# KIT gene

| No. cases | Mutation   | Exon | DNA                | Protein               | Variant effect                  |
|-----------|------------|------|-------------------|-----------------------|---------------------------------|
| 4         | Y503_F504ins | 9    | c.1509_1510insGCCTAT | p.Tyr503_Phe504insAY   | Insertion - In frame            |
| 1         | E561del    | 11   | c.1681_1683delGAG  | p.Glu561del           | Deletion - In frame             |
| 1         | G565_L576del | 11   | c.1694_1727del33   | p.Gly565_Leu576del    | Deletion - In frame             |
| 1         | HS80_K581ins4 | 11   | c.1740_1741ins12   | p.His580_Lys581insPYDH| Insertion - In frame            |
| 1         | K558_Q575del | 11   | c.1672_1725del54   | p.Lys558_Gln575del    | Deletion - In frame             |
| 4         | K558_V559del | 11   | c.1672_1677delAAAGTT | p.Lys558_Val559del    | Deletion - In frame             |
| 1         | K558_V560>N | 11   | c.1674_1679delGGTTGT | p.Lys558_Val560>Asn   | Complex - deletion              |
|           |            |      |                   |                       | insertion inframe               |
| 1         | K558>NP    | 11   | c.1673_1674insTCC  | p.Lys558>AsnPro       | Complex - insertion              |
|           |            |      |                   |                       | inframe                         |
| 1         | R588_L589ins | 11   | c.1669_1674delTGGAAG | p.Arg588_Leu589ins17  | Insertion - In frame            |
| 1         | M552_V559>I | 11   | c.1654_1677del24   | p.Met552_Val559>Ile   | Complex - deletion              |
|           |            |      |                   |                       | inframe                         |
| 1         | M552_Y553del | 11   | c.1654_1659delATGTAT | p.Met552_Tyr553del    | Complex - deletion              |
|           |            |      |                   |                       | inframe                         |
| 1         | N564_L576del | 11   | c.1690_1728del39   | p.Asn564_Leu576del    | Deletion - In frame             |
| 1         | N566_P573del | 11   | c.1697_1720del24   | p.Asn566_Pro573       | Deletion - In frame             |
|           |            |      |                   | delINNYYIDP           |                                 |
| 2         | P551_E554del | 11   | c.1651_1662del12   | p.Pro551_Glu554del    | Deletion - In frame             |
| 2         | P551_V555del | 11   | c.1651_1665del15   | p.Pro551_Val555del    | Deletion - In frame             |
| 1         | P585_R586ins | 11   | c.1755_1756ins30   | p.Pro585_Arg586       | Insertion - In frame             |
|           |            |      |                   | insLPYDHKWEFP         |                                 |
| 5         | V555_V559del | 11   | c.1663_1677del15   | p.Val555_Val559del    | Deletion - In frame             |
| 1         | V559_G565del | 11   | c.1675_1692del18   | p.Val559_Gly565del    | Deletion - In frame             |
| 1         | V559A + G565A | 11   | c.1676T>C + c.1694G>C | p.Val559Aa+p.Gly565Aa | Substitution-Missense           |
| 10        | V559D      | 11   | c.1676T>A          | p.Val559Asp           | Substitution-Missense           |
| 1         | V559del    | 11   | c.1675_1677delGT   | p.Val559del           | Deletion - In frame             |
| 1         | V560_N566del | 11   | c.1677_1695del18   | p.Val560_Asn566del    | Deletion - In frame             |
| 1         | V560del    | 11   | c.1678_1680delGT   | p.Val560del           | Deletion - In frame             |
| 2         | V560G      | 11   | c.1679T>G          | p.Val560G             | Substitution-Missense           |
| 4         | V569_Y578del | 11   | c.1706_1733del29 + | p.Val569_Tyr578del    | Deletion - In frame             |
|           |            |      | c.1706_1710insAAGTT|                       |                                 |
| 2         | W557_J563del | 11   | c.1669_1680del12   | p.Trp557_Iso563del    | Deletion - In frame             |
| 21        | W557_K558del | 11   | c.1669_1674delTGGAAG | p.Trp557_Lys558del | Deletion - In frame             |
| 2         | W557_V559del | 11   | c.1669_1677del9    | p.Trp557_Val559del    | Deletion - In frame             |
| 1         | W557_V559>F | 11   | c.1670_1675delGGAAAG | p.Trp557_Val559>Phe  | Complex - deletion              |
|           |            |      |                   |                       | inframe                         |
| 1         | W557_V560>C | 11   | c.1671_1679del9    | p.Trp557_Val560>Cys   | Complex - deletion              |
|           |            |      |                   |                       | inframe                         |
| 3         | W557R      | 11   | c.1669T>A          | p.Trp557Arg           | Substitution-Missense           |
| 1         | K642E      | 13   | c.1924A>G          | p.Lys642Glu           | Substitution-Missense           |

# PDGFRA gene

| No. cases | Mutation | Exon | DNA    | Protein   | Variant effect |
|-----------|----------|------|--------|-----------|---------------|
| 1         | V561D    | 12   | c.1682T>A | p.Val561Asp | Substitution-Missense |
| 4         | D842V    | 18   | c.2525A>T | p.Asp842Val | Substitution-Missense |