamy S, Aglan M, Zabel B, Eysel P, Koerber F, Leikin S, Garcia KC, Netzer C, Schönau E, Ruiz-Perez VL, Mundlos S, Amling M, Kornak U, Marini J, Wollnik B.

### **Unmasking Kabuki syndrome.**

Clin Genet. 2013 Mar;83(3):201-11. doi: 10.1111/cge.12051. Epub 2012 Nov 26.

Bögershausen N, Wollnik B.

### **Treatment of Obstructive Sleep Apnea in Infants With Trisomy 21 Using Oral Appliances.**

Cleft Palate Craniofac J. 2012 Oct 23. [Epub ahead of print]

Linz A, Urschitz MS, Bacher M, Brockmann PE, Buchenau W, Poets CF.