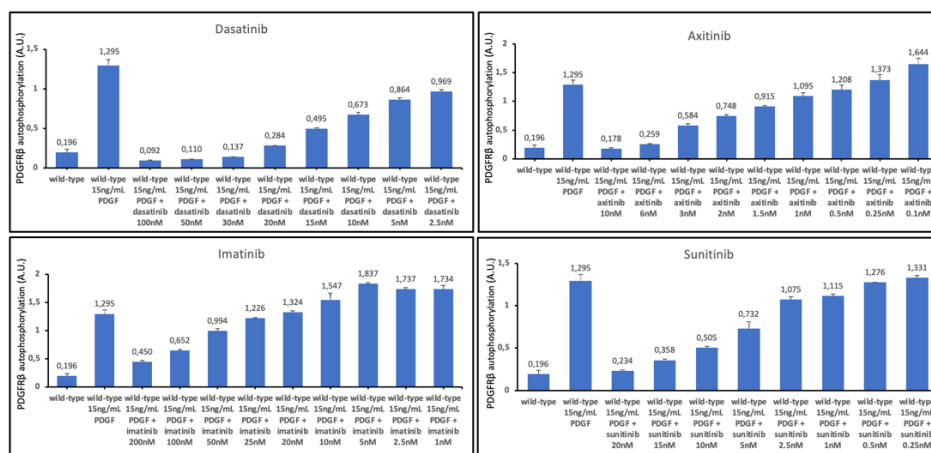


**SUPPLEMENTAL FIGURES:**

Individual	Clinical Findings	Age of Onset	Visual Acuity	p.Ser548Tyr <i>PDGFRB</i> variant
II.1	O.d. a temporal pterygium extending over the visual axis.	early 20s	o.d. hand motions; o.s. 20/20	+
III.2	O.d. a temporal pterygium extending over the visual axis and a small nasal pterygium. o.s. a temporal pterygium without visual axis involvement.	6 years old	o.d. counting fingers o.s. 20/25	+
III.3	O.d. an inferotemporal pterygium extending over the visual axis and a small nasal pterygium.	4 years old	o.d. counting fingers o.s. 20/40	+
II.3	No history of pterygium. Central corneal scarring with associated peripheral vascularization.	NIA	blind	-
III.6	No history of pterygium. In the left eye dense corneal opacity, lateral rectus palsy, and horizontal histagmus.	NIA	o.d. 20/20 o.s. counting fingers	-

Supplementary material Table 1. Summary of clinical data for individuals described in the case report <sup>16</sup>. Abbreviations used: o.d. and o.s. denotes the right eye and the left eye respectively, and NIA indicates that no information is available.

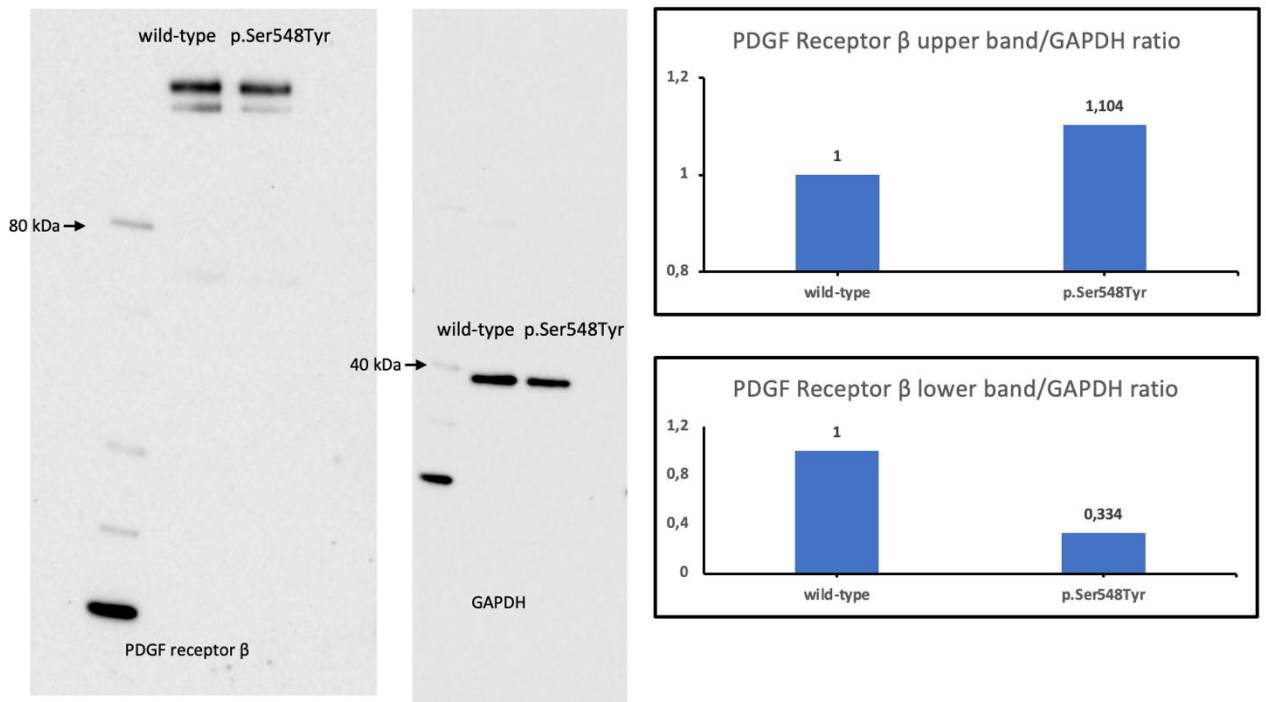


Supplementary Fig. 1. Titration study establishing IC<sub>50</sub> in-house concentrations of dasatinib, imatinib, sunitinib, and axitinib. HeLa cells were stably transduced with a wild-type *PDGFRB* vector and treated with various concentrations of axitinib, dasatinib, imatinib, and sunitinib for 6 hours. Twenty minutes prior to harvesting cells were stimulated with 15ng/mL PDGF. The TKI concentrations inhibiting 50% of the ligand-induced PDGFR $\beta$  activation were calculated from at least

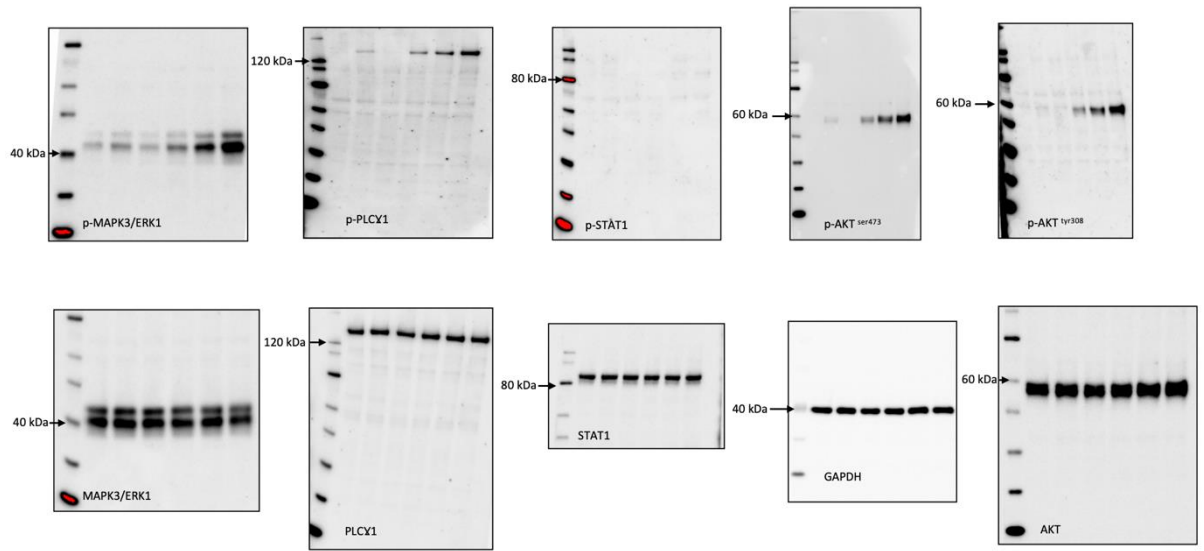
three independent experiments. The phosphorylation of PDGFR $\beta$  was measured with ELISA analysis.

	550
human	K V V V I <b>S</b> A I L A L V V L
chimpanzee	K V V V I <b>S</b> A I L A L V V L
rhesus	K A V V I <b>S</b> A I L A L V V L
wolf	K V V V I <b>S</b> A I L A L V V L
cow	K V V V I <b>S</b> A I L A L V V L
mouse	K V V V I <b>S</b> A I L A L V V L
rat	K V V V I <b>S</b> A I L A L V V L
rooster	K V V I I <b>S</b> V I L A L L V L
frog	K I V L V <b>S</b> A I L A L V V L
zebrafish	Q V A V L <b>A</b> A V L A L V V I
consensus	K V V V I <b>S</b> A I L A L V V L

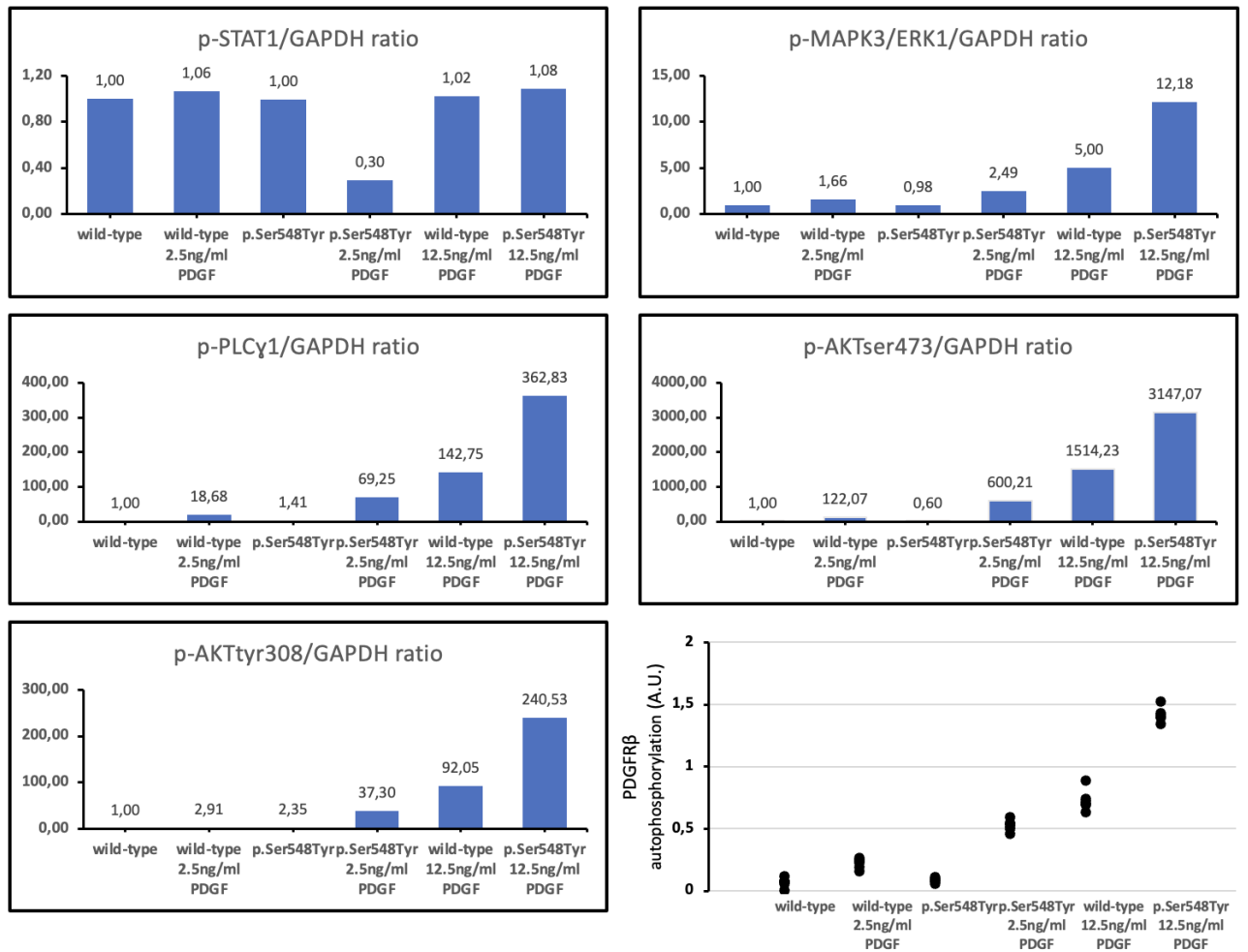
Supplementary Fig. 2. Amino acid alignment of a part of the transmembrane domain of PDGFR $\beta$  expressed from exon 11 of *PDGFRB*. All species presented have serine at the 548 position (marked in yellow) except for zebrafish.



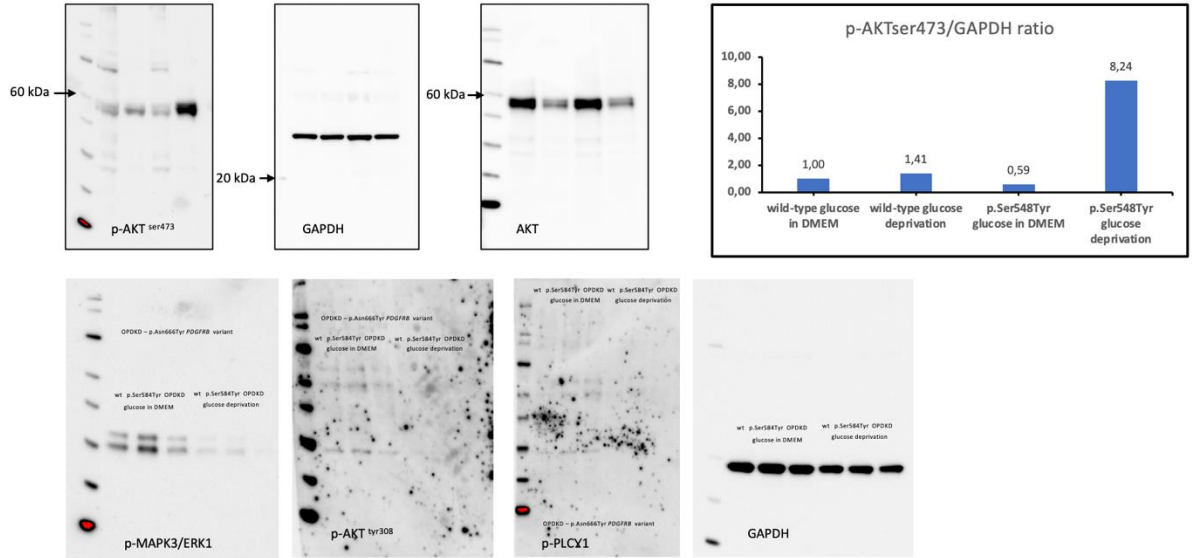
Supplementary Fig. 3. Immunoblot analysis showing the expression levels of PDGFR $\beta$  in wild-type and p.Ser548Tyr stably transduced HeLa cells. Immunoblot quantification is illustrated on the right side.



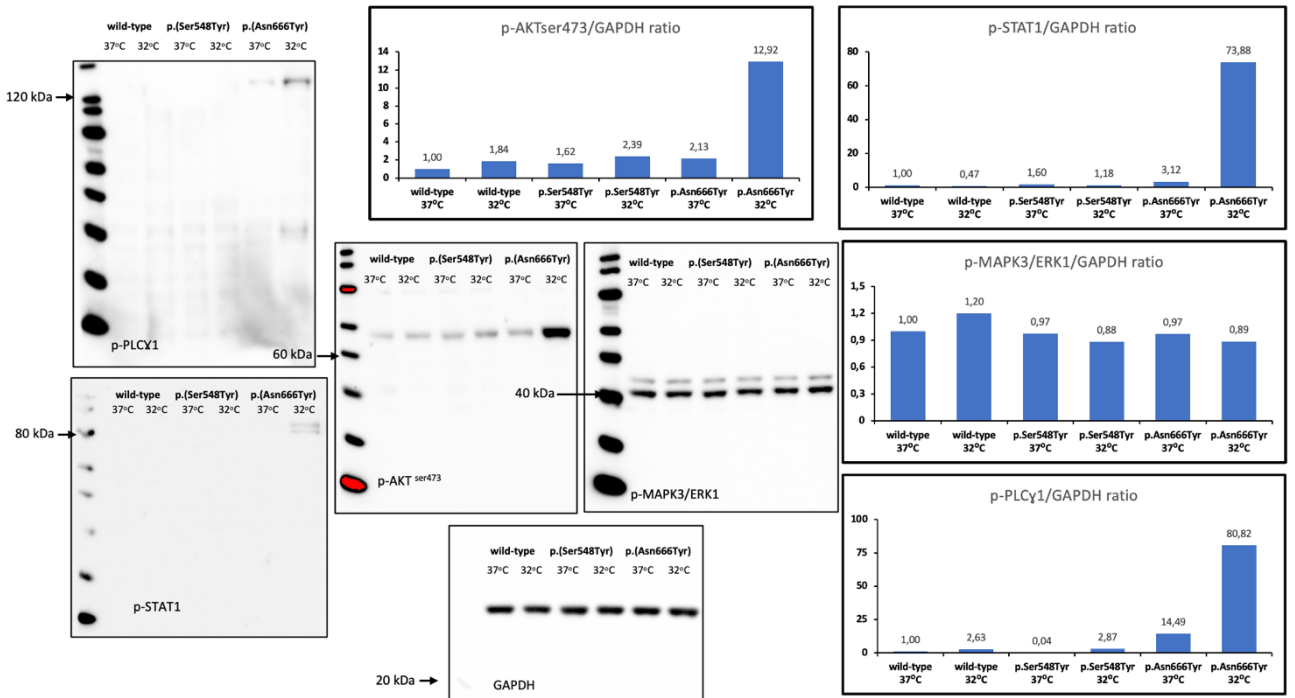
Supplementary Fig. 4. Complete blots corresponding to Figure 2., Panel B.



Supplementary Fig. 5. Quantification of blots corresponding to Figure 2., Panel B and individual ELISA analysis data point distribution corresponding to Figure 2., Panel A.

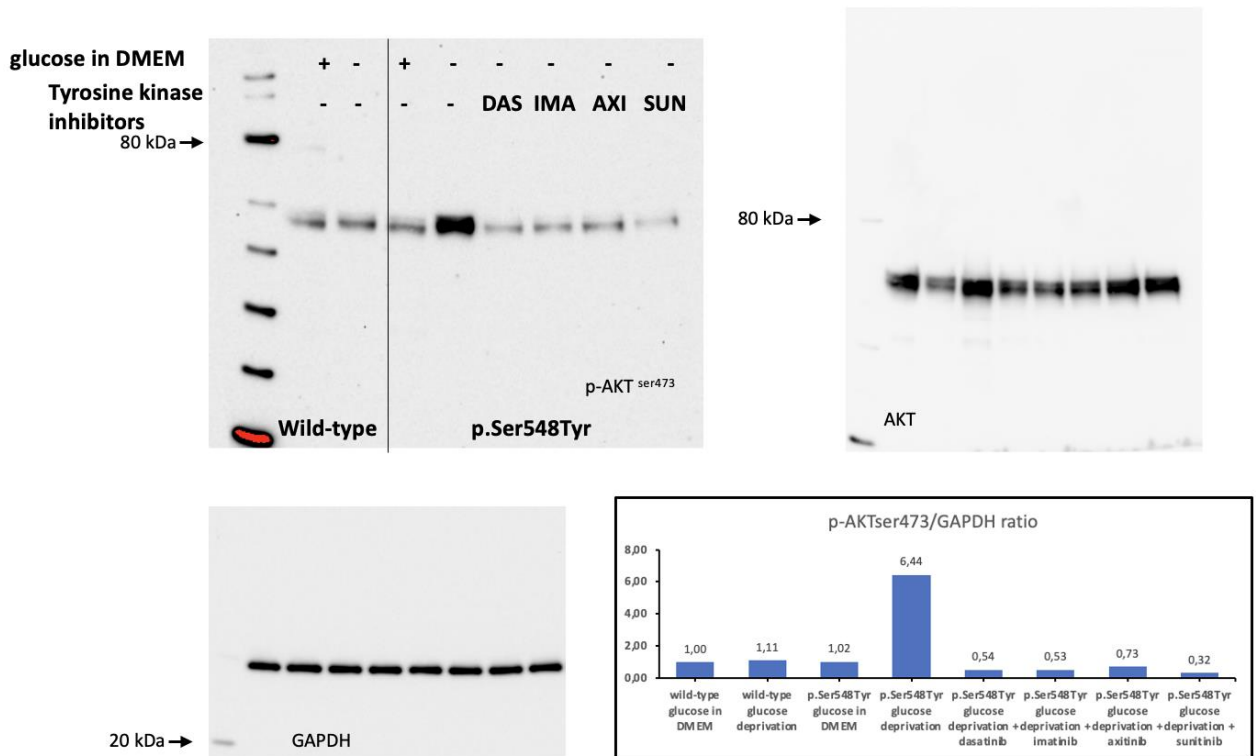


Supplementary Fig. 6. Complete blots and quantification of p-AKT<sup>ser473</sup> corresponding to Figure 3. Additional blots with p-MAPK3/ERK1, p-AKT<sup>tyr308</sup>, and p-PLC $\gamma$ 1, which were not included in the main figure, are shown here.



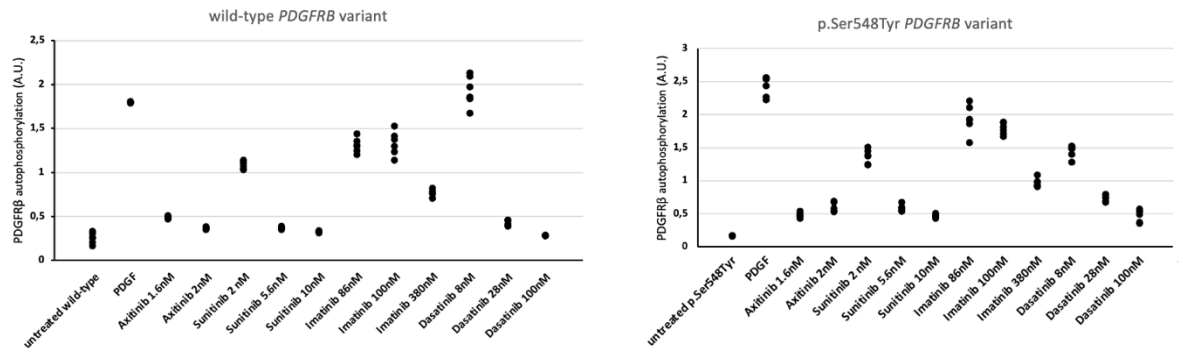
Supplementary Fig. 7. Effect of temperature on PDGFR $\beta$  downstream signaling.

Immunoblot analysis and quantifications show that the p.(Ser548Tyr) variant is not temperature sensitive, as levels of PDGFR $\beta$  downstream signaling proteins, p-AKT<sup>Ser473</sup>, p-STAT1, p-PLC  $\gamma$ 1, and p-MAPK3/ERK1 remained unaltered. The HeLa cells stably expressing the p.(Asn666Tyr) substitution were used as a positive control.

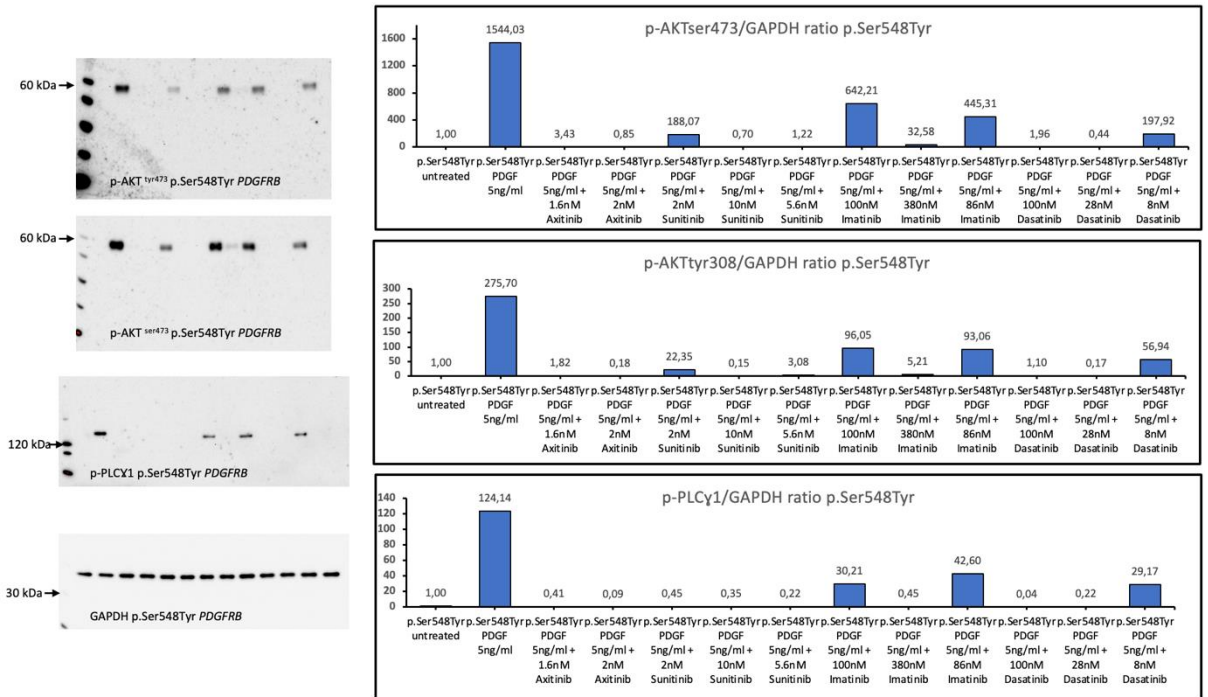


Supplementary Fig. 8. Immunoblot analysis and quantifications showing the effect of

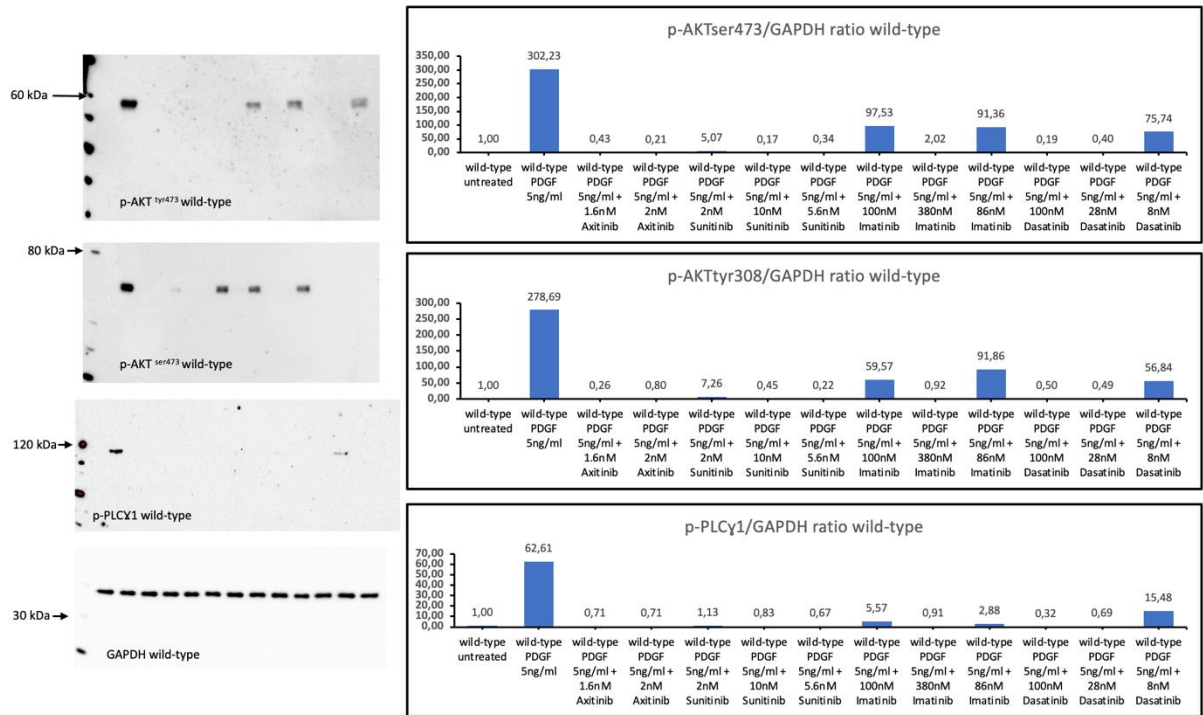
tyrosine kinase inhibitors on glucose withdrawal-induced upregulation of p-AKT<sup>Ser473</sup>. Axitinib 2nM, Dasatinib 8nM, Imatinib 86nM, and Sunitinib 5.6nM were used in this experiment.



Supplementary Fig. 9. Individual ELISA analysis data point distribution corresponding to Figure 4., Panel A and B.



Supplementary Fig. 10. Complete blots and quantifications of blots corresponding to Figure 4., Panel C.



Supplementary Fig. 11. Complete blots and quantifications of blots corresponding to Figure 4., Panel D.