## **Backhouse et al. Supplementary Material**

## **Supplementary Table 1. Diagnostic and candidate genes for MRKH**

|  |  |  |
| --- | --- | --- |
| **Gene** | **Diagnostic or Candidate?** | **References** |
| ***AMH*** | Candidate | (Mishina et al., 1996) |
| ***AMHR2*** | Candidate | (Mishina et al., 1996) |
| ***BMP2*** | Candidate | (Atsuta et al., 2016) |
| ***BMP4*** | Candidate | (Atsuta et al., 2016) |
| ***CTNNB1*** | Candidate | (Arango et al., 2005; Deutscher and Hung-Chang Yao, 2007; Jeong et al., 2009) |
| ***DACH1*** | Candidate | (Davis et al., 2008) |
| ***DACH2*** | Candidate | (Davis et al., 2008) |
| ***DERL2*** | Candidate | (Ledig et al., 2017) |
| ***DOCK4*** | Candidate | (Ledig et al., 2017) |
| ***EMX2*** | Candidate | (Liu et al., 2015; Miyamoto et al., 1997) |
| ***ESR1*** | Candidate | (Brucker et al., 2017) |
| ***FGFR2*** | Candidate | (Atsuta et al., 2016) |
| ***HNF1B*** | Diagnostic | (Bingham et al., 2002; Lindner et al., 1999; Lokmane et al., 2010) |
| ***HOXA10*** | Candidate (Diagnostic for other Müllerian anomalies i.e. Uterus Didelphys) | (Benson et al., 1996; Cheng et al., 2011; Ekici et al., 2013) |
| ***HOXA11*** | Candidate | (Gendron et al., 1997) |
| ***HOXA13*** | Candidate | (Warot et al., 1997) |
| ***HNRNPCL1*** | Candidate | (Chen et al., 2015) |
| ***ITIH5*** | Candidate | (Morcel et al., 2012; Morcel et al., 2011) |
| ***LAMC1*** | Candidate | (Willem et al., 2002) |
| ***LHX1*** | Diagnostic | (Kobayashi et al., 2004; Ledig et al., 2012; Sandbacka et al., 2013) |
| ***LRP10*** | Candidate | (Rall et al., 2015) |
| ***MMP14*** | Candidate | (Rall et al., 2015) |
| ***MSL3*** | Candidate | (Ledig et al., 2017) |
| ***NAALADL2*** | Candidate | (Ledig et al., 2017) |
| ***OR2T2*** | Candidate | (Chen et al., 2015; McGowan et al., 2015) |
| ***OR4M2*** | Candidate | (Chen et al., 2015) |
| ***OXTR*** | Candidate | (Brucker et al., 2017) |
| ***PAX2*** | Candidate | (Atsuta et al., 2016; Torres et al., 1995) |
| ***PAX8*** | Candidate | (Heidarpour and Tavanafar, 2014; Ma et al., 2014) |
| ***PBX1*** | Candidate | (Ma et al., 2015; Schnabel et al., 2003) |
| ***PDE11A*** | Candidate | (Chen et al., 2015) |
| ***PLCH1*** | Candidate | (Ledig et al., 2017) |
| ***RARA*** | Candidate | (Mendelsohn et al., 1994) |
| ***RARB*** | Candidate | (Mendelsohn et al., 1994) |
| ***RARC*** | Candidate | (Mendelsohn et al., 1994) |
| ***RBM8A*** | Candidate | (Tewes et al., 2015) |
| ***RSPO4*** | Candidate | (Ledig et al., 2017) |
| ***SHOX*** | Candidate | (Gervasini et al., 2010) |
| ***SNAP29*** | Candidate | (Ledig et al., 2017) |
| ***TBX6*** | Candidate | (Sandbacka et al., 2013) |
| ***VPS13A*** | Candidate | (Ledig et al., 2017) |
| ***WNT4*** | Diagnostic | (Biason-Lauber et al., 2004; Philibert et al., 2011) |
| ***WNT5A*** | Candidate | (Mericskay et al., 2004) |
| ***WNT7A*** | Candidate | (Miller and Sassoon, 1998) |
| ***WNT9B*** | Candidate | (Ma et al., 2015) |
| ***WT1*** | Candidate | (Nachtigal et al., 1998) |
| ***ZNF277*** | Candidate | (Ledig et al., 2017) |
| ***ZNF816*** | Candidate | (Chen et al., 2015) |

## **Supplementary Table 2. Diagnostic genes for syndromes with Müllerian involvement**

|  |  |  |  |
| --- | --- | --- | --- |
| **Syndrome** | **Common Syndromic Phenotype** | **Genes** | **References** |
| Acromesomelic Dysplasia (Demihran Type) | Hypoplastic long bones (radius and fibula), brachydactyly, and uterine hypoplasia | ***BMPR1B*** | (Demirhan et al., 2005) |
| Antley-Bixer Syndrome | Craniosynostosis and radiohumral synostosis, with various genital malformations | ***POR*** | (Fluck et al., 2004) |
| Austosomal Recessive Lethal Fatal Ciliopathy | IUGR, microcephaly, renal aplasia, brain malformations, and uterine hypoplasia | ***KIF14*** | (Filges et al., 2014) |
| Bardet-Biedl Syndrome | Retinal dystrophy, obesity, polydactyly, cognitive impairment, and various urogenital anomalies | ***BBS1, BBS2, ARL6, BBS4, BBS5, MKKS, BBS7, TTC8, PTHB1, BBS10, TRIM32, BBS12, MKS1, CEP290, WDPCP, LZTFL1, BBIP1, IFT27*** | (Stoler et al., 1995) |
| Cornelia de Lange Syndrome | Short stature, developmental delay, limb anomalies, cardiac anomalies, hearing and visual impairment, dysmorphic features, and hypoplastic genitals | ***NIPBL, SMC1L1, SMC3, RAD21, HDAC8*** | (Krantz et al., 2004) |
| Currarino Triad | Anorectal abnormalities, partial sacral agenesis, and pre-sacral mass | ***HLXB9*** | (Kochling et al., 2001) |
| Fraser Syndrome | Cryptopthalmos, cutaneous syndactyly, laryngeal and urogenital malformations | ***FRAS1, FREM2, GRIP1*** | (Fraser, 1962) |
| Johanson-Blizzard Syndrome | Exocrine pancreatic insufficiency, dental anomalies, hypothyroidism, sensorineural hearing loss, urogenital and anorectal anomalies, short stature, cognitive impairment, and dysmorphic features | ***UBR1*** | (Zenker et al., 2005) |
| Limb-Mammary Syndrome | Severe hand and foot anomalies, and hypoplasia/aplasia of the mammary glands and nipples. | ***p63*** | (Guazzarotti et al., 2008) |
| McKusick-Kaufman Syndrome | Partial vaginal atresia with imperforate hymen, hydrometrocolpos, hydronephrosis, polydactyly, and cardiac anomalies | ***MKKS*** | (McKusick et al., 1964) |
| Meckel-Gruber Syndrome | Cystic renal dysplasia, occipital encepholocele, and postaxial polydactyly | ***TMEM216, TMEM67, RPGRIP1L, CC2D2A, NPHP3, TCTN2, B9D1, B9D2, TMEM231, KIF14, TMEM107*** | (Balci et al., 1992) |
| Oculo-auriculo-vertebral Dysplasia | Facial asymmetry, microtia, pre-auricular skin tags, ocular defects, conductive hearing loss, vertebral and rib defects | ***TCOF1, SALL1*** | (van Bever et al., 1992) |
| Opitz GBBB syndrome, type II | Hypertelorism or telecanthus; laryngotracheoesophageal cleft; clefts of lip, palate, and uvula; swallowing difficulty and hoarse cry; genitourinary defects, especially hypospadias in males and splayed labia majora in females; mental retardation; developmental delay; and congenital heart defects. | ***SPECC1L*** | (Kruszka et al., 2015) |
| Peters’-plus Syndrome | Eye anomalies, short stature, brachydactyly, developmental delay, and dysmorphic features | ***B3GALTL, PAX6, PITX2*** | (Siala et al., 2013) |
| PDAC Syndrome | Pulmonary hypoplasia, diaphragmatic hernia, anopthalmia, and cardiac defects | ***STRA6*** | (Seller et al., 1996) |
| Roberts Syndrome | Tetramelic, symmetrical limb reduction, radial defects, oligodactyly or syndactyly, microcephaly, cleft lip, and dysmorphic features | ***ESCO2*** | (Van Den Berg and Francke, 1993) |
| Robinow Syndrome | Mesomelic limb shortening, hypoplastic external genitalia, renal and vertebral anomalies, and dysmorphic features | ***DVL1, DVL3*** | (Patton and Afzal, 2002) |
| Setleis Syndrome | Unusual facies, periorbital swelling, wrinkled skin, bulbous nose, and downturned mouth | ***TWIST2, CYP26C1*** | (Girisha et al., 2014) |
| Short-rib Thoracic Dysplasia Syndrome, (Saldino-Noonan type) | Thoracic dysplasia with narrow chest, short, flipper-like limbs with polydactyly, and frequent genitourinary abnormalities | ***DYNC2H1*** | (Bernstein et al., 1985) |
| Smith-Lemli-Opitz Syndrome | Cognitive impairment, microcephaly, dysmorphic features, syndactyly of toes, hypospadias and ambiguous genitalia, photosensitivity, and autistic and aggressive behaviour | ***DHCR7*** | (Bialer et al., 1987) |
| STAR Syndrome | Syndactyly of toes, telecanthus, and anogenital and renal malformations | ***FAM58A*** | (Unger et al., 2008) |
| Terminal Osseous Dysplasia | Skeletal dysplasia of limbs, pigmentary defects of the skin, recurrent digital fibroma | ***FLNA*** | (Robertson et al., 2003) |
| Tetra-amelia | Tetra-amelia, absent pelvic bones, hydrocephalus, facial cleft, absent ears/nose/nipples, anal atresia, and vertebral and rib anomalies | ***WNT3*** | (Kosaki et al., 1996) |
| Ulnar-mammary Syndrome | Asymmetric ulnar hypoplasia, mammary hypoplasia, hypoplasia of axillary hair and sweat glands, scoliosis, genital abnormalities, delayed puberty, and growth retardation | ***TBX3*** | (Bamshad et al., 1997) |
| Waardenburg Syndrome (Type 1) | Pigmentary abnormalities of the skin, hair, and eyes, sensorineural hearing loss, and dystopia canthorum | ***PAX3*** | (Goodman et al., 1988) |

# **References**

Atsuta, Y., & Takahashi, Y. Early formation of the Mullerian duct is regulated by sequential actions of BMP/Pax2 and FGF/Lim1 signaling. *Development, 143*(19), 3549-3559. doi:10.1242/dev.137067 (2016).

Arango N.A, Szotek P.P, Manganaro T.F, Oliva E, Donahoe P.K and Teixeira J: Conditional deletion of beta-catenin in the mesenchyme of the developing mouse uterus results in a switch to adipogenesis in the myometrium. Dev Biol 288: 276-283 (2005).

Balci S, Onol B, Ercal M.D, Beksac S, Erzen C and Akhan O: Meckel Gruber syndrome: a case diagnosed in utero. The Turkish journal of pediatrics34: 179-185 (1992).

Bamshad M, Lin R.C, Law D.J, Watkins W.S, Krakowiak P.A, Moore M.E, Franceschini P, Lala R, Holmes L.B, Gebuhr T.C: Mutations in human TBX3 alter limb, apocrine and genital development in ulnar-mammary syndrome. Nat Genet16: 311-315 (1997).

Benson G.V, Lim H, Paria B.C, Satokata I, Dey S.K and Maas R.L: Mechanisms of reduced fertility in Hoxa-10 mutant mice: uterine homeosis and loss of maternal Hoxa-10 expression. Development122: 2687-2696 (1996).

Bernstein R, Isdale J, Pinto M, Du Toit Zaaijman J and Jenkins T: Short rib-polydactyly syndrome: a single or heterogeneous entity? A re-evaluation prompted by four new cases. J Med Genet22: 46-53 (1985).

Bialer M.G, Penchaszadeh V.B, Kahn E, Libes R, Krigsman G and Lesser M.L: Female external genitalia and mullerian duct derivatives in a 46,XY infant with the smith-lemli-Opitz syndrome. Am J Med Genet28: 723-731 (1987).

Biason-Lauber A, Konrad D, Navratil F and Schoenle E.J: A WNT4 mutation associated with Mullerian-duct regression and virilization in a 46,XX woman. The New England journal of medicine 351: 792-798 (2004).

Bingham C, Ellard S, Cole T.R, Jones K.E, Allen L.I, Goodship J.A, Goodship T.H, Bakalinova-Pugh D, Russell G.I, Woolf A.S, Nicholls A.J and Hattersley A.T: Solitary functioning kidney and diverse genital tract malformations associated with hepatocyte nuclear factor-1beta mutations. Kidney international 61: 1243-1251 (2002).

Bombard D.S and Mousa S.A: Mayer–Rokitansky–Kuster–Hauser syndrome: complications, diagnosis and possible treatment options: a review. Gynecological Endocrinology 30: 618-623 (2014).

Brucker SY, Frank L, Eisenbeis S, Henes M, Wallwiener D, Riess O, et al. Sequence variants in ESR1 and OXTR are associated with Mayer-Rokitansky-Küster-Hauser syndrome. Acta Obstet Gynecol Scand 2017;96(11):1338–46.

Chen M.J, Wei S.Y, Yang W.S, Wu T.T, Li H.Y, Ho H.N, Yang Y.S and Chen P.L: Concurrent exome-targeted next-generation sequencing and single nucleotide polymorphism array to identify the causative genetic aberrations of isolated Mayer–Rokitansky–Küster–Hauser syndrome. Human Reproduction 30: 1732-1742 (2015).

Cheng Z, Zhu Y, Su D, Wang J, Cheng L, Chen B, Wei Z, Zhou P, Wang B, Ma X and Cao Y: A novel mutation of HOXA10 in a Chinese woman with a Müllerian duct anomaly. Human Reproduction 26: 3197-3201 (2011).

Christopoulos P, Gazouli M, Fotopoulou G and Creatsas G: The role of genes in the development of Mullerian anomalies: where are we today? Obstetrical & gynecological survey 64: 760-768 (2009).

Davis R.J, Harding M, Moayedi Y and Mardon G: Mouse Dach1 and Dach2 are redundantly required for Müllerian duct development. genesis 46: 205-213 (2008).

Demirhan O, Türkmen S, Schwabe G.C, Soyupak S, Akgül E, Taştemir D, Karahan D, Mundlos S and Lehmann K: A homozygous BMPR1B mutation causes a new subtype of acromesomelic chondrodysplasia with genital anomalies. Journal of Medical Genetics 42: 314-317 (2005).

Deutscher E and Hung-Chang Yao H: Essential roles of mesenchyme-derived beta-catenin in mouse Mullerian duct morphogenesis. Dev Biol 307: 227-236 (2007).

Ekici A.B, Strissel P.L, Oppelt P.G, Renner S.P, Brucker S, Beckmann M.W and Strick R: HOXA10 and HOXA13 sequence variations in human female genital malformations including congenital absence of the uterus and vagina. Gene 518: 267-272 (2013).

Filges I, Nosova E, Bruder E, Tercanli S, Townsend K, Gibson W.T, Rothlisberger B, Heinimann K, Hall J.G, Gregory-Evans C.Y, Wasserman W.W, Miny P and Friedman J.M: Exome sequencing identifies mutations in KIF14 as a novel cause of an autosomal recessive lethal fetal ciliopathy phenotype. Clin Genet 86: 220-228 (2014).

Fluck C.E, Tajima T, Pandey A.V, Arlt W, Okuhara K, Verge C.F, Jabs E.W, Mendonca B.B, Fujieda K and Miller W.L: Mutant P450 oxidoreductase causes disordered steroidogenesis with and without Antley-Bixler syndrome. Nat Genet 36: 228-230 (2004).

Fraser G.R: Our genetical ‘load’. A review of some aspects of genetical variation. Annals of Human Genetics 25: 387-415 (1962).

Gendron R.L, Paradis H, Hsieh-Li H.M, Lee D.W, Potter S.S and Markoff E: Abnormal uterine stromal and glandular function associated with maternal reproductive defects in Hoxa-11 null mice. Biology of reproduction 56: 1097-1105 (1997).

Gervasini C, Grati F.R, Lalatta F, Tabano S, Gentilin B, Colapietro P, De Toffol S, Frontino G, Motta F, Maitz S, Bernadini L, Dallapiccola B, Fedele L, Larizza L and Miozzo M: SHOX duplications found in some cases with type I Mayer-Rokitansky-Kuster-Hauser syndrome. Genet Med 12: 634-640 (2010).

Gilad Y and Lancet D: Population Differences in the Human Functional Olfactory Repertoire. Molecular Biology and Evolution 20: 307-314 (2003).

Girisha K.M, Bidchol A.M, Sarpangala M.K and Satyamoorthy K: A novel frameshift mutation in TWIST2 gene causing Setleis syndrome. Indian journal of pediatrics 81: 302-304 (2014).

Goodman R.M, Oelsner G, Berkenstadt M and Admon D: Absence of a vagina and right sided adnexa uteri in the Waardenburg syndrome: a possible clue to the embryological defect. J Med Genet 25: 355-357 (1988).

Goyal D, Yadav D.K, Shukla U and Sethi S.K: Coffin-Siris syndrome with Mayer-Rokitansky-Küster-Hauser syndrome: a case report. Journal of Medical Case Reports 4: 354-354 (2010).

Guazzarotti L, Caprio C, Rinne T.K, Bosoni M, Pattarino G, Mauri S, Tadini G.L, van Bokhoven H and Zuccotti G.V: Limb-mammary syndrome (LMS) associated with internal female genitalia dysgenesia: a new genotype/phenotype correlation? Am J Med Genet A 146a: 2001-2004 (2008).

Heidarpour M and Tavanafar Z: Diagnostic utility of PAX8 in differentiation of mullerian from non-mullerian tumors. Advanced Biomedical Research 3: 96 (2014).

Jeong J.W, Lee H.S, Franco H.L, Broaddus R.R, Taketo M.M, Tsai S.Y, Lydon J.P and DeMayo F.J: Beta-catenin mediates glandular formation and dysregulation of beta-catenin induces hyperplasia formation in the murine uterus. Oncogene 28: 31-40 (2009).

Kobayashi A, Shawlot W, Kania A and Behringer R.R: Requirement of Lim1 for female reproductive tract development. Development 131: 539-549 (2004).

Kochling J, Karbasiyan M and Reis A: Spectrum of mutations and genotype-phenotype analysis in Currarino syndrome. Eur J Hum Genet 9: 599-605 (2001).

Kosaki K, Jones M.C and Stayboldt C: Zimmer phocomelia: delineation by principal coordinate analysis. Am J Med Genet 66: 55-59 (1996).

Krantz I.D, McCallum J, DeScipio C, Kaur M, Gillis L.A, Yaeger D, Jukofsky L, Wasserman N, Bottani A, Morris C.A, Nowaczyk M.J, Toriello H, Bamshad M.J, Carey J.C, Rappaport E, Kawauchi S, Lander A.D, Calof A.L, Li H.H, Devoto M and Jackson L.G: Cornelia de Lange syndrome is caused by mutations in NIPBL, the human homolog of Drosophila melanogaster Nipped-B. Nat Genet 36: 631-635 (2004).

Ledig S, Brucker S, Barresi G, Schomburg J, Rall K and Wieacker P: Frame shift mutation of LHX1 is associated with Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome. Human Reproduction 27: 2872-2875 (2012).

Ledig S, Tewes A-C, Hucke J, Römer T, Kapczuk K, Schippert C, et al. Array-CGH Analysis in Patients with Müllerian Fusion Anomalies. Clin Genet (2017)

Lindner T.H, Njolstad P.R, Horikawa Y, Bostad L, Bell G.I and Sovik O: A novel syndrome of diabetes mellitus, renal dysfunction and genital malformation associated with a partial deletion of the pseudo-POU domain of hepatocyte nuclear factor-1beta. Hum Mol Genet 8: 2001-2008 (1999).

Liu S, Gao X, Qin Y, Liu W, Huang T, Ma J, Simpson J.L and Chen Z.J: Nonsense mutation of EMX2 is potential causative for uterus didelphysis: first molecular explanation for isolated incomplete müllerian fusion. Fertility and Sterility 103: 769-774.e762 (2015).

Lokmane L, Heliot C, Garcia-Villalba P, Fabre M and Cereghini S: vHNF1 functions in distinct regulatory circuits to control ureteric bud branching and early nephrogenesis. Development 137: 347-357 (2010).

Ma D, Marion R, Punjabi N.P, Pereira E, Samanich J, Agarwal C, Li J, Huang C.K, Ramesh K.H, Cannizzaro L.A and Naeem R: A de novo 10.79 Mb interstitial deletion at 2q13q14.2 involving PAX8 causing hypothyroidism and mullerian agenesis: a novel case report and literature review. Molecular Cytogenetics 7: 1-6 (2014).

Ma W, Li Y, Wang M, Li H, Su T, Li Y and Wang S: Associations of Polymorphisms in WNT9B and PBX1 with Mayer-Rokitansky-Küster-Hauser Syndrome in Chinese Han. PLoS One 10 (2015).

McGowan R, Tydeman G, Shapiro D, Craig T, Morrison N, Logan S, Balen A.H, Ahmed S.F, Deeny M, Tolmie J and Tobias E: DNA copy number variations are important in the complex genetic architecture of müllerian disorders. Fertility and Sterility 103: 1021-1030.e1021 (2015).

McKusick V.A, Bauer R.L, Koop C.E and Scott R.B: Hydrometrocolpos as a simply inherited malformation. Jama 189: 813-816 (1964).

Mendelsohn C, Lohnes D, Decimo D, Lufkin T, LeMeur M, Chambon P and Mark M: Function of the retinoic acid receptors (RARs) during development (II). Multiple abnormalities at various stages of organogenesis in RAR double mutants. Development 120: 2749-2771 (1994).

Mericskay M, Kitajewski J and Sassoon D: Wnt5a is required for proper epithelial-mesenchymal interactions in the uterus. Development 131: 2061-2072 (2004).

Miller C and Sassoon D.A: Wnt-7a maintains appropriate uterine patterning during the development of the mouse female reproductive tract. Development 125: 3201-3211 (1998).

Mishina Y, Rey R, Finegold M.J, Matzuk M.M, Josso N, Cate, R.L and Behringer R.R: Genetic analysis of the Mullerian-inhibiting substance signal transduction pathway in mammalian sexual differentiation. Genes & development 10: 2577-2587 (1996).

Miyamoto N, Yoshida M, Kuratani S, Matsuo I and Aizawa S: Defects of urogenital development in mice lacking Emx2. Development 124: 1653-1664 (1997).

Morcel K, Watrin T, Jaffre F, Deschamps S, Omilli F, Pellerin I, Leveque J and Guerrier D: Involvement of ITIH5, a candidate gene for congenital uterovaginal aplasia (Mayer-Rokitansky-Kuster-Hauser syndrome), in female genital tract development. Gene expression 15: 207-214 (2012).

Morcel K, Watrin T, Pasquier L, Rochard L, Le Caignec C, Dubourg C, Loget P, Paniel B.J, Odent S, David V, Pellerin I, Bendavid C and Guerrier D: Utero-vaginal aplasia (Mayer-Rokitansky-Kuster-Hauser syndrome) associated with deletions in known DiGeorge or DiGeorge-like loci. Orphanet Journal of Rare Diseases 6: 9 (2011).

Murry J, Santos X, Wang X, Wan Y-W, Van den Veyver I and Dietrich J: A genome-wide screen for copy number alterations in an adolescent pilot cohort with müllerian anomalies. Fertility and Sterility 103(2): 487-93 (2015).

Nachtigal M.W, Hirokawa Y, Enyeart-VanHouten D.L, Flanagan J.N, Hammer G.D and Ingraham H.A: Wilms' tumor 1 and Dax-1 modulate the orphan nuclear receptor SF-1 in sex-specific gene expression. Cell 93: 445-454 (1998).

Nik-Zainal S, Strick R, Storer M, Huang N, Rad R, Willatt L, Fitzgerald T, Martin V, Sandford R, Carter N.P, Janecke A, Renner S, Oppelt P, Oppelt P, Schulze C, Brucker S, Hurles M, Beckmann M, Strissel P and Shaw-Smith C: High incidence of recurrent copy number variants in patients with isolated and syndromic Müllerian aplasia. Journal of Medical Genetics 48: 197-204 (2011).

Patton M.A and Afzal A.R: Robinow syndrome. J Med Genet 39: 305-310 (2002).

Philibert P, Biason-Lauber A, Gueorguieva I, Stuckens C, Pienkowski C, Lebon-Labich B, Paris F and Sultan C: Molecular analysis of WNT4 gene in four adolescent girls with mullerian duct abnormality and hyperandrogenism (atypical Mayer-Rokitansky-Küster-Hauser syndrome). Fertility and Sterility 95: 2683-2686 (2011).

Rall K, Eisenbeis S, Barresi G, Rückner D, Walter M, Poths S, Wallwiener D, Riess O, Bonin M and Brucker S: Mayer-Rokitansky-Küster-Hauser syndrome discordance in monozygotic twins: matrix metalloproteinase 14, low-density lipoprotein receptor–related protein 10, extracellular matrix, and neoangiogenesis genes identified as candidate genes in a tissue-specific mosaicism. Fertility and Sterility 103: 494-502.e493 (2015).

Rall K, Eisenbeis S, Henninger V, Henes M, Wallwiener D, Bonin M and Brucker S: Typical and Atypical Associated Findings in a Group of 346 Patients with Mayer-Rokitansky-Kuester-Hauser Syndrome. Journal of Pediatric and Adolescent Gynecology 28: 362-368 (2015).

Robertson S.P, Twigg S.R.F, Sutherland-Smith A.J, Biancalana V, Gorlin R.J, Horn D, Kenwrick S.J, Kim C.A, Morava E, Newbury-Ecob R, Orstavik K, Quarrell O, Schwartz C, Shears D, Suri M, Kendrick-Jones J and Wilkie A: Localized mutations in the gene encoding the cytoskeletal protein filamin A cause diverse malformations in humans. Nat Genet 33: 487-491 (2003).

Sadedin S.P, Dashnow H, James P.A, Bahlo M, Bauer D.C, Lonie A, Lunke S, Macciocca I, Ross J.P, Siemering K.R, Stark Z, White S.M, Taylor G, Gaff C, Oshlack A and Thorne N.P: Cpipe: a shared variant detection pipeline designed for diagnostic settings. Genome medicine 7: 68 (2015).

Sandbacka M, Laivuori H, Freitas É, Halttunen M, Jokimaa V, Morin-Papunen L, Rosenberg C and Aittomäki K: TBX6, LHX1 and copy number variations in the complex genetics of Müllerian aplasia. Orphanet Journal of Rare Diseases 8: 125-125 (2013).

Schnabel C.A, Selleri L and Cleary M.L: Pbx1 is essential for adrenal development and urogenital differentiation. Genesis 37: 123-130 (2003).

Seller M.J, Davis T.B, Fear C.N, Flinter F.A, Ellis I and Gibson A.G: Two sibs with anophthalmia and pulmonary hypoplasia (the Matthew-Wood syndrome). Am J Med Genet 62: 227-229 (1996).

Siala O, Belguith N and Fakhfakh F: An Unusual Case of Peters Plus Syndrome with Sexual Ambiguity and Absence of Mutations in the B3GALTL Gene. Iranian Journal of Pediatrics 23: 485-488 (2013).

Spenlé C, Simon-Assmann P, Orend G and Miner J.H: Laminin α5 guides tissue patterning and organogenesis. Cell Adhesion & Migration 7: 90-100 (2013).

Stoler J.M, Herrin J.T and Holmes L.B: Genital abnormalities in females with Bardet-Biedl syndrome. Am J Med Genet 55: 276-278 (1995).

Strubbe E.H, Cremers C.W, Willemsen W.N, Rolland R and Thijn C.J: The Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome without and with associated features: two separate entities? Clinical dysmorphology 3: 192-199 (1994).

Tewes A.C, Rall K.K, Römer T, Hucke J, Kapczuk K, Brucker S, Wieacker P and Ledig S: Variations in RBM8A and TBX6 are associated with disorders of the müllerian ducts. Fertility and Sterility 103: 1313-1318 (2015).

Torres, M., Gomez-Pardo, E., Dressler, G. R., & Gruss, P. (1995). Pax-2 controls multiple steps of urogenital development. Development, 121(12), 4057-4065.

Unger S, Bohm D, Kaiser F.J, Kaulfuss S, Borozdin W, Buiting K, Burfeind P, Bohm J, Barrionuevo F, Craig, A, Borowski K, Keppler-Noreuil K, Schmitt-Mechelke T, Steiner B, Bartholdi D, Lemke J, Mortier G, Sandford R, Zabel B, Superti-Furga A and Kohlhase J: Mutations in the cyclin family member FAM58A cause an X-linked dominant disorder characterized by syndactyly, telecanthus and anogenital and renal malformations. Nat Genet 40: 287-289 (2008).

Van Bever Y, Van den Ende J.J and Richieri-Costa A: Oculo-auriculo-vertebral complex and uncommon associated anomalies: report on 8 unrelated Brazilian patients. Am J Med Genet 44: 683-690 (1992).

Van Den Berg D.J and Francke U: Roberts syndrome: a review of 100 cases and a new rating system for severity. Am J Med Genet 47: 1104-1123 (1993).

Van der Auwera G.A, Carneiro M.O, Hartl C, Poplin R, del Angel G, Levy-Moonshine A, Jordan T, Shakir K, Roazen D, Thibault J, Banks E, Garimella K, Altshuler D, Gabriel S and DePristo M: From FastQ data to high confidence variant calls: the Genome Analysis Toolkit best practices pipeline. Current protocols in bioinformatics / editoral board, Andreas D Baxevanis [et al] 11: 11.10.11-11.10.33 (2013).

Warot X, Fromental-Ramain C, Fraulob V, Chambon P and Dolle P: Gene dosage-dependent effects of the Hoxa-13 and Hoxd-13 mutations on morphogenesis of the terminal parts of the digestive and urogenital tracts. Development 124: 4781-4791 (1997).

Willem M, Miosge N, Halfter W, Smyth N, Jannetti I, Burghart E, Timpl R and Mayer U: Specific ablation of the nidogen-binding site in the laminin γ1 chain interferes with kidney and lung development. Development 129: 2711-2722 (2002).

Zenker M, Mayerle J, Lerch M.M, Tagariello A, Zerres K, Durie P.R, Beier M, Hulskamp G, Guzman C, Rehder H, Beemer F, Hamel B, Vanlieferinghen P, Gershoni-Baruch R, Vieira M, Dumic M, Auslender R, Gil-da-Silva-Lopes V, Steinlicht S, Rauh M, Shalev S, Thiel C, Winterpacht A, Kwon Y, Varshavsky A and Reis A: Deficiency of UBR1, a ubiquitin ligase of the N-end rule pathway, causes pancreatic dysfunction, malformations and mental retardation (Johanson-Blizzard syndrome). Nat Genet 37: 1345-1350 (2005).